PRENATALAUTISM

Elenco dei geni e delle malattie genetiche a trasmissione ereditaria investigati

PrenatalAutism - Elenco dei geni e delle malattie genetiche a trasmissione ereditaria investigati

Gene	Associated genetic disorder	inheritance	prevalence	OMIM gene
AAAS	Achalasia-addisonianism-alacrima syndrome	AR	1/1,000,000	605378
AASS	Hyperlysinemia	AR	Unknown	605113
ABAT	Deficit di acido gamma-aminobutirrico transaminasi	AR	1/1 000 000	137150
ABCA1	Tangier disease	AR	1/1 000 000	600046
ABCA12	Harlequin ichthyosis	AR	1/300 000	607800
ABCA3	Interstitial lung disease due to ABCA3 deficiency	AR/AD/XLR	Unknown	601615
ABCA4	Cone-rod dystrophy / Stargardt disease / Retinitis pigmentosa	AR	1-9/100 000	601691
ABCB11	Progressive familial intrahepatic cholestasis type 2	AR	1-9/100 000	603201
ABCB4	Progressive familial intrahepatic cholestasis type 3	AR	1-9/100 000	171060
ABCC6	Pseudoxanthoma elasticum	AR	1/25,000	603234
ABCD1	Adrenoleukodystrophy	XLR	Unknown	300371
ABCD4	Methylmalonic acidemia with homocystinuria	AR	Unknown	603214
ABHD12	Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome	AR	1/1 000 000	613599
ACAD8	Isobutyryl-CoA dehydrogenase deficiency	AR	Unknown	604773
ACAD9	Acyl-CoA dehydrogenase deficiency, type 9	AR	Unknown	611103
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency (MCADD)	AR	1-9/100 000	607008
ACADS	Short-chain acyl-CoA dehydrogenase deficiency (SCADD)	AR	Unknown	606885
ACADSB	2-Methylbutyryl-CoA dehydrogenase deficiency	AR	<1 / 1 000 000	600301
ACADVL	Very long-chain acyl-CoA dehydrogenase deficiency (VLCADD)	AR	1-9/100 000	609575
ACAT1	Beta-ketothiolase deficiency	AR	Unknown	607809
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	AR	<1 / 1 000 000	609751
ACSF3	Combined malonic and methylmalonic aciduria	AR	Unknown	614265
ACSL4	X-linked non-syndromic intellectual disability	XLR	Unknown	300157
ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency	AR	1-9 / 1 000 000	608958
ADAMTS13	Congenital thrombotic thrombocytopenic purpura	AR	<1 / 1 000 000	604134
ADAMTS2	Ehlers-Danlos syndrome, type VIIC	AR	1/1,000,000	604539
ADAMTSL2	Geleophysic dysplasia	AR	<1 / 1 000 000	612277
ADK	Encephalopathy associated with hypermethioninaemia due to adenosine kinase deficiency	AR	Unknown	102750
AFF2	FRAXE-related intellectual disability	XLR	1-9 / 1 000 000	300806
AGA	Aspartylglucosaminuria	AR	Unknown	613228
AGK	Sengers syndrome (Congenital cataract – hypertrophic cardiomyopathy – mitochondrial myopathy)	AR	<1 / 1 000 000	610345
AGL	Glycogen storage disease type 3a (GSD 3a) and type 3b (GSD 3b)	AR	Unknown	610860
AGPS	Rhizomelic chondrodysplasia punctata, type 3	AR	Unknown	603051
AGXT	Primary hyperoxaluria, type 1	AR	Unknown	604285
AHCY	S-adenosylhomocysteine hydrolase deficiency	AR	<1 / 1 000 000	180960
AHI1	Joubert syndrome with ocular involvement	AR	Unknown	608894
AIPL1	Leber congenital amaurosis / Cone-rod dystrophy	AR/AD	1-9 / 100 000	604392
AIRE	Autoimmune polyendocrine syndrome type 1	AR	1-9 / 1 000 000	607358
ALDH3A2	Sjögren–Larsson syndrome	AR	1-9 / 1 000 000	609523
ALDH4A1	Hyperprolinemia, type 2	AR		606811
ALDH7A1	Pyridoxine-dependent epilepsy	AR	1/100,000	107323
ALDOB	Hereditary fructose intolerance	AR	1-9 / 100 000	612724
ALG1	Congenital disorder of glycosylation, type 1k	AR	<1 / 1 000 000	605907
ALG12	Congenital disorder of glycosylation, type 1g	AR	<1 / 1 000 000	607144
ALG2	Congenital disorder of glycosylation, type 1i	AR	<1 / 1 000 000	607905
ALG3	Congenital disorder of glycosylation, type 1d	AR	<1 / 1 000 000	608750
ALG6	Congenital disorder of glycosylation, type 1c	AR	<1 / 1 000 000	604566
ALG8	Congenital disorder of glycosylation, type 1h	AR	<1 / 1 000 000	608103
ALG9	Congenital disorder of glycosylation, type 1L	AR	<1 / 1 000 000	606941
ALMS1	Alström syndrome	AR	1-9 / 1 000 000	606844
ALPL	Infantile hypophosphatasia / Odontohypophosphatasia	AR/AD	Unknown	171760
ALS2	Hereditary spastic paraplegia with infantile onset / Juvenile amyotrophic lateral sclerosis / Juvenile primary lateral sclerosis	AR	<1 / 1 000 000	205100
AMACR	Congenital defect in bile acid synthesis, type 4	AR	<1 / 1 000 000	604489
AMH	Persistent Müllerian duct syndrome, type 1	AR	1/1,000,000	600957
AMHR2	Persistent Müllerian duct syndrome, type 2	AR	1/1,000,000	600956
AMT	Glycine encephalopathy	AR	1-9 / 1 000 000	238310
ANO5	Limb-girdle muscular dystrophy, autosomal recessive type 2L / Gnathodiaphyseal dysplasia	AR/AD	Unknown	608662

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ANTXR2	Juvenile hyaline fibromatosis	AR	<1 / 1 000 000	608041
AP1S1	Mental retardation, enteropathy, deafness, peripheral neuropathy, ichthyosis, and keratoderma (MEDNIK)	AR	<1/1,000,000	603531
AP1S2	Fried syndrome	XLR	<1 / 1 000 000	300629
AP3B1	Hermansky–Pudlak syndrome due to AP-3 deficiency	AR	<1 / 1 000 000	603401
APTX	Ataxia with oculomotor apraxia	AR		606350
AQP2	Familial nephrogenic diabetes insipidus, AQP2-related	AR	<1/1,000,000	107777
AR	Androgen insensitivity syndrome	XLR		313700
ARG1	Argininemia	AR	<1/1.000.000	608313
ARHGEF6	X-linked non-syndromic intellectual disability	XLR	Unknown	300267
ARHGEF9	Hyperekplexia and epilepsy syndrome	XLR	<1 / 1 000 000	300429
ARL13B	Joubert syndrome	AR		608922
ARL6	Retinitis pigmentosa / Bardet–Biedl syndrome	AR	1-5 / 10 000	608845
ARSA	Metachromatic leukodystrophy	AR	1-9 / 1 000 000	607574
ARSB	Mucopolysaccharidosis type VI	AR	1-9/100.000	611542
ARSF	Chondrodysplasia punctata	XLR	Unknown	300003
ARX	Early infantile epileptic encephalopathy / X-linked non-syndromic intellectual disability / Agenesis of the corpus callosum–genital anomalies syndrome / Partington syndrome	XLR	Unknown	300382
ASAH1	Farber lipogranulomatosis	AR	<1 / 1 000 000	613468
ASL	Argininosuccinic aciduria	AR	1-9 / 100 000	608310
ASNS	Asparagine synthetase deficiency	AR	1/1,000,000	108370
ASPA	Canavan disease	AR	Unknown	608034
ASPM	Autosomal recessive primary microcephaly	AR	Unknown	605481
ASS1	Citrullinaemia	AR	1-9 / 100 000	603470
ATM	Ataxia–telangiectasia	AR	1-9/100.000.000	607585
ATP6V0A2	Autosomal recessive cutis laxa, type 2, classic form / Wrinkly skin syndrome	AR	Unknown	611716
ATP6V1B1	Renal tubular acidosis and deafness, ATP6V1B1-related	AR	1/100,000	192132
ATP7A	Menkes disease	XLR	Unknown	300011
ATP7B	Wilson disease	AR	1-9 / 100 000	606882
ATP8B1	Progressive familial intrahepatic cholestasis, type 1 / Benign recurrent intrahepatic cholestasis, type 1	AR	1-9 / 100 000	602397
ATR	Seckel syndrome / Familial cutaneous telangiectasia and oropharyngeal tumour predisposition syndrome	AR	<1/1.000.000	601215
ATRX	"Hypnotic facies intellectual disability syndrome, Juberg–Marsidi syndrome,	XL	Unknown	300032
AUH	Carpenter–Waziri syndrome, Holmes–Gang syndrome,	AR	<1 / 1 000 000	600529
B4GALT1	Smith–Fineman–Myers syndrome,	AR	<1 / 1 000 000	137060
BAG3	Alpha-thalassaemia / intellectual disability syndrome"	AR	1-5 / 10 000	603883
BBS1	Congenital disorder of glycosylation, type IId	AR	Unknown	209901
BBS10	Familial isolated dilated cardiomyopathy / Selcen-type muscular dystrophy	AR	Unknown	610148
BBS12	Williams–Beuren syndrome	AR	Unknown	610683
BBS2	Bardet–Biedl syndrome, type 1	AR	Unknown	606151
BBS4	Bardet–Biedl syndrome, type 10	AR	Unknown	600374
BBS5	Bardet–Biedl syndrome, type 12	AR	Unknown	603650
BBS7	Bardet–Biedl syndrome, type 2	AR	Unknown	607590
BBS9	Bardet–Biedl syndrome, type 4	AR	Unknown	607968
BCHE	Pseudocholinesterase deficiency	AR	1/50,000	177400
BCKDHA	Bardet–Biedl syndrome, type 5	AR	1-9/100.000.000	608348
BCKDHB	Bardet–Biedl syndrome, type 7	AR	1-9/100.000.000	248611
BCOR	Maple syrup urine disease, type Ib	XLR	Unknown	300485
BCS1L	Branched-chain ketoacid dehydrogenase kinase deficiency (autism–epilepsy syndrome)	AR	<1 / 1 000 000	603647
BIN1	Björnstad syndrome / GRACILE syndrome / Isolated complex III deficiency	AR	Unknown	601248
BLM	3q29 deletion/duplication syndrome	AR		604610
BLOC1S6	22q11.2 deletion syndrome	AR	1-9 / 1 000 000	604310
BMP1	Autosomal recessive centronuclear myopathy	AR	Unknown	112264
BRIP1	Fanconi anemia, group J	AR	<1/1,000,000	605882
BRWD3	Unspecified syndromic intellectual disability	XLR	Unknown	300553
BSND	Bartter syndrome, type 4	AR	Unknown	606412
BTD	Biotinidase deficiency	AR	1-9/100.000	609019
BTK	X-linked agammaglobulinemia, Short stature due to isolated growth hormone deficiency associated with X-linked hypogammaglobulinemia	XLR	1-9 / 1 000 000	300300
CA2	Osteopetrosis with renal tubular acidosis	AR	<1 / 1 000 000	611492
CANT1	Desbuquois dysplasia 1	AR	<1/1,000,000	613165
CAPN3	Limb-girdle muscular dystrophy type D4 related to calpain-3, Limb-girdle muscular dystrophy, autosomal recessive type 2A	AD/AR	<1 / 1 000 000	114240

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CASK	X-linked intellectual disability, Najm type / Early infantile epileptic encephalopathy	XLD	<1/1.000.000	300172
CASQ2	Tachicardia ventricolare polimorfa catecolaminergica	AR	1-5 / 10 000	114251
CBS	Classical homocystinuria	AR	1-9/100.000	613381
CC2D1A	Mental retardation, autosomal recessive 3	AR	<1/1,000,000	610055
CC2D2A	Joubert syndrome, type 9, Meckel syndrome, COACH syndrome	AR	<1 / 1 000 000	612013
CCDC103	Primary ciliary dyskinesia, type 17	AR	Unknown	614677
CCDC39	Primary ciliary dyskinesia, type 14	AR	Unknown	613798
CCDC40	Primary ciliary dyskinesia, type 15	AR	Unknown	613799
CCDC8	3M syndrome	AR	Unknown	614145
CD19	Common variable immunodeficiency	AR	1-9 / 100 000	107265
CD3D	Severe combined immunodeficiency (T-, B+, N+)	AR	Unknown	186790
CD3E	Severe combined immunodeficiency (T-, B+, NK+)	AR	Unknown	186830
CD3G	Combined immunodeficiency due to CD3-gamma deficiency	AR	Unknown	186740
CD40LG	X-linked hyper-IgM syndrome	XLR	Unknown	300386
CD79A	Agammaglobulinemia 3	AR	1-9 / 1 000 000	112205
CDH23	Usher syndrome, type 1	AR	1-9/100.000	605516
CDK5RAP2	Primary microcephaly, type 3	AR	Unknown	608201
CEP135	Primary microcephaly, type 8	AR	Unknown	611423
CEP290	Joubert syndrome with oculorenal defect	AR	<1 / 1 000 000	610142
CERKL	Retinite pigmentosa	AR	1-5 / 10 000	608381
CFP	Properdin deficiency	XLR		300383
CFTR	Cystic fibrosis	AR	1-5/10.000	602421
CHM	Choroideremia, X-linked	XLR	1/50,000	300390
CHRNA1	Congenital myasthenic syndrome	AD/AR	1-9 / 1 000 000	100690
CHRND	Congenital myasthenic syndrome (duplicate)	AD/AR	1-9 / 1 000 000	100720
CHRNE	Congenital myasthenic syndrome, CHRNE-related	AR	<1/1,000,000	100725
CHRNG	Lethal multiple pterygium syndrome	AR	<1 / 1 000 000	100730
CHST6	Macular corneal dystrophy	AR	1-9 / 100 000	605294
CIITA	Bare lymphocyte syndrome, CIITA-related	AR	1/1,000,000	600005
CLCN1	Recessive congenital myotonia	AR	1-9 / 100 000	118425
CLCN5	Dent disease, type 1	XLR	Unknown	300008
CLDN1	Ichthyosis–hypotrichosis–sclerosing cholangitis syndrome	AR	<1 / 1 000 000	603718
CLDN16	Renal hypomagnesemia, type 3	AR	<1 / 1 000 000	603959
CLDN19	Hypercalciuria syndrome with bilateral macular coloboma	AR	<1 / 1 000 000	610036
CLN3	Neuronal ceroid lipofuscinosis, type 3	AR	Unknown	607042
CLN5	Infantile neuronal ceroid lipofuscinosis	AR	1/20.000	608102
CLN6	Late-infantile neuronal ceroid lipofuscinosis	AR	Unknown	606725
CLN8	Late-infantile neuronal ceroid lipofuscinosis	AR	Unknown	607837
CLRN1	Usher syndrome, type 3A	AR	1-9/100.000	606397
CNGA1	Retinitis pigmentosa	AR	1-5 / 10 000	123825
CNGA3	Achromatopsia, CNGA3-related	AR	1/30,000	600053
CNGB1	Retinitis pigmentosa	AR	1-5 / 10 000	600724
CNGB3	Stargardt disease	AR	1-5 / 10 000	605080
COG1	Congenital disorder of glycosylation, type Iig	AR	<1 / 1 000 000	606973
COG7	Congenital disorder of glycosylation, type IIe	AR	<1 / 1 000 000	606978
COG8	Congenital disorder of glycosylation, type IIh	AR	<1 / 1 000 000	606979
COL11A2	Fibrochondrogenesis, type 2	AR	1/1,000,000	120290
COL17A1	Late-onset junctional epidermolysis bullosa	AR	<1 / 1 000 000	113811
COL4A3	Alport syndrome	AD/AR	1/50.000	120070
COL4A4	Alport syndrome (duplicate)	AD/AR	1/50.000	120131
COL4A5	X-linked Alport syndrome	XLD	1-9/100.000	303630
COL6A1	Ullrich congenital muscular dystrophy / Bethlem myopathy	AD/AR	1-9 / 1 000 000	120220
COL6A2	Ullrich congenital muscular dystrophy / Bethlem myopathy (duplicate)	AD/AR	1-9 / 1 000 000	120240
COL6A3	Ullrich congenital muscular dystrophy / Bethlem myopathy (duplicate)	AD/AR	1-9 / 1 000 000	120250
COL7A1	Epidermolysis bullosa	AD/AR	<1 / 1 000 000	120120
COQ2	Multiple system atrophy	AD/AR	1-9 / 100 000	609825
COQ9	Encephalopathy associated with hypertrophic cardiomyopathy and renal tubular disease	AR	<1 / 1 000 000	612837
COX10	Isolated cytochrome C oxidase deficiency	AR	Unknown	602125

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COX15	Lethal infantile cytochrome C oxidase deficiency	AR	Unknown	603646
COX6B1	Isolated cytochrome C oxidase deficiency (duplicate)	AR	Unknown	124089
CP	Cerebellar ataxia	AR	<1/1.000.000	117700
CPS1	Carbamoyl phosphate synthetase I deficiency	AR	Unknown	608307
CPT1A	Carnitine palmitoyltransferase 1A deficiency	AR	<1/1.000.000	600528
CPT2	Carnitine palmitoyltransferase II deficiency	AR	<1/1.000.000	600650
CRB1	Retinitis pigmentosa	AR	1-5 / 10 000	604210
CRLF1	Cold-induced sweating syndrome	AR	<1 / 1 000 000	604237
CRTAP	Osteogenesis imperfecta, type 2 / type 3 / type 4	AR	Unknown	605497
CSTB	Progressive myoclonic epilepsy, type 1	AR	Unknown	601145
CTH	Cystathioninuria	AR	1-9 / 100 000	607657
CTNS	Cystinosis / Congenital neuronal ceroid lipofuscinosis	AR	1-9 / 100 000	606272
CTSA	Galactosialidosis	XL	Unknown	613111
CTSC	Papillon-Lefevre syndrome	AR	1/1,000,000	602365
CTSD	Infantile neuronal ceroid lipofuscinosis	AR	Unknown	116840
CTSK	Pycnodysostosis	AR	1-9 / 1 000 000	601105
CUL4B	X-linked intellectual disability, Cabezas type	XLR	<1 / 1 000 000	300304
CUL7	3M syndrome	AR	Unknown	609577
CYBA	Chronic granulomatous disease, CYBA-related	AR	1/1,000,000	608508
CYBB	Chronic granulomatous disease, X-linked	XLR	1/200,000	300481
CYP11A1	46,XY disorder of sex development – adrenal insufficiency due to CYP11A1 deficiency	AD/AR	<1 / 1 000 000	118485
CYP11B1	11β-hydroxylase deficiency	AR	1-9 / 1 000 000	610613
CYP11B2	Corticosterone methyloxidase deficiency	AR	1/1,000,000	124081
CYP17A1	17α-hydroxylase deficiency	AR	1-9/100.000	609300
CYP19A1	Aromatase deficiency	AR	<1/1,000,000	107910
CYP1B1	Primary congenital glaucoma	AR	1/15,000	601771
CYP21A2	Congenital adrenal hyperplasia	AR	1-9/100.000	613815
CYP27A1	Sterol 27-hydroxylase deficiency	AR	Unknown	606530
CYP27B1	Vitamin D–dependent hypocalcaemic rickets, type 1A	AR	1-5 / 10 000	609506
CYP2R1	Vitamin D–dependent hypocalcaemic rickets, type 1B	AR	1-5 / 10 000	608713
DBH	Dopamine beta-hydroxylase deficiency	AR	<1 / 1 000 000	609312
DBT	Maple syrup urine disease, type 2	AR	1-9 / 1 000 000	
DCLRE1C	Omenn syndrome, Severe combined immunodeficiency (SCID)	AR	<1 / 1 000 000	605988
DCX	Lissencephaly, type 1, due to doublecortin gene mutation	XLR	Unknown	300121
DDB2	Xeroderma pigmentosum, type E	AR	Unknown	600811
DDC	Aromatic L-amino acid decarboxylase deficiency	AR	<1 / 1 000 000	107930
DECR1	2,4-Dienoyl-CoA reductase deficiency	AR	MICRODELETION	222745
DES	Myofibrillar myopathy, type 1	AD/AR	1-5 / 10 000	125660
DGUOK	Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency	AR	Unknown	601465
DHCR24	Desmosterolosis	AR	<1 / 1 000 000	606418
DHCR7	Smith-Lemli-Opitz syndrome	AR	Unknown	602858
DHDDS	Retinitis pigmentosa	AR	1-5 / 10 000	608172
DKC1	Dyskeratosis congenita	XLR	1-9 / 1 000 000	300126
DLD	Dihydrolipoamide dehydrogenase deficiency	AR	Unknown	238331
DLG3	X-linked non-syndromic intellectual disability	XLR	Unknown	300189
DLL3	Autosomal recessive spondylocostal dysostosis	AR	Unknown	602768
DMD	Duchenne and Becker muscular dystrophy	XLR	1-9 / 100 000	300377
DMP1	Autosomal recessive hypophosphataemic rickets	AR	Unknown	600980
DNAAF1	Primary ciliary dyskinesia, type 13	AR	Unknown	613190
DNAAF2	Primary ciliary dyskinesia, type 10	AR	Unknown	612517
DNAAF3	Primary ciliary dyskinesia, type 2	AR	Unknown	614566
DNAH11	Primary ciliary dyskinesia, type 7	AR	Unknown	603339
DNAH5	Primary ciliary dyskinesia, type 3	AR	Unknown	603335
DNAI1	Kartagener syndrome	AR	Unknown	604366
DNAI2	Primary ciliary dyskinesia, type 9	AR	Unknown	605483
DNAJC19	3-Methylglutaconic aciduria, type V	AR	Unknown	608977
DNAL1	Primary ciliary dyskinesia, type 16	AR	Unknown	610062
DNMT3B	ICF syndrome (Immunodeficiency, Centromeric instability, and Facial anomalies)	AR	<1 / 1 000 000	602900

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DOCK6	Adams-Oliver syndrome 2	AR	Unknown	614194
DOCK8	Combined immunodeficiency due to DOCK8 deficiency	AR	<1 / 1 000 000	611432
DOK7	Congenital myasthenic syndrome	AR	1-9 / 1 000 000	610285
DOLK	Congenital disorder of glycosylation, type 1m	AR	<1 / 1 000 000	610746
DPAGT1	Congenital myasthenic syndrome (duplicate)	AR	1-9 / 1 000 000	191350
DPM1	Congenital disorder of glycosylation, type 1e	AR	<1 / 1 000 000	603503
DPYD	Dihydropyrimidine dehydrogenase deficiency	AR	Unknown	612779
DPYS	Dihydropyrimidinase deficiency	AR	<1 / 1 000 000	613326
DSC2	Arrhythmogenic ventricular dysplasia	AD/AR	Unknown	125645
DSG2	Dilated cardiomyopathy	AD/AR	1-5 / 10 000	125671
DSP	Dilated cardiomyopathy (duplicate)	AD/AR	1-5 / 10 000	125647
DUOX2	Familial thyroid dyshormonogenesis, type 6	AR	1-9 / 100 000	606759
DUOXA2	Familial thyroid dyshormonogenesis, type 5	AR	1-9 / 100 000	612772
DYNC2H1	Short-rib polydactyly syndrome, Verma-Naumoff type	AR	Unknown	603297
DYSF	Limb-girdle muscular dystrophy, type 2B	AR	1-9 / 1 000 000	603009
EBP	Chondrodysplasia punctata, X-linked (MEND syndrome)	XLD, XLR	1-9 / 1 000 000	300205
EDA	X-linked hypohidrotic ectodermal dysplasia	XLR	1-9 / 1 000 000	300451
EDAR	Hypohidrotic ectodermal dysplasia	AR; AD	1/100,000	604095
EFEMP2	Cutis laxa, type 1	AR	<1 / 1 000 000	604633
EFNB1	Craniofrontonasal dysplasia	XLD	Unknown	300035
EIF2AK3	Wolcott-Rallison syndrome	AR	<1 / 1 000 000	604032
EIF2B5	Leukoencephalopathy with vanishing white matter	AR	<1/1,000,000	606686
EMD	Emery-Dreifuss muscular dystrophy 1, X-linked	XLR	<1/1,000,000	300384
ENPP1	Autosomal recessive hypophosphataemic rickets	AR	Unknown	173335
EPM2A	Lafora disease, epilepsy	AR	Unknown	607566
ERBB3	Lethal congenital contracture syndrome, type 2	AR	1-5 / 10 000	190151
ERCC1	COFS syndrome type 4 (Cerebro-oculo-facio-skeletal syndrome, type 4)	AR	<1 / 1 000 000	126380
ERCC2	Xeroderma pigmentosum, group D	AR	Unknown	126340
ERCC3	Xeroderma pigmentosum	AR	Unknown	133510
ERCC4	Cockayne syndrome / Xeroderma pigmentosum, group F	AR	<1 / 1 000 000	133520
ERCC5	Cockayne syndrome / Xeroderma pigmentosum, group G, COFS syndrome type 3	AR	<1 / 1 000 000	133530
ERCC6	Cockayne syndrome, type B / COFS syndrome type 1	AR	<1 / 1 000 000	609413
ERCC8	Cockayne syndrome, type A	AR	Unknown	609412
ESCO2	Roberts syndrome	AR	Unknown	609353
ETFA	Multiple acyl-CoA dehydrogenase deficiency (Glutaric aciduria type IIA)	AR	1-9 / 1 000 000	608053
ETFB	Multiple acyl-CoA dehydrogenase deficiency (Glutaric aciduria type IIB)	AR	1-9 / 1 000 000	130410
ETFDH	Multiple acyl-CoA dehydrogenase deficiency (Glutaric aciduria type IIC)	AR	1-9 / 1 000 000	231675
ETHE1	Ethylmalonic encephalopathy	AR	<1 / 1 000 000	608451
EVC	Ellis–van Creveld syndrome (EVC)	AR	Unknown	604831
EVC2	Ellis–van Creveld syndrome (EVC) (duplicate)	AR	Unknown	607261
EXOSC3	Pontocerebellar hypoplasia, type 1B	AR	1/1,000,000	606489
EYS	Retinitis pigmentosa	AR	1-5 / 10 000	612424
F11	Congenital factor XI deficiency	AD/AR	1-9 / 1 000 000	264900
F2	Congenital factor II deficiency	AR	<1 / 1 000 000	176930
F5	Budd-Chiari syndrome	AR	1-9 / 100 000	612309
F8	Congenital factor VIII deficiency / Haemophilia A	XLR	1-9 / 100 000	300841
F9	Congenital factor IX deficiency / Haemophilia B	XLR	1-9 / 100 000	300746
FAH	Tyrosinaemia, type I	AR	Unknown	613871
FAM126A	Hypomyelination with congenital cataract	AR	<1 / 1 000 000	610531
FAM161A	Retinitis pigmentosa 28	AR	1/1,000,000	613596
FAM20C	Lethal osteosclerotic bone dysplasia	AR	<1 / 1 000 000	611061
FANCA	Fanconi anaemia	AR	1-9 / 1 000 000	607139
FANCC	Fanconi anaemia (duplicate)	AR	1-9 / 1 000 000	613899
FANCG	Fanconi anemia, group G	AR	<1/1,000,000	602956
FASTKD2	Infantile mitochondrial encephalopathy related to FASTKD2	AR	<1 / 1 000 000	612322
FBP1	Fructose 1,6-bisphosphatase deficiency	AR	Unknown	611570
FERMT3	Leukocyte adhesion deficiency, type III	AR	<1 / 1 000 000	607901
FGA	Afib amyloidosis / Familial afibrinogenaemia	AD/AR	Unknown	134820

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FGD1	Aarskog–Scott syndrome	XLR	Unknown	300546
FGD4	Charcot–Marie–Tooth disease, type 4H	AR	<1 / 1 000 000	611104
FH	Fumaric aciduria	AR	<1 / 1 000 000	136850
FHL2	Familial isolated dilated cardiomyopathy	AD/AR	1-5 / 10 000	602633
FKBP10	Osteogenesis imperfecta, types 3 and 4 / Bruck syndrome	AR	<1 / 1 000 000	607063
FKBP14	Ehlers–Danlos syndrome with progressive kyphoscoliosis, myopathy and hearing loss	AR	<1 / 1 000 000	614505
FKRP	Walker–Warburg syndrome	AR	1-9 / 1 000 000	606596
FKTN	Walker–Warburg syndrome	AR	1-9 / 1 000 000	607440
FMR1	Fragile X syndrome, point mutations	XLR	1-5 / 10 000	300805
FOLR1	Neurodegenerative syndrome due to cerebral folate transport deficiency	AR	<1 / 1 000 000	136430
FOXE1	Bamforth–Lazarus syndrome	AR	<1 / 1 000 000	602617
FOXN1	Severe T-cell immunodeficiency with congenital alopecia and nail dystrophy	AR	Unknown	600838
FOXP3	X-linked immunodysregulation polyendocrinopathy enteropathy syndrome (IPEX)	XLR	Unknown	300292
FRAS1	Fraser syndrome	AR	Unknown	607830
FREM2	Fraser syndrome (duplicate)	AR	Unknown	608945
FTCD	Glutamate formiminotransferase deficiency	AR	Unknown	606806
FTL	Hyperferritinaemia	AD/AR	<1 / 1 000 000	134790
FTSJ1	X-linked non-syndromic intellectual disability	XLR	Unknown	300499
FUCA1	Alpha-fucosidase deficiency	AR	Unknown	612280
FXN	Friedreich's ataxia, point mutations	AR	1-9 / 100 000	606829
G6PC	Glycogen storage disease due to glucose-6-phosphatase deficiency, type Ia	AR	Unknown	613742
G6PC3	Severe congenital neutropenia due to G6PC3 deficiency	AR	<1 / 1 000 000	611045
G6PD	Glucose-6-phosphate dehydrogenase deficiency	XL	<1 / 1 000 000	305900
GAA	Glycogen storage disease due to acid maltase deficiency, infantile onset (Pompe disease)	AR	1-9 / 100 000	606800
GALC	Krabbe disease	AR	1-9 / 100 000	606890
GALE	Galactose epimerase deficiency	AR	Unknown	606953
GALK1	Galactokinase deficiency	AR	Unknown	604313
GALNS	Mucopolysaccharidosis type IVA (Morquio A syndrome)	AR	1-5 / 10 000	612222
GALNT3	Hyperphosphatemic familial tumoral calcinosis	AR	1/1,000,000	601756
GALT	Galactosaemia	AR	Unknown	606999
GAMT	Guanidinoacetate methyltransferase deficiency	AR	<1 / 1 000 000	601240
GATAD1	Familial isolated dilated cardiomyopathy	AR	1-5 / 10 000	614518
GBA	Gaucher disease	AR	1-9 / 100 000	606463
GBE1	Glycogen storage disease	AR	Unknown	607839
GCDH	Glutaryl-CoA dehydrogenase deficiency	AR	Unknown	608801
GCH1	Autosomal dominant dopa-responsive dystonia, BH4 deficiency	AR/AD	1-9 / 1 000 000	600225
GCSH	Glycine encephalopathy	AR	1-9 / 1 000 000	238330
GDAP1	Charcot–Marie–Tooth disease	AR/AD	1-5/10 000	606598
GDF5	Grebe syndrome	AR	1/1,000,000	601146
GDI1	X-linked non-syndromic intellectual disability (duplicate)	XLR	Unknown	300104
GFM1	Combined oxidative phosphorylation deficiency 1	AR	Unknown	606639
GH1	Isolated growth hormone deficiency, type IA/II	AR; AD	1/100,000	139250
GHRHR	Isolated growth hormone deficiency, type IB	AR	1/100,000	600962
GJB1	Hereditary deafness, type 1A	AR	1/1 000	304040
GJB2	Hereditary deafness, type 1B	AR	1/1 000	121011
GJB3	Non-syndromic genetic deafness	AR/AD	1/1 000	603324
GJB6	Non-syndromic genetic deafness (duplicate)	AR/AD	1/1 000	604418
GJC2	Autosomal recessive spastic paraplegia, type 44	AR	Unknown	608803
GLA	Fabry disease	XL	1-5/10.000	300644
GLB1	GM1 gangliosidosis, type 1	AR	Unknown	611458
GLDC	Glycine encephalopathy (duplicate)	AR	1-9/1 000 000	238300
GLE1	Arthrogryposis–anterior horn cell disease syndrome	AR	<1/1.000.000	603371
GLIS3	Neonatal diabetes mellitus	AR	Unknown	610192
GNE	Inclusion body myopathy 2	AR	<1/1,000,000	603824
GNMT	Glycine N-methyltransferase deficiency	AR	Unknown	606628
GNPAT	Rhizomelic chondrodysplasia punctata, type 2	AR	Unknown	602744
GNPTAB	Mucolipidosis type II	AR	<1/10.000-1.000.000	607840
GNPTG	Mucolipidosis type II (duplicate)	AR	<1/10.000-1.000.000	607838

GNRHR	Normosmic congenital hypogonadotropic hypogonadism	AR	Unknown	138850
GNS	Sanfilippo syndrome, type D (Mucopolysaccharidosis type III)	AR	1-9/1.000.000	607664
GORAB	Geroderma osteodysplasticum	AR	<1 / 1 000 000	607983
GP1BA	Bernard-Soulier syndrome, type A2	AR	1/1,000,000	606672
GP1BB	Bernard–Soulier syndrome	AR	<1 / 1 000 000	138720
GP9	Bernard-Soulier syndrome, type C	AR	1/1,000,000	173515
GPC3	Simpson–Golabi–Behmel syndrome	XLR	Unknown	300037
GPR143	X-linked recessive ocular albinism	XLR	1-9 / 1 000 000	300808
GRHPR	Primary hyperoxaluria, type 2	AR	1/1,000,000	604296
GRIA3	X-linked intellectual disability due to GRIA3 mutations	XLR	<1 / 1 000 000	305915
GRIK2	Autosomal recessive non-syndromic intellectual disability	AR	Unknown	138244
GSS	Glutathione synthetase deficiency	AR	<1 / 1 000 000	601002
GTF2H5	Trichothiodystrophy	AR	Unknown	608780
GUCY2D	Cone-rod dystrophy	AR/AD	1-9 / 100 000	600179
GUSB	Mucopolysaccharidosis type VII	AR	<1 / 1 000 000	611499
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency	AR	Unknown	601609
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	AR	1-9/100.000	600890
HADHB	Mitochondrial trifunctional protein deficiency	AR	1-9/100.000	143450
HAL	Histidinemia	AR	1-9 / 100 000	609457
HAMP	Hemochromatosis	AR	<1/1.000.000	606464
HAX1	Kostmann syndrome	AR	<1 / 1 000 000	605998
HBA1	Alpha Thalassemia	AR	1/10 000	141800
HBA2	Alpha Thalassemia	AR	1/10 000	141850
HBB	Beta-thalassemia	AR	1-9/1.000.000	141900
HESX1	Septo-optic dysplasia	AR/AD	Unknown	601802
HEXA	Tay-Sachs disease	AR	1/320.000	606869
HEXB	Sandhoff disease	AR	1-9 / 1 000 000	606873
HFE	Hemochromatosis, type 1	AR	1/200	613609
HFE2	Hemochromatosis, type 2A	AR	1/1,000,000	608374
HGD	Alkaptonuria	AR	1-9 / 1 000 000	607474
HGSNAT	Sanfilippo syndrome type C	AR	1-9/1.000.000	610453
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency	AR	<1 / 1 000 000	610690
HLCS	Holocarboxylase synthetase deficiency	AR	Unknown	609018
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	AR	Unknown	613898
HMGCS2	3-hydroxy-3-methylglutaryl-CoA synthase deficiency	AR	<1 / 1 000 000	600234
HMOX1	Heme oxygenase-1 deficiency	AR	1/1,000,000	141250
HOGA1	Hyperoxaluria, primary, type III	AR	<1 / 1 000 000	613597
HPD	Tyrosinemia, type III	AR	<1 / 1 000 000	609695
HPRT1	Kelley-Seegmiller syndrome/Lesch-Nyhan syndrome	AR	1-9 / 1 000 000	308000
HPS1	Hermansky-Pudlak syndrome 1	AR	1/500,000	604982
HPS3	Hermansky-Pudlak syndrome 3	AR	1/500,000	606118
HPS4	Hermansky-Pudlak syndrome 4	AR	1/500,000	606682
HSD11B2	11-beta-hydroxysteroid dehydrogenase deficiency, type 2	AR	<1 / 1 000 000	614232
HSD17B10	17-beta-hydroxysteroid dehydrogenase deficiency	XL	<1 / 1 000 000	300256
HSD17B3	46,XY difference of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	AR	Unknown	605573
HSD17B4	Perrault syndrome 1/D-bifunctional protein deficiency	AR	<1 / 1 000 000	601860
HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR	<1 / 1 000 000	613890
HSPG2	Dyssegmental dysplasia, Silverman-Handmaker type/ Schwartz-Jampel syndrome	AR	<1 / 1 000 000	142461
HUWE1	Non-specific syndromic intellectual disability	XL	Unknown	300697
HYAL1	Hyaluronidase deficiency,Mucopolysaccharidosis type 9	AR	<1 / 1 000 000	607071
HYDIN	Ciliary dyskinesia, primary, 5	AR	Unknown	610812
HYLS1	Hydrolethalus syndrome	AR	<1/1,000,000	610693
ICOS	Common variable immunodeficiency	AR	1-9 / 100 000	604558
IDH3B	Retinitis pigmentosa	AR	1-5 / 10 000	604526
IDS	Mucopolysaccharidosis II	XLR	1-9/1.000.000	300823
IDUA	Hurler-Scheie syndrome	AR	1-9 / 1 000 000	252800
IFNGR1	Mendelian susceptibility to mycobacterial diseases	AR	<1 / 1 000 000	107470
IFNGR2	Mendelian susceptibility to mycobacterial diseases	AR	<1 / 1 000 000	147569

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IFT80	Asphyxiating thoracic dystrophy of the newborn	AR	Unknown	611177
IGBP1	Corpus callosum agenesis	XLR	<1 / 1 000 000	300139
IGHM	Agammaglobulinemia 1	AR	<1 / 1 000 000	147020
IGHMBP2	Charcot-Marie-Tooth disease type 2S	AR	<1 / 1 000 000	600502
IGLL1	Agammaglobulinemia 2	AR	<1 / 1 000 000	146770
IHH	Acrocapitofemoral dysplasia	AR	<1 / 1 000 000	600726
IKBKAP	Dysautonomia, familial (IKBKAP or ELP1)	AR	1/30,000	603722
IKBKG	Ectodermal dysplasia and immunodeficiency 1/ Incontinentia pigmenti	AR/AD	1-9 / 1 000 000	300248
IL12B	Mendelian susceptibility to mycobacterial diseases	AR	<1 / 1 000 000	161561
IL12RB1	Mendelian susceptibility to mycobacterial diseases	AR	<1 / 1 000 000	601604
IL17RA	Chronic mucocutaneous candidiasis	AR	Unknown	605461
IL1RAPL1	X-linked non-syndromic intellectual disability	XLR	Unknown	300206
IL1RN	Sterile multifocal osteomyelitis with periostitis and pustulosis	AR	<1 / 1 000 000	147679
IL2RA	Interleukin-2 receptor alpha deficiency (Immunodeficiency)	AR	<1 / 1 000 000	147730
IL2RG	X-linked combined immunodeficiency	XL	1-9/1.000.000	308380
IL7R	T– B+ severe combined immunodeficiency due to IL-7Rα deficiency	AR	Unknown	146661
INVS	Infantile nephronophthisis	AR	Unknown	243305
IQCB1	Senior–Løken syndrome	AR	1-9 / 1 000 000	609237
IQSEC2	X-linked non-syndromic intellectual disability	XLR	Unknown	300522
ITGA6	Epidermolysis bullosa with pyloric atresia	AR	<1 / 1 000 000	147556
ITGB3	Glanzmann thrombasthenia	AR	1/200,000	173470
ITGB4	Epidermolysis bullosa	AR	<1 / 1 000 000	147557
IVD	Isovaleric acidaemia	AR	1-9/1.000.000	607036
IYD	Thyroid dyshormonogenesis type 4	AR	1-9 / 100 000	612025
JAK3	T– B+ severe combined immunodeficiency due to JAK3 deficiency	AR	Unknown	600173
JPH2	Familial isolated dilated cardiomyopathy	AR/AD	1-5 / 10 000	605267
KCNJ1	Bartter syndrome type 2	AR	Unknown	600359
KDM5C	X-linked syndromic intellectual disability	AR	<1 / 1 000 000	314690
L1CAM	X-linked complex agenesis of the corpus callosum	XLR	1/30.000	308840
LAMA2	Congenital muscular dystrophy, type 1A	AR	1-9 / 1 000 000	156225
LAMA3	Epidermolysis bullosa	AR	1-9 / 1 000 000	600805
LAMB2	Congenital myasthenic syndrome	AR	1-9 / 1 000 000	150325
LAMB3	Epidermolysis bullosa (duplicate)	AR	1-9 / 1 000 000	150310
LAMC2	Epidermolysis bullosa (duplicate)	AR	1-9 / 1 000 000	150292
LAMP2	Danon disease	XLR	<1 / 1 000 000	309060
LBR	Greenberg dysplasia	AR	<1 / 1 000 000	600024
LCA5	Leber congenital amaurosis, type LCA5	AR	1/250,000	611408
LCT	Congenital lactase deficiency	AR	Unknown	603202
LDHA	Glycogen storage disease, type XI	AR	Unknown	150000
LDLR	Familial hypercholesterolaemia	AR/AD	1-9 / 1 000 000	606945
LDLRAP1	Familial hypercholesterolaemia (duplicate)	AR	1-9 / 1 000 000	605747
LEPRE1	Osteogenesis imperfecta, types 2 and 3	AR	1-5 / 10 000	610339
LHCGR	Leydig cell hypoplasia due to LH resistance	AR	Unknown	152790
LHX3	Hypothyroidism, type 3	AR	Unknown	600577
LIFR	Stüve–Wiedemann syndrome	AR	<1 / 1 000 000	151443
LIG4	Dubowitz syndrome	AR	Unknown	601837
LIPA	Wolman disease / Cholesteryl ester storage disease	AR	Unknown	613497
LIPH	Woolly hair/hypotrichosis syndrome	AR	1/1,000,000	607365
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	AR	<1 / 1 000 000	612625
LOXHD1	Deafness, autosomal recessive 77	AR	1/1,000,000	613072
LPL	Lipoprotein lipase deficiency	AR	1/1,000,000	238600
LRAT	Retinite Pigmentosa giovanile, tipo 14	AR	1-5 / 10 000	604863
LRP1	Siemens follicular keratosis spinulosa decalvans	AR	<1 / 1 000 000	107770
LRP2	Donnai–Barrow syndrome	AR	<1 / 1 000 000	600073
LRP5	Exudative vitreoretinopathy, type 4	AR	Unknown	603506
LRPPRC	Congenital lactic acidosis, Saguenay–Lac-Saint-Jean type	AR	Unknown	614930
LRRC6	Primary ciliary dyskinesia, type 19	AR	Unknown	614930
LYST	Chédiak–Higashi syndrome	AR	Unknown	606897

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MAN2B1	Alpha-mannosidosis, types I and II	AR	1-9 / 1 000 000	609458
MANBA	Beta-mannosidosis	AR	Unknown	609489
MAT1A	Methionine adenosyltransferase deficiency	AR	<1 / 1 000 000	610550
MBTPS2	IFAP syndrome (Follicular ichthyosis–atrichia–photophobia) / Osteogenesis imperfecta	XLR	<1 / 1 000 000	300294
MCCC1	3-Methylcrotonyl-CoA carboxylase deficiency	AR	1-9 / 100 000	609010
MCCC2	3-Methylcrotonyl-CoA carboxylase deficiency	AR	1-9 / 100 000	609014
MCEE	Methylmalonyl-CoA epimerase deficiency	AR	<1 / 1 000 000	608419
MCOLN1	Mucolipidosis IV	AR	Unknown	605248
MCPH1	Primary microcephaly 1	AR	Unknown	607117
MECP2	Rett syndrome	XLD	1-9 / 100 000	300005
MED12	Opitz-Kaveggia syndrome (FGS1), Lujan-Fryns syndrome	XLR	Unknown	300188
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy	AR	1/1,000,000	603811
MEFV	Familial Mediterranean fever	AR/AD	1-5 / 10 000	608107
MEGF10	Congenital myopathy	AR	<1 / 1 000 000	612453
MESP2	Spondylothoracic dysostosis, MESP2-related	AR	<1/1,000,000	605195
MFSD8	Ceroid lipofuscinosis	AR	Unknown	611124
MGAT2	N-acetylglucosaminyltransferase 2 deficiency	AR	<1 / 1 000 000	602616
MID1	Opitz GBBB syndrome	XLR	Unknown	300552
MKKS	Bardet–Biedl syndrome	AR	Unknown	604896
MKS1	Meckel syndrome, type 1	AR	<1 / 1 000 000	609883
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	AR	<1 / 1 000 000	605908
MLYCD	Malonyl-CoA decarboxylase deficiency	AR	<1 / 1 000 000	606761
MMAA	Methylmalonic acidaemia, type cblA	AR	<1 / 1 000 000	607481
MMAB	Methylmalonic acidaemia, type cblB	AR	Unknown	607568
MMACHC	Methylmalonic aciduria with homocystinuria, type cblC	AR	Unknown	609831
MMADHC	Methylmalonic aciduria with homocystinuria, type cblD	AR	<1 / 1 000 000	611935
MOCOS	Hereditary xanthinuria, type II	AR	Unknown	613274
MOCS1	Sulfite oxidase deficiency due to molybdenum cofactor deficiency, type A	AR	<1 / 1 000 000	603707
MOCS2	Sulfite oxidase deficiency due to molybdenum cofactor deficiency, type B	AR	Unknown	603708
MOGS	Congenital disorder of glycosylation, type IIb	AR	<1 / 1 000 000	601336
MPDU1	Congenital disorder of glycosylation, type If	AR	<1 / 1 000 000	604041
MPI	Congenital disorder of glycosylation, type Ib	AR	<1 / 1 000 000	154550
MPL	Hereditary isolated aplastic anaemia	AD/AR	Unknown	159530
MPV17	Navajo neuropathy	AR	<1 / 1 000 000	137960
MRPS16	Combined oxidative phosphorylation deficiency, type 2	AR	<1 / 1 000 000	609204
MRPS22	Gonadal dysgenesis 46,XX; Hypotonia with lactic acidaemia and hyperammonaemia	AR	Unknown	605810
MTHFD1	Severe combined immunodeficiency	AR	Unknown	172460
MTHFR	Homocystinuria	AR	Unknown	607093
MTM1	X-linked myotubular myopathy	XLR	1-9/1.000.000	300415
MTMR2	Charcot–Marie–Tooth disease, type 4B1	AR	<1 / 1 000 000	603557
MTR	Methylmalonic acidaemia, type cblG	AR	<1 / 1 000 000	156570
MTRR	Homocystinuria–megaloblastic anaemia, type cblE	AR	<1 / 1 000 000	602568
MTTP	Abetalipoproteinaemia	AR	<1 / 1 000 000	157147
MUT	Methylmalonic aciduria, type mut(0)	AR	<1/1,000,000	609058
MVK	Mevalonate kinase deficiency	AR	<1 / 1 000 000	251170
MYD88	Recurrent pyogenic bacterial infections due to MyD88 deficiency	AR	Unknown	602170
MYH7	Hypertrophic cardiomyopathy, type 1, with myopathy	AD/AR	1-5 / 10 000	160760
MYL2	Congenital fibre-type disproportion myopathy	AD/AR	Unknown	160781
MYO15A	Deafness, autosomal recessive, 3	AR	1/1,000,000	602666
MYO18B	Klippel–Feil anomaly–myopathy–facial dysmorphism syndrome	AR	<1 / 1 000 000	607295
MYO5A	Griscelli syndrome	AR	<1 / 1 000 000	160777
MYO7A	Usher syndrome, type 1	AR	1-9 / 100 000	276903
MYPN	Familial isolated dilated cardiomyopathy (duplicate)	AD/AR	1-5 / 10 000	608517
NAGA	Alpha-N-acetylgalactosaminidase deficiency	AR	<1 / 1 000 000	104170
NAGLU	Sanfilippo syndrome, type B (Mucopolysaccharidosis type IIIB)	AR	1-9 / 100 000	609701
NAGS	Hyperammonaemia due to N-acetylglutamate synthase deficiency	AR	<1 / 1 000 000	608300
NBN	Nijmegen breakage syndrome	AR	1/330 000	602667
NDP	Norrie disease	XLR	Unknown	300658

NDRG1	Charcot–Marie–Tooth disease, type 4D	AR	Unknown	605262
NDUFA1	Isolated complex I deficiency	XLR	Unknown	300078
NDUFAF2	Isolated complex I deficiency	AR	Unknown	609653
NDUFAF4	Isolated complex I deficiency	AR	Unknown	611776
NDUFAF5	Mitochondrial complex I deficiency, NDUFAF5-related	AR	1/1,000,000	612360
NDUFS3	Isolated complex I deficiency	AR	Unknown	603846
NDUFS4	Isolated complex I deficiency	AR	Unknown	602694
NDUFS6	Isolated complex I deficiency	AR	Unknown	603848
NDUFS7	Isolated complex I deficiency	AR	Unknown	601825
NDUFS8	Isolated complex I deficiency	AR	Unknown	602141
NDUFV1	Isolated complex I deficiency	AR	Unknown	161015
NEB	Lethal multiple pterygium syndrome	AR	<1 / 1 000 000	161650
NEU1	Sialidosis, types I and II	AR	<1 / 1 000 000	608272
NEUROG3	Enteric anendocrinosis	AR	<1 / 1 000 000	604882
NFKBIA	X-linked hypohidrotic ectodermal dysplasia with immunodeficiency	AR	Unknown	164008
NHEJ1	Combined immunodeficiency with microcephaly, growth retardation, and T- and B-cell lymphopenia	AR	<1 / 1 000 000	611290
NHLRC1	Lafora disease	AR	Unknown	608072
NHS	Nance–Horan syndrome	XLD	Unknown	300457
NLRP7	Hydatidiform mole, recurrent	AR	1/100,000	609661
NME8	Primary ciliary dyskinesia, type 6	AR	Unknown	607421
NPC1	Niemann–Pick disease, type C1	AR	1-9 / 100 000	607623
NPC2	Niemann–Pick disease, type C2	AR	1-9 / 100 000	601015
NPHP1	Joubert syndrome, type 4	AR	Unknown	607100
NPHP3	Nephronophthisis	AR	Unknown	608002
NPHP4	Nephronophthisis (duplicate)	AR	Unknown	607215
NPHS1	Nephrotic syndrome, type 1	AR	Unknown	602716
NPHS2	Nephrotic syndrome, type 2	AR	Unknown	604766
NR0B1	Congenital adrenal hypoplasia, X-linked	XLR	<1/1,000,000	300473
NR2E3	Retinitis pigmentosa	AR/AD	1-5 / 10 000	604485
NSUN2	Dubowitz syndrome	AR	Unknown	610916
NTRK1	Hereditary sensory and autonomic neuropathy type 4 (HSAN4)	AR	Unknown	191315
NUP62	Familial infantile bilateral striatal necrosis	AR	<1 / 1 000 000	605815
OAT	Gyrate atrophy of the choroid and retina with or without hyperornithinaemia	AR	Unknown	613349
OBSL1	3M syndrome	AR	Unknown	610991
OCA2	Oculocutaneous albinism, type 2	AR	1-9 / 100 000	611409
OCRL	Oculo–cerebro–renal syndrome of Lowe	AR	1-9 / 1 000 000	300535
OFD1	Joubert syndrome, type 10	XLR	Unknown	300170
OPA3	3-Methylglutaconic aciduria, type III	AR	Unknown	606580
OPHN1	X-linked intellectual disability with cerebellar hypoplasia	XLD	<1 / 1 000 000	300127
ORAI1	Combined immunodeficiency due to ORAI1 deficiency	AR	<1 / 1 000 000	610277
OSTM1	Osteopetrosis with neuroaxonal dysplasia, infantile form	AR	<1 / 1 000 000	607649
OTC	Ornithine transcarbamylase deficiency	XL	1-9 / 100 000	300461
OTOF	Auditory neuropathy, type 1	AR	Unknown	603681
OXCT1	Succinyl-CoA:acetoacetate transferase deficiency	AR	<1 / 1 000 000	601424
PAH	Phenylketonuria	AR	1-9 / 100 000	612349
PAK3	X-linked non-syndromic intellectual disability	XLR	Unknown	300142
PANK2	Pantothenate kinase-associated neurodegeneration (PKAN)	AR	1-9 / 1 000 000	606157
PAX3	Craniofacial–deafness–hand syndrome / Waardenburg syndrome, type 1 / Waardenburg syndrome, type 3	AD/AR	1-9 / 100 000	606597
PC	Pyruvate carboxylase deficiency	AR	Unknown	608786
PCBD1	Hyperphenylalaninaemia	AR	Unknown	126090
PCCA	Propionic acidaemia	AR	1-9 / 1 000 000	232000
PCCB	Propionic acidaemia (duplicate)	AR	1-9 / 1 000 000	232050
PCDH15	Usher syndrome, type 1	AR	1-9 / 1 000 000	605514
PCDH19	Female-limited epilepsy with intellectual disability	XL	<1 / 1 000 000	300460
PDE6A	Retinitis pigmentosa	AR	1-5 / 10 000	180071
PDHA1	Pyruvate dehydrogenase deficiency	XLD	Unknown	300502
PDHB	Pyruvate dehydrogenase deficiency (duplicate)	AR	Unknown	179060
PDHX	Pyruvate dehydrogenase deficiency (duplicate)	AR	Unknown	608769
PDP1	Pyruvate dehydrogenase deficiency (duplicate)	AR	Unknown	605993

PDSS1	Coenzyme Q10 deficiency	AR	<1 / 1 000 000	607429
PDSS2	Coenzyme Q10 deficiency (duplicate)	AR	<1 / 1 000 000	610564
PEPD	Prolidase deficiency	AR	Unknown	613230
PEX1	Neonatal adrenoleukodystrophy	AR	Unknown	602136
PEX10	Peroxisome biogenesis disorder, 6A (Zellweger spectrum), 6B	AR	1/50 000	602859
PEX12	Peroxisome biogenesis disorder, 3A (Zellweger spectrum), 3B	AR	1/50 000	601758
PEX13	Peroxisome biogenesis disorder, 11A (Zellweger spectrum), 11B	AR	1/50 000	601789
PEX2	Peroxisome biogenesis disorder, 5A (Zellweger spectrum), 5B		1/50 000	170993
PEX26	Zellweger syndrome	AR	1/50 000	608666
PEX3	Peroxisome biogenesis disorder, 10A (Zellweger spectrum), 10B	AR	1/50 000	603164
PEX5	Rhizomelic chondrodysplasia punctata, type 5 / Peroxisome biogenesis disorder, type 2 (Zellweger spectrum), 2B	AR	1/50 000	600414
PEX6	Peroxisome biogenesis disorder, 4A (Zellweger spectrum), 4B	AR	1/50 000	601498
PEX7	Rhizomelic chondrodysplasia punctata, type 1 / Peroxisome biogenesis disorder, type 9	AR	1/50 000	601757
PFKM	Glycogen storage disease, type VII	AR	<1 / 1 000 000	610681
PGAM2	Glycogen storage disease, type X (myopathic form)	AR	<1 / 1 000 000	612931
PGK1	Phosphoglycerate kinase deficiency	XLR	<1 / 1 000 000	311800
PHF6	Borjeson–Forssman–Lehmann syndrome	XLR	<1 / 1 000 000	300414
PHF8	X-linked intellectual disability, Siderius type	XLR	<1 / 1 000 000	300560
PHGDH	Phosphoglycerate dehydrogenase deficiency	AR	<1/1,000,000	606879
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1	AR	<1/1,000,000	606097
PKHD1	Autosomal recessive polycystic kidney disease	AR	Unknown	606702
PKLR	Hemolytic anaemia due to erythrocyte pyruvate kinase deficiency	AR	1-9 / 100 000	609712
PLA2G6	Infantile neuroaxonal dystrophy	AR	Unknown	603604
PLEC	Congenital myasthenic syndrome	AR	<1 / 1 000 000	601282
PLEKHG5	Charcot–Marie–Tooth disease, intermediate type C / Infantile-onset lower motor neuron disease	AR	<1 / 1 000 000	611101
PLG	Congenital plasminogen deficiency	AR	1-9 / 100 000	173350
PLOD1	Ehlers–Danlos syndrome	AR	Unknown	153454
PLOD2	Osteogenesis imperfecta, type 3	AR	<1 / 1 000 000	601865
PLP1	Pelizaeus–Merzbacher disease	XLR	1-9 / 1 000 000	300401
PMM2	Congenital disorder of glycosylation, type Ia	AR	Unknown	601785
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency	AR	<1 / 1 000 000	164050
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency	AR	Unknown	603287
POLG	Alpers–Huttenlocher syndrome	AR	<1 / 1 000 000	174763
POLH	Xeroderma pigmentosum variant	AR	1/1,000,000	603968
POLR1C	Treacher Collins syndrome, type 3	AR	1-9 / 100 000	610060
POMGNT1	Walker–Warburg syndrome	AR	1-9 / 1 000 000	606822
POMT1	Walker–Warburg syndrome	AR	1-9 / 1 000 000	607423
POMT2	Walker–Warburg syndrome	AR	1-9 / 1 000 000	607439
POR	Antley–Bixler syndrome, type 2	AR	1-9 / 100 000	124015
POU1F1	Alpers–Huttenlocher syndrome	AR	Unknown	173110
POU3F4	Ayazi syndrome	XLR	<1 / 1 000 000	300039
PPIB	Osteogenesis imperfecta, types 2, 3, and 4	AR	1-5 / 10 000	123841
PPT1	Neuronal ceroid lipofuscinosis, type 1	AR	Unknown	600722
PQBP1	Renpenning syndrome	XLR	<1 / 1 000 000	605557
PRDM5	Ehlers–Danlos syndrome, type 6B	AR	<1 / 1 000 000	605557
PREPL	Myasthenic syndrome, congenital, 22	AR	<1/1,000,000	609557
PRF1	Familial haemophagocytic lymphohistiocytosis	AR	Unknown	170280
PRODH	Hyperprolinemia type 1	AR	Unknown	606810
PROP1	Pituitary hormone deficiency, combined, 2	AR	Unknown	601538
PRPS1	X-linked Charcot-Marie-Tooth disease type 5	XLR	<1 / 1 000 000	311850
PRSS12	Intellectual developmental disorder, autosomal recessive 1	AR	Unknown	606709
PRX	Charcot-Marie-Tooth disease type 4F	AR	<1 / 1 000 000	605725
PSAP	Atypical Gaucher disease due to saposin C deficiency, Krabbe disease, Encephalopathy due to prosaposin deficiency	AR	1-9 / 100 000	176801
PTH1R	Chondrodysplasia, Blomstrand type	AR	<1 / 1 000 000	168468
PTPRC	T-B+ severe combined immunodeficiency due to CD45 deficiency	AR	1 / 1 000 000	151460
PTS	Hyperphenylalaninemia, BH4-deficient, A	AR	1-9 / 1 000 000	612719
PUS1	Mitochondrial myopathy and sideroblastic anemia (MLASA1)	AR	<1/1,000,000	608109
PYGM	Glycogen storage disease due to muscle glycogen phosphorylase deficiency	AR	Unknown	608455

PrenatalAutism - Elenco dei geni e delle malattie genetiche a trasmissione ereditaria investigati

RAB23	Carpenter syndrome	AR	<1 / 1 000 000	606144
RAB27A	Griscelli syndrome	AR	<1 / 1 000 000	603868
RAB39B	Rare non-syndromic intellectual disability	XLR	Unknown	300774
RAB3GAP1	Micro syndrome/ Martsolf syndrome 2	AR	<1 / 1 000 000	602536
RAB3GAP2	Micro syndrome/ Martsolf syndrome 1	AR	<1 / 1 000 000	609275
RAG1	Combined immunodeficiency with granulomatosis, Omenn syndrome	AR	<1 / 1 000 000	179615
RAG2	Combined immunodeficiency with granulomatosis, Omenn syndrome	AR	<1 / 1 000 000	179616
RAPSN	Congenital myasthenic syndrome	AR	1-9 / 1 000 000	601592
RARS2	Pontocerebellar hypoplasia, type 1 and 6, RARS2-related	AR	1/1,000,000	611524
RAX	Colobomatous microphthalmia	AR	Unknown	601881
RBM8A	Thrombocytopenia-absent radius syndrome	AR	Unknown	605313
RDH12	Leber congenital amaurosis/Retinitis pigmentosa	AR/AD	1-9 / 100 000	608830
RECQL4	Baller-Gerold syndrome	AR	<1 / 1 000 000	603780
RELN	Epilepsy with auditory features/Microlissencephaly	AD/AR	Unknown	600514
RFT1	Congenital disorder of glycosylation, type In	AR	<1 / 1 000 000	611908
RLBP1	Retinal dystrophies, RLBP1-associated	AR	1/1,000,000	180090
RMRP	Omenn syndrome	AR	<1 / 1 000 000	157660
RNASEH2A	Aicardi-Goutières syndrome	AR	Unknown	606034
RNASEH2B	Aicardi-Goutières syndrome	AR	Unknown	610326
RNASEH2C	Aicardi-Goutières syndrome	AR	Unknown	610330
RP1	Retinitis pigmentosa 1	AR/AD	1-5 / 10 000	603937
RP2	Retinitis pigmentosa 2	XL	1-5 / 10 000	300757
RPE65	Retinitis pigmentosa 20	AR	1-5 / 10 000	180069
RPGR	Retinitis pigmentosa 3	XL	1-5 / 10 000	312610
RPGRIP1L	Joubert syndrome 7	AR	<1 / 1 000 000	610937
RPL10	Intellectual developmental disorder, X-linked syndromic 35	XLR	<1 / 1 000 000	312173
RPS6KA3	Coffin-Lowry syndrome	XLD	1-9 / 100 000	300075
RRM2B	Mitochondrial neurogastrointestinal encephalomyopathy	AR	1-9 / 1 000 000	604712
RS1	X-linked retinoschisis	XLR	1-9 / 100 000	300839
RSPH4A	Ciliary dyskinesia, primary, 11	AR	Unknown	612647
SACS	Spastic ataxia, Charlevoix-Saguenay type	AR	Unknown	604490
SAMD9	MIRAGE syndrome	AD	1/1,000,000	610456
SAMHD1	Aicardi-Goutières syndrome	AR	Unknown	606754
SBDS	Shwachman-Diamond syndrome 1	AR	1-9 / 1 000 000	607444
SCN4A	Myasthenic syndrome, congenital, 16	AR	1-9 / 1 000 000	603967
SCO1	Fatal infantile cytochrome C oxidase deficiency	AR	Unknown	603644
SCO2	Autosomal recessive axonal Charcot-Marie-Tooth disease due to copper metabolism defect, Fatal infantile cytochrome C oxidase deficiency	AR	<1 / 1 000 000	604272
SDHA	Isolated succinate-CoQ reductase deficiency	AR	<1 / 1 000 000	600857
SEPSECS	Pontocerebellar hypoplasia, type 2D	AR	1/1,000,000	613009
SERPINA1	Alpha-1-antitrypsin deficiency	AR	1-5 / 10 000	107400
SERPINA7	Deficit ereditario della tireoglobulina	XL		314200
SERPINF1	Osteogenesis imperfecta, type VI	AR	Unknown	172860
SERPINH1	Osteogenesis imperfecta type 3	AR	Unknown	600943
SFTPB	Surfactant metabolism dysfunction, pulmonary, 1	AR	Unknown	178640
SGCA	Alpha-sarcoglycan-related limb-girdle muscular dystrophy R3	AR	Unknown	600119
SGCB	Beta-sarcoglycan-related limb-girdle muscular dystrophy R4	AR	1-9 / 1 000 000	600900
SGCD	Delta-sarcoglycan-related limb-girdle muscular dystrophy R6	AR	1-9 / 1 000 000	601411
SGCG	Limb-girdle muscular dystrophy, type 2C	AR	1/200,000	608896
SGSH	Sanfilippo syndrome type A	AR	1-9 / 1 000 000	605270
SH2D1A	X-linked lymphoproliferative disease due to SAP deficiency	XLR	<1 / 1 000 000	300490
SH3TC2	Charcot-Marie-Tooth disease type 4C	AR	Unknown	608206
SHROOM4	X-linked intellectual disability, Stocco Dos Santos type	XLR	<1 / 1 000 000	300579
SIL1	Marinesco-Sjogren syndrome	AR	<1 / 1 000 000	608005
SIX6	Colobomatous optic disc-macular atrophy-chorioretinopathy syndrome	AR	<1 / 1 000 000	606326
SLC12A1	Bartter syndrome, type 1	AR	Unknown	600839
SLC12A3	Gitelman syndrome	AR	1/40,000	600968
SLC12A6	Corpus callosum agenesis-neuronopathy syndrome	AR	<1 / 1 000 000	604878
SLC16A2	Allan-Herndon-Dudley syndrome	XLR	Unknown	300095

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SLC17A5	Salla disease	AR	<1 / 1 000 000	604322
SLC19A2	Megaloblastic anemia syndrome	AR	<1/1,000,000	603347
SLC22A5	Carnitine deficiency, systemic primary	AR	Unknown	603377
SLC25A13	Citrullinemia type II	AR	Unknown	603859
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	AR	Unknown	603861
SLC25A20	Deficit di carnitina-acilcarnitinatraslocasi	AR	<1 / 1 000 000	613698
SLC25A22	Early infantile developmental and epileptic encephalopathy	AR	Unknown	609302
SLC26A2	Achondrogenesis 1B	AR	Unknown	606718
SLC26A3	Congenital chloride diarrhea	AR	<1/1,000,000	126650
SLC26A4	Rare autosomal recessive non-syndromic sensorineural deafness type DFNB	AR	Unknown	605646
SLC2A1	Classic glucose transporter type 1 deficiency syndrome	AD/AR	1-9 / 1 000 000	138140
SLC35A1	Congenital disorder of glycosylation, type IIf	AR	<1 / 1 000 000	605634
SLC35C1	Congenital disorder of glycosylation, type IIc	AR	<1 / 1 000 000	605881
SLC35D1	Schneckenbecken dysplasia	AR	<1 / 1 000 000	610804
SLC37A4	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib	AR	Unknown	602671
SLC39A4	Acrodermatitis enteropathica	AR	Unknown	607059
SLC3A1	Cystinuria	AD/AR	1-5 / 10 000	104614
SLC45A2	Oculocutaneous albinism, type 4	AR	1/100,000	606202
SLC46A1	Hereditary folate malabsorption	AR	<1 / 1 000 000	611672
SLC4A11	Corneal dystrophy-perceptive deafness syndrome	AR	<1 / 1 000 000	610206
SLC5A5	Familial thyroid dyshormonogenesis	AR	1-9 / 100 000	601843
SLC6A19	Hartnup disease	AR	1-9 / 100 000	608893
SLC6A8	X-linked creatine transporter deficiency	XLR	Unknown	300036
SLC7A7	Lysinuric protein intolerance	AR	Unknown	603593
SLC7A9	Cystinuria	AD/AR	1-5 / 10 000	604144
SLC9A6	Christianson syndrome	XLR	1-9 / 100 000	300231
SMARCAL1	Schimke immunoosseous dysplasia	AR	<1/1,000,000	606622
SMN1	Proximal spinal muscular atrophy	AR	1-9 / 100 000	600354
SMPD1	Niemann-Pick disease	AR	Unknown	607608
SMS	X-linked intellectual disability, Snyder type	XLR	<1 / 1 000 000	300105
SNAP29	CEDNIK syndrome	AR	<1 / 1 000 000	604202
SP110	Hepatic veno-occlusive disease-immunodeficiency syndrome	AR	<1 / 1 000 000	604457
SP7	Osteogenesis imperfecta, type XII	AR	Unknown	606633
SPATA7	Retinitis pigmentosa 94, variable age at onset, autosomal recessive	AR	Unknown	609868
SPG11	Autosomal recessive spastic paraplegia type 11	AR	Unknown	610844
SPG7	Spastic paraplegia 7	AD/AR	Unknown	602783
SPR	Dopa-responsive dystonia due to sepiapterin reductase deficiency	AR	<1 / 1 000 000	182125
SRCAP	Floating-Harbor syndrome	AR	Unknown	611421
SRD5A2	Pseudovaginal perineoscrotal hypospadias	AR	Unknown	607306
SRD5A3	Congenital disorder of glycosylation, type Iq	AR	<1 / 1 000 000	611715
ST3GAL3	Intellectual developmental disorder, autosomal recessive 12	AR	Unknown	606494
ST3GAL5	GM3 synthase deficiency	AR	Unknown	604402
STAR	Lipoid adrenal hyperplasia	AR	Unknown	600617
STIL	Microcephaly 7, primary	AR	Unknown	181590
STIM1	Combined immunodeficiency due to STIM1 deficiency	AR	<1 / 1 000 000	605921
STK4	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations	AR	<1 / 1 000 000	604965
STRA6	Microphthalmia, syndromic 9	AR	Unknown	610745
STRC	Deafness, autosomal recessive 16	AR	1/1,000,000	606440
STX11	Hemophagocytic lymphohistiocytosis, familial, 4	AR	Unknown	605014
STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease	AR	Unknown	601717
SUCLA2	Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria	AR	<1 / 1 000 000	603921
SUCLG1	Mitochondrial DNA depletion syndrome 9	AR	Unknown	611224
SUMF1	Multiple sulfatase deficiency	AR	1-9 / 1 000 000	607939
SUOX	Sulfite oxidase deficiency	AR	<1 / 1 000 000	606887
SURF1	SURF1-related Charcot-Marie-Tooth disease type 4	AR	<1 / 1 000 000	185620
SYN1	Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders	XLR	Unknown	313440
SYP	Intellectual developmental disorder, X-linked 96	XLR	Unknown	313475
TAT	Tyrosinemia type 2	AR	<1 / 1 000 000	613018

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TAZ	Barth syndrome	XL	1-9 / 1 000 000	300394
TBCE	Kenny-Caffey syndrome/Sanjad-Sakati syndrome/ Early-onset progressive encephalopathy-spastic ataxia-distal spinal muscular atrophy syndrome	AR	<1 / 1 000 000	604934
TCAP	Cardiomyopathy, hypertrophic, 25/ Muscular dystrophy, limb-girdle, autosomal recessive 7	AD/AR	1-5 / 10 000	604488
TCIRG1	Osteopetrosis	AR	<1 / 1 000 000	604592
TCN2	Transcobalamin deficiency	AR	<1 / 1 000 000	613441
TF	Congenital atransferrinemia	AR	<1 / 1 000 000	190000
TFR2	Digenic hemochromatosis	AR	<1 / 1 000 000	604720
TG	Familial thyroid dyshormonogenesis	AR	1-9 / 100 000	188450
TGM1	Lamellar ichthyosis	AR	1-9 / 1 000 000	190195
TH	Autosomal recessive dopa-responsive dystonia	AR	1-9 / 1 000 000	191290
THRB	Resistance to thyroid hormone due to a mutation in thyroid hormone receptor beta	AR	Unknown	190160
TIMM8A	Mohr-Tranebjaerg syndrome	XLR	<1 / 1 000 000	300356
TK2	Autosomal recessive progressive external ophthalmoplegia/Mitochondrial DNA depletion syndrome, myopathic form	AR	Unknown	188250
TMC1	Deafness, autosomal dominant 36, autosomal recessive 7	AD; AR	<1/1,000,000	606706
TMEM216	Joubert syndrome 2/Meckel syndrome 2	AR	<1 / 1 000 000	613277
TMEM67	Joubert syndrome 6/Meckel syndrome 3	AR	<1 / 1 000 000	609884
TMIE	Deafness, autosomal recessive 6	AR	Unknown	607237
TNFRSF11B	Juvenile Paget disease	AR	<1 / 1 000 000	602643
TNFRSF4	Combined immunodeficiency due to OX40 deficiency	AR	<1 / 1 000 000	600315
TNNI3	Familial isolated dilated cardiomyopathy/Familial isolated restrictive cardiomyopathy	AD/AR	1-5 / 10 000	191044
TPM1	Cardiomyopathy, hypertrophic, 3/Cardiomyopathy, dilated, 1Y/Left ventricular noncompaction 9	AD/AR	1-5 / 10 000	191010
TPM3	Congenital myopathy 4A, autosomal dominant/Congenital myopathy 4B, autosomal recessive	AR/AD	Unknown	191030
TPO	Thyroid dyshormonogenesis 2A	AR	1-9 / 100 000	606765
TPP1	Ceroid lipofuscinosis, neuronal, 2	AR	Unknown	607998
TRAPPC9	Intellectual developmental disorder, autosomal recessive 13	AR	<1 / 1 000 000	611966
TRDN	Catecholaminergic polymorphic ventricular tachycardia	AR/AD	1-5 / 10 000	603283
TREX1	Aicardi-Goutières syndrome	AR/AD	Unknown	606609
TRHR	Resistance to thyrotropin-releasing hormone syndrome	AR	<1 / 1 000 000	188545
TRIM32	Bardet-Biedl syndrome 11/ Muscular dystrophy, limb-girdle, autosomal recessive 8	AR	Unknown	602290
TRIM37	Mulibrey nanism	AR	Unknown	605073
TRMU	Acute infantile liver failure, TRMU-related	AR	<1/1,000,000	610230
TSEN54	Pontocerebellar hypoplasia	AR	Unknown	608755
TSFM	Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3	AR	<1 / 1 000 000	604723
TSHB	Isolated thyroid-stimulating hormone deficiency	AR	Unknown	188540
TSHR	Hyperthyroidism, familial gestational/Hyperthyroidism, nonautoimmune/Hypothyroidism, congenital, nongoitrous, 1	AD/AR	Unknown	603372
TSPAN7	Intellectual developmental disorder, X-linked 58	XLR	Unknown	300096
TSPYL1	Sudden infant death with dysgenesis of the testes syndrome	AR	<1 / 1 000 000	604714
TTC21B	Short-rib thoracic dysplasia 4 with or without polydactyly	AR	Unknown	612014
TTC37	Tricho-hepato-enteric syndrome	AR	1/1,000,000	609193
TTC8	Bardet-Biedl syndrome 8	AR	Unknown	608132
TTPA	Ataxia with isolated vitamin E deficiency	AR	1-9 / 1 000 000	600415
TUFM	Combined oxidative phosphorylation defect type 4	AR	<1 / 1 000 000	602389
TULP1	Leber congenital amaurosis 15/Retinitis pigmentosa 14	AR	1-9 / 100 000	602280
TUSC3	Intellectual developmental disorder, autosomal recessive 7	AR	Unknown	601385
TXNRD2	Glucocorticoid deficiency 5	AR	Unknown	606448
TYK2	Immunodeficiency 35	AR	Unknown	176941
TYMP	Mitochondrial DNA depletion syndrome 1	AR	1-9 / 1 000 000	131222
TYR	Albinism, oculocutaneous, type IA/IB	AR	1-9 / 100 000	606933
TYRP1	Oculocutaneous albinism, type 3	AR	<1/1,000,000	115501
UBA1	VEXAS syndrome	XLR	<1 / 1 000 000	314370
UBE2A	Intellectual developmental disorder, X-linked syndromic, Nascimento type	XLR	<1 / 1 000 000	312180
UBR1	Johanson-Blizzard syndrome	AR	Unknown	605981
UGT1A1	Crigler-Najjar syndrome	AR	1/1,000,000	191740
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3	AR	Unknown	608897
UPB1	Beta-ureidopropionase deficiency	AR	1/1,000,000	613161
UPF3B	Intellectual developmental disorder, X-linked syndromic 14	XLR	Unknown	300298
UQCRB	Isolated complex III deficiency	AR	Unknown	191330
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4	AR	Unknown	612080

PrenatalAutism - Elenco dei geni e delle malattie genetiche a trasmissione ereditaria investigati

UROS	Porphyria, congenital erythropoietic	AR	<1 / 1 000 000	606938
USH1C	Usher syndrome, type 1C	AR	1-9 / 100 000	605242
USH1G	Sindrome di Usher, tipo 1	AR	1-9 / 100 000	607696
USH2A	Retinite pigmentosa/ Sindrome di Usher, tipo 2	AR	1-9 / 100 000	608400
USP9X	X-linked female restricted facial dysmorphism-short stature-choanal atresia-intellectual disability	XLD, XLR	<1 / 1 000 000	300072
VDR	Hypocalcemic vitamin D-resistant rickets	AR	Unknown	601769
VIPAS39	VPS33B interacting protein, apical-basolateral polarity regulator, spe-39 homolog - VIPAS39	AR	<1 / 1 000 000	613401
VLDLR	Atassia cerebellare non progressiva - deficit cognitivo	AR	<1 / 1 000 000	192977
VPS13A	Choreo-acanthocytosis	AR	1/1,000,000	605978
VPS13B	Cohen syndrome	AR	Unknown	607817
VPS33B	Arthrogryposis-renal dysfunction-cholestasis syndrome	AR	<1 / 1 000 000	608552
VRK1	Pontocerebellar hypoplasia, type 1A	AR	1/1,000,000	602168
VSX2	Microphthalmia/Anophthalmia, VSX2-related	AR	1/1,000,000	142993
VWF	Von Willebrand disease	AD; AR	1/1000	613160
WAS	X-linked severe congenital neutropenia/Wiskott-Aldrich syndrome/ X-linked thrombocytopenia with normal platelets	XLR	Unknown	300392
WDR19	Cranioectodermal dysplasia/ Jeune syndrome/Senior-Loken syndrome	AR	1-9 / 1 000 000	608151
WDR35	Cranioectodermal dysplasia 2/Short rib-polydactyly syndrome type 5/ Short rib-polydactyly syndrome, Verma-Naumoff type	AR	Unknown	613602
WDR62	Isolated congenital microcephaly	AR	Unknown	613583
WISP3	Progressive pseudorheumatoid dysplasia	AR	<1/1,000,000	603400
WNT10A	Schopf-Schulz-Passarge syndrome/Ectodermal dysplasia 16 (odontoonychodermal dysplasia)/Tooth agenesis, selective, 4	AR	<1 / 1 000 000	606268
WNT3	Tetraamelia-multiple malformations syndrome	AR	<1 / 1 000 000	165330
WNT7A	Phocomelia, Schinzel type/Fuhrmann syndrome	AR	Unknown	601570
WRN	Werner Syndrome	AR	1-9 / 1 000 000	277700
XDH	Xanthinuria type I	AR	Unknown	607633
XIAP	X-linked lymphoproliferative disease due to XIAP deficiency	XLR	<1 / 1 000 000	300079
XPA	Xeroderma pigmentosum, group A	AR	Unknown	611153
XPC	Xeroderma pigmentosum, group C	AR	Unknown	613208
ZAP70	Combined immunodeficiency due to ZAP70 deficiency	AR	Unknown	176947
ZDHHC9	Lujan-Fryns syndrome	XLR	Unknown	300646
ZFYVE26	Spastic paraplegia type 15	AR	1/500,000	612012
ZIC3	Congenitally uncorrected transposition of the great arteries/Situs ambiguus	XLR	Unknown	300265
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy/Restrictive dermopathy 1	XLR	Unknown	606480
ZNF469	Brittle cornea syndrome 1	AR	<1 / 1 000 000	612078
ZNF711	Intellectual developmental disorder, X-linked 97	XLR	Unknown	314990

PRENATALAUTISM

Elenco dei geni e delle malattie genetiche ad insorgenza de novo investigate

PrenatalAutism - elenco dei geni e delle malattie genetiche ad insorgenza de novo investigate

Gene Symbol	Disease(s)	Disease category	Model_Of_Inheritance
ACO2	Infantile cerebellar-retinal degeneration, OMIM:614559;Infantile cerebellar-retinal degeneration, MONDO:0013802	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ACTB	Dystonia, juvenile-onset, 607371Baraitser-Winter syndrome 1, 243310;BARAITSER-WINTER SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ACTG1	Deafness, autosomal dominant 20/26, 604717Baraitser-Winter syndrome 2, 614583;BARAITSER-WINTER SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ADAR	Aicardi-Goutieres syndrome 6, OMIM:615010;Dyschromatosis symmetrica hereditaria, OMIM:127400	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ADD1	Global developmental delay;Intellectual disability;Seizures;Ventriculomegaly;Abnormality of the corpus callosum	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ADNP	MENTAL RETARDATION, AUTOSOMAL DOMINANT, 28; MRD28	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
AFF3	KINSSHIP syndrome, OMIM:619297	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AGO1	Neurodevelopmental disorder with language delay and behavioral abnormalities, with or without seizures, OMIM:620292	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AGO2	Intellectual disability;regression;seizures	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, OMIM:615937	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ALDH18A1	Cutis laxa, autosomal recessive, type IIIA, 219150;SPASTIC PARAPLEGIA 9, AUTOSOMAL DOMINANT; SPG9	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ANK2	Complex neurodevelopmental disorder, MONDO:0100038	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ANK3	Intellectual developmental disorder, autosomal recessive 37, OMIM:615493;intellectual disability-hypotonia-spasticity-sleep disorder syndrome, MONDO:0014210	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ANKRD11	KBG syndrome, 148050;KBG SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ARID1A	Mental retardation, autosomal dominant 14, 614607;COFFIN-SIRIS SYNDROME; CSS	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ARID1B	Mental retardation, autosomal dominant 12, 614562;COFFIN SIRIS SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ASXL1	Bohring-Opitz syndrome, 605039Myelodysplastic syndrome, somatic, 614286;BOHRING-OPITZ SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ATN1	Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, OMIM:618494	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ATP1A2	Migraine, familial hemiplegic, 2 602481;Alternating hemiplegia of childhood 1, 104290	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ATP1A3	Alternating Hemiplegia of Childhood (AHC), intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ATP6V0A1	ATP6V0A1-related developmental disorder (monoallelic)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
AUTS2	SYNDROMIC INTELLECTUAL DISABILITY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
BAP1	Kury-Isidor syndrome, OMIM:619762	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BCL11A	INTELLECTUAL DISABILITY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
BMP4	Microphthalmia, syndromic 6 607932;Global developmental delay	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BRAF	Melanoma, malignant, somaticColorectal cancer, somaticAdenocarcinoma of lung, somatic, 211980Nonsmall cell lung cancer, somaticCardiofaciocutaneous syndrome, 115150Noonan syndrome 7, 613706LEOPARD syndrome 3, 613707;NOONAN SYNDROME TYPE 7 (NS7)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
BRSK2	Global developmental delay;Autism;Behavioral abnormality;Global developmental delay, Intellectual disability, Autism, Behavioral abnormality;Intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CACNA1A	Developemental and epileptic encephalopathy 42, OMIM:617106;developmental and epileptic encephalopathy, 42, MONDO:0014917	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CACNA1C	Timothy syndrome, OMIM:601005;Timothy syndrome, MONDO:0010979;Long QT syndrome 8, OMIM:618447;long qt syndrome 8, MONDO:0032756;Brugada syndrome 3, OMIM:611875;Brugada syndrome 3, MONDO:0012742;CACNA1C-related disorder	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CACNA1D	Primary aldosteronism, seizures, and neurologic abnormalities 615474 AD; Sinoatrial node dysfunction and deafness 614896 AR	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CACNA1E	Global developmental delay;Intellectual disability;Seizures;Dystonia;Congenital contracture;Macrocephaly;Epileptic encephalopathy, early infantile, 69, 618285	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CACNA1G	Spinocerebellar ataxia 42 616795;Cerebellar atrophy, epilepsy, intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CAMK4	Global developmental delay;Intellectual disability;Autism;Behavioral abnormality;Abnormality of movement;Dystonia;Ataxia;Chorea;Myoclonus	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CAMTA1	CEREBELLAR ATAXIA, NONPROGRESSIVE, WITH MENTAL RETARDATION	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;NOONAN SYNDROME-LIKE DISORDER WITH OR WITHOUT JUVENILE MEYLOMONOCYTIC LEUKEMIA	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CDON	Holoprosencephaly 11, 614226;HOLOPROSENCEPHALY 11	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CEP85L	Lissencephaly 10, OMIM:618873;Lissencephaly 10, MONDO:0030031	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CHD2	Epileptic encephalopathy, childhood-onset, 615369;EPILEPTIC ENCEPHALOPATHY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CHD3	Global developmental delay;Intellectual disability;Macrocephaly;Snijders Blok-Campeau syndrome, 618205	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHD7	CHARGE syndrome, 214800{Scoliosis, idiopathic 3}, 608765Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;KALLMANN SYNDROME TYPE 5 (KAL5)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CHD8	Overgrowth with Intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CIC	Mental retardation, autosomal dominant 45 617600	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CNNM2	Hypomagnesemia, seizures, and mental retardation, OMIM:616418;Hypomagnesemia, seizures, and mental retardation, MONDO:0014631	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CNOT3	CNOT3 syndrome;intellectual disability, global developmental delay;Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL4A1	Porencephaly 1, 175780Brain small vessel disease with hemorrhage, 607595Angiopathy, hereditary, with nephropathy, aneurysms, and muscle, 611773Brain small vessel disease with Axenfeld-Rieger anomaly, 607595{Hemorrhage, intracerebral, susceptibility to}, 614519;PORENCEPHALY 1	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
COL4A2	Porencephaly 2, 614483{Hemorrhage, intracerebral, susceptibility to}, 614519;PORENCEPHALY 2	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CREBBP	Rubinstein-Taybi syndrome, 180849;RUBINSTEIN-TAYBI SYNDROME TYPE 1 (RSTS1)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CSDE1	Autism;Global developmental delay;Intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CTNNB1	Neurodevelopmental disorder with spastic diplegia and visual defects, OMIM:615075	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CTNND1	developmental delay	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CUL3	Neurodevelopmental disorder with or without autism or seizures, OMIM:619239	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DEAF1	?Dyskinesia, seizures, and intellectual developmental disorder, 617171;Mental retardation, autosomal dominant 24, 615828	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
DHDDS	Developmental delay and seizures with or without movement abnormalities, OMIM:617836	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
DLL1	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, OMIM:618709	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DNM1	Developmental and epileptic encephalopathy 31, OMIM:616346	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
DNM1L	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, MIM#614388	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
DNMT3A	Tatton-Brown-Rahman syndrome OMIM:615879;Heyn-Sproul-Jackson syndrome OMIM:618724;MONDO:0032882	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
DOCK4	neuronevelopmental disorder, MONDO:0700092;intellectual disability, MONDO:0001071	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
DYNC1H1	Mental Retardation, Dominant;Charcot-Marie-Tooth disease, axonal, type 20, 614228;Mental retardation, autosomal dominant 13, 614563;Spinal muscular atrophy, lower extremity-predominant, AD, 158600;SPINAL MUSCULAR ATROPHY, LOWER EXTREMITY-PREDOMINANT, AD	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
DYRK1A	Mental retardation, autosomal dominant 7, 614104;MENTAL RETARDATION AUTOSOMAL DOMINANT TYPE 7	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type, 610536;MANDIBULOFACIAL DYSOSTOSIS WITH MICROCEPHALY; MFDM	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
EHMT1	Kleefstra syndrome, 610253;9Q SUBTELOMERIC DELETION SYNDROME (9Q- SYNDROME)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
EMX2	Schizencephaly 269160	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EP300	RUBINSTEIN-TAYBI SYNDROME TYPE 2 (RSTS2)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ERBB4	intellectual disability MONDO:0001071	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
EZH2	WEAVER SYNDROME 2	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FBXW11	Neurodevelopmental, jaw, eye, and digital syndrome, OMIM:618914;neurodevelopmental, jaw, eye, and digital syndrome, MONDO:003005	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FBXW7	Developmental delay, hypotonia, and impaired language, OMIM:620012	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXG1	Rett Syndrome, congenital variant OMIM:613454;Rett syndrome, congenital variant MONDO:0013270	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FOXP1	Mental retardation with language impairment and autistic features, 613670;Mental Retardation with Language Impairment and Autistic Features;MENTAL RETARDATION WITH LANGUAGE IMPAIRMENT AND AUTISTIC FEATURES (MRLIAF)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FOXP2	Speech-Language Disorder 1;SPEECH-LANGUAGE DISORDER 1	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
GABRA1	JUVENILE MYOCLONIC EPILEPSY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
GABRA5	Epileptic encephalopathy, early infantile, 79, 618559;developmental delay	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GABRB3	CHILDHOOD ABSENCE EPILEPSY TYPE 5	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
GABRD	{Epilepsy, idiopathic generalized, 10}, OMIM:613060;{Epilepsy, juvenile myoclonic, susceptibility to}, OMIM:613060	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
GABRG2	Epilepsy, generalized, with febrile seizures plus, type 3 611277;Febrile seizures, familial, 8 611277;{Epilepsy, childhood absence, susceptibility to, 2} 607681	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230Hyperphenylalaninemia, BH4-deficient, B, 233910;DYSTONIA TYPE 5 (DYT5)	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GFAP	Alexander disease, 203450;ALEXANDER DISEASE	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
GLI2	Holoprosencephaly-9, 610829;GLI2-RELATED HOLOPROSENCEPHALY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted

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GNAS	Pseudohypoparathyroidism Ia, OMIM:103580;pseudohypoparathyroidism type 1A, MONDO:0007078;Pseudohypoparathyroidism Ic, OMIM:612462;pseudohypoparathyroidism type 1C, MONDO:0012911;Pseudopseudohypoparathyroidism, OMIM:612463;pseudopseudohypoparathyroidism, MONDO:0012912	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GRIK2	Mental retardation, autosomal recessive, 6, OMIM:611092;non-syndromic neurodevelopmental disorder (NDD), autosomal dominant	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant OMIM:614254;intellectual disability, autosomal dominant 8 MONDO:0013655;Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive OMIM:617820;neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive MONDO:0060629	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GRIN2A	Epilepsy with neurodevelopmental defects, 613971;EPILEPSY WITH NEURODEVELOPMENTAL DEFECTS	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
GRIN2B	Intellectual developmental disorder, autosomal dominant 6, with or without seizures, OMIM:613970;Developmental and epileptic encephalopathy 27, OMIM:616139	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
HCN1	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 24; EIEE24	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
HDAC4	Brachydactyly-mental retardation syndrome, 600430;BRACHYDACTYLY-MENTAL RETARDATION SYNDROME (BDMR)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A, OMIM:613925;Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, OMIM:613926	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
HESX1	Growth hormone deficiency with pituitary anomalies, OMIM:182230;Pituitary hormone deficiency, combined, 5, OMIM:182230;Septooptic dysplasia, OMIM:182230	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
HK1	Neurodevelopmental disorder with visual defects and brain anomalies, OMIM:618547	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
HNRNPU	Epileptic encephalopathy, early infantile, 54, 617391;intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
HPD	Tyrosinemia, type III, 276710Hawkinsinuria, 140350;HAWKINSINURIA (HAWK)	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
HRAS	{Bladder cancer, somatic}, 109800Costello syndrome, 218040{Thyroid carcinoma, follicular, somatic}, 188470Congenital myopathy with excess of muscle spindles, 218040{Nevus sebaceous, somatic}, 162900Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;COSTELLO SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
IDH2	D-2-hydroxyglutaric aciduria 2613657	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
IFIH1	Aicardi-Goutieres syndrome 7, OMIM:615846	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
IMPDH2	Neurodevelopmental disorder with dystonia	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ITPR1	Gillespie syndrome, OMIM:206700;Spinocerebellar ataxia 15, OMIM:606658;Spinocerebellar ataxia 29, congenital nonprogressive, OMIM:117360	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KANSL1	Koolen-De Vries syndrome, 610443;Intellectual Disability Syndrome;CHROMOSOME 17Q21.31 MICRODELETION SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KAT6B	SBBYSS syndrome, 603736Genitopatellar syndrome, 606170;BLEPHAROPHIMOSIS/INTELLECTUAL DISABILITY PHENOTYPE WHICH IS NOONAN-LIKE	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KCNA3	Neurodevelopmental disorder, MONDO:0700092;intellectual disability, MONDO:0001071	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KCND2	global developmental delay, HP:0001263	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNJ11	Hyperinsulinemic hypoglycemia, familial, 2, 601820Diabetes, permanent neonatal, 606176Diabetes mellitus, permanent neonatal, with neurologic features, 606176{Diabetes mellitus, type 2, susceptibility to}, 125853Diabetes mellitus, transient neonatal, 3, 610582;FAMILIAL HYPERINSULINISM	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KCNJ6	Keppen-Lubinsky syndrome 614098	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNK9	Birk-Barel mental retardation dysmorphism syndrome 612292	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)
KCNMA1	Cerebellar atrophy, developmental delay, and seizures, OMIM:617643;Cerebellar atrophy, developmental delay, and seizures, MONDO:0060551;Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, OMIM:609446;Generalized epilepsy-paroxysmal dyskinesia syndrome, MONDO:0012276;Liang-Wang syndrome, OMIM:618729;Liang-Wang syndrome, MONDO:0032886;{Epilepsy, idiopathic generalized, susceptibility to, 16}, OMIM:618596;Epilepsy, idiopathic generalized, susceptibility to, 16, MONDO:0032827	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KCNN3	Zimmermann-Laband syndrome 3, OMIM:618658	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KCNQ2	Developmental and epileptic encephalopathy 7, OMIM:613720	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KCNQ3	Seizures, benign neonatal, 2; MIM: 121201	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KDM5A	autism spectrum disorder, MONDO:0005258;intellectual disability, MONDO:0001071;El Hayek-Chahrour neurodevelopmental syndrome, OMIM:620820	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KDM6B	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, OMIM:618505;neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, MONDO:0032790	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950;AUTOSOMAL-DOMINANT MICROCEPHALY ASSOCIATED WITH LYMPHEDEMA AND/OR CHORIORETINOPATHY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KIF1A	NESCAV syndrome, OMIM:614255;Spastic paraplegia 30, autosomal dominant, OMIM:610357	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KIF5A	Myoclonus, intractable, neonatal, 617235;intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KIRREL3	Intellectual developmental disorder, autosomal dominant 4, OMIM:612581;intellectual disability, autosomal dominant 4, MONDO:0012947	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KLF7	Intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KMT2C	Kleefstra syndrome 2, 617768	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KMT2D	Kabuki syndrome 1, OMIM:147920	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KMT2E	O'Donnell-Luria-Rodan syndrome, 618512;Global developmental delay;Intellectual disability;Autism;Seizures;Abnormality of skull size	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KRAS	Lung cancer, somatic, 211980Bladder cancer, somatic, 109800Pancreatic carcinoma, somatic, 260350Gastric cancer, somatic, 137215Leukemia, acute myelogenousNoonan syndrome 3, 609942Cardiofaciocutaneous syndrome 2, 615278Breast cancer, somatic, 114480SFM syndrome, somatic mosaic, 163200;CARDIOFACIOCUTANEOUS SYNDROME (CFC SYNDROME)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
LMNB1	Microcephaly 26, primary, autosomal dominant, OMIM:619179	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MAF	CATARACT, DEAFNESS, INTELLECTUAL DISABILITY, SEIZURES, AND A DOWN SYNDROME-LIKE FACIES	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MAGEL2	PRADER WILLI SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, maternally imprinted (paternal allele expressed)
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279;CARDIOFACIOCUTANEOUS SYNDROME (CFC SYNDROME)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MAP2K2	Cardiofaciocutaneous syndrome 4, 615280;CARDIOFACIOCUTANEOUS SYNDROME (CFC SYNDROME)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MAST4	neurodevelopmental disorder MONDO:0700092, MAST4-related	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MAT1A	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/IIIdeficiency, 250850Methionine adenosyltransferase deficiency, autosomal recessive, 250850;METHIONINE ADENOSYLTRANSFERASE DEFICIENCY	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MBD5	Mental Retardation, Dominant;Autosomal Dominant Mental Retardation syndrome type 1;Mental retardation, autosomal dominant 1, 156200;EHMT1-LIKE INTELLECTUAL DISABILITY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MED13	Delayed speech and language development;Motor delay;Intellectual disability;Autistic behavior;Attention deficit hyperactivity disorder;Abnormality of the eye;Constipation	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MED13L	Impaired intellectual development and distinctive facial features with or without cardiac defects, OMIM:616789	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MEF2C	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443;MRSME;Chromosome 5q14.3 deletion syndrome, 613443	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MEIS2	Cleft palate, cardiac defects, and mental retardation, OMIM:600987;Cardiac malformation, cleft lip/palate, microcephaly, and digital anomalies, MONDO:0010970	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MYCN	Feingold syndrome 1, OMIM:164280	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MYT1L	Mental retardation, autosomal dominant 39, 616521;MRD39;Intellectual disability;obesity	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NBEA	Global developmental delay;Intellectual disability;Seizures;No OMIM number	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NCKAP1	Intellectual disability;Autism	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NEDD4L	Periventricular nodular heterotopia 7, 617201	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NF1	Neurofibromatosis, type 1, 162200Leukemia, juvenile myelomonocytic, 607785Melanoma, desmoplastic neurotrophic (2)Neurofibromatosis, familial spinal, 162210Neurofibromatosis-Noonan syndrome, 601321Watson syndrome, 193520;NEUROFIBROMATOSIS-NOONAN SYNDROME (NFNS)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NFIA	Brain malformations with or without urinary tract defects, 613735;BRMUTD;Intellectual disability;Chromosome 1p32-p31 deletion syndrome, included	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NFIX	SOTOS-LIKE SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NIPBL	Cornelia de Lange syndrome 1, 122470;CORNELIA DE LANGE SYNDROME TYPE 1 (CDLS1)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NKX2-1	Goiter, familial, due to TTF-1 defect (1)Chorea, hereditary benign, 118700Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978;BENIGN HEREDITARY CHOREA	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NR2F1	BOSCH-BOONSTRA OPTIC ATROPHY SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NR4A2	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, OMIM:619911	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NRAS	Autoimmune lymphoproliferative syndrome type IV, 614470Noonan syndrome 6, 613224Epidermal nevus, somatic, 162900Thyroid carcinoma, follicular, somatic, 188470Colorectal cancer, somatic, 114500;NOONAN SYNDROME TYPE 6	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NRXN1	Pitt-Hopkins-like syndrome 2, OMIM:614325 (AR);Complex neurodevelopmental disorder (AD)	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
NSD1	Sotos syndrome 1, 117550Leukemia, acute myeloid, 601626 (1)Beckwith-Wiedemann syndrome, 130650;WEAVER SYNDROME (WES)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NTRK2	Epileptic encephalopathy, early infantile, 58, 617830;Obesity, hyperphagia, and developmental delay, 613886	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

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ODC1	Global developmental delay;Intellectual disability;Macrocephaly;Alopecia;Ectodermal dysplasia	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
OTX2	MICROPHTHALMIA SYNDROMIC TYPE 5 (MCOPS5)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PAFAH1B1	Lissencephaly 1, OMIM:607432;Subcortical laminar heterotopia, OMIM:607432	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700;CONGENITAL HYPOTHYROIDISM NON-GOITROUS TYPE 2 (CHNG2)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PDE4D	Acrodysostosis 2, with or without hormone resistance, 614613;Acrodysostosis Orphanet:950	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PEX6	Peroxisome biogenesis disorder 4A (Zellweger), OMIM:614862;Peroxisome biogenesis disorder 4B, OMIM:614863	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PHIP	INTELLECTUAL DISABILITY;Developmental delay, intellectual disability, obesity, and dysmorphic features, 617991	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PIK3CA	Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501;Megalencephaly-capillary malformation-polymicrogyria syndrome, Orphanet:60040	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, OMIM:603387	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PIP5K1C	neurodevelopmental disorder, MONDO:0700092;intellectual disability, MONDO:0001071	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
POGZ	White-Sutton syndrome, OMIM:616364	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, OMIM:614381;POLR3B-related neurodevelopmental disorder;Ataxia, spasticity, and demyelinating neuropathy	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
POLRMT	Combined oxidative phosphorylation deficiency 55, OMIM:619743	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PPM1D	Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold, 617450;IDDGIP	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PPP2R1A	INTELLECTUAL DISABILITY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PRICKLE2	Neurodevelopmental disorder;global developmental delay	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PRPF8	PRPF8-related developmental disorder (monoallelic);Retinitis pigmentosa 13, OMIM:600059	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PTCH1	Holoprosencephaly 7, OMIM:610828	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PTEN	Cowden syndrome 1, 158350Lhermitte-Duclos syndrome, 158350Bannayan-Riley-Ruvalcaba syndrome, 153480{Meningioma}, 607174{Glioma susceptibility 2}, 613028Macrocephaly/autism syndrome, 605309PTEN hamartoma tumor syndromeVATER association with macrocephaly and ventriculomegaly, 276950{Prostate cancer, somatic}, 176807Thyroid carcinoma, follicular, somatic, 188470Malignant melanoma, somatic, 155600Endometrial carcinoma, somatic, 608089Squamous cell carcinoma, head and neck, somatic, 275355;PROTEUS SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PTPN11	Noonan syndrome 1, 163950LEOPARD syndrome 1, 151100Leukemia, juvenile myelomonocytic, 607785Metachondromatosis, 156250;LEOPARD SYNDROME TYPE 1	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
RAC1	intellectual disability; developmental delay	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RAD21	Cornelia de Lange syndrome 4, OMIM:614701	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
RAF1	Noonan syndrome 5, 611553LEOPARD syndrome 2, 611554;NOONAN SYNDROME 5	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
RAI1	Immunodeficiency 9, 612782;SMITH-MAGENIS SYNDROME (SMS)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
RELN	Lissencephaly 2 (Norman-Roberts type), OMIM:257320;neurodevelopmental disorder, MONDO:0700092	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
RORA	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SAMD9	MIRAGE syndrome, 617053	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SATB2	Cleft palate and mental retardation, 119540;CLEFT PALATE ISOLATED (CPI)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SCN1A	Epilepsy, generalized, with febrile seizures plus, type 2, 604403Dravet syndrome, 607208Migraine, familial hemiplegic, 3, 609634Febrile seizures, familial, 3A, 604403;SCN1A-RELATED SEIZURE DISORDERS	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SCN2A	Seizures, benign familial infantile, 3, 607745Epileptic encephalopathy, early infantile, 11, 613721;NONSPECIFIC SEVERE ID	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SCN3A	Epileptic encephalopathy, early infantile, 62, 617938;intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SCN8A	Cognitive impairment with or without cerebellar ataxia, OMIM:614306;Developmental and epileptic encephalopathy 13, OMIM:614558	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SETBP1	Schinzel-Giedion midface retraction syndrome, 269150;SCHINZEL-GIEDION MIDFACE RETRACTION SYNDROME (SGMFS)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SETD2	Luscan-Lumish syndrome, 616831;intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SHANK2	SUSCEPTIBILITY TO AUTISM TYPE 17 (AUTS17)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SHANK3	Phelan-McDermid syndrome, 606232{Schizophrenia 15}, 613950;PHELAN-MCDERMID SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SHH	Holoprosencephaly-3, 142945Single median maxillary central incisor, 147250Microphthalmia with coloboma 5, 611638Schizencephaly, 269160;TRIPHALANGEAL THUMB-POLYSYNDACTYLY SYNDROME (TPTPS)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SHOC2	Noonan-like syndrome with loose anagen hair, 607721;NOONAN-LIKE SYNDROME WITH LOOSE ANAGEN HAIR	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SIX3	Holoprosencephaly-2, 157170;Schizensephaly, 269160;HOLOPROSENCEPHALY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SKI	Shprintzen-Goldberg syndrome, 182212;SHPRINTZEN-GOLDBERG CRANIOSYNOSTOSIS SYNDROME; SGS	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SLC1A2	Epileptic encephalopathy, early infantile, 41, OMIM:617105;developmental and epileptic encephalopathy, 41, MONDO:0014916	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SLC2A1	GLUT1 deficiency syndrome 1, 606777;GLUT1 deficiency syndrome 2, 612126;{Epilepsy, idiopathic generalized, suscpetibility to, 12}, 614847;Dystonia 9, 601042;GLUT1 DEFICIENCY SYNDROME TYPE 1 (GLUT1DS1)	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SLC6A1	Myoclonic-atonic epilepsy, OMIM:616421	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SMAD4	Myhre syndrome, 139210;Includes intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMARCA2	Nicolaides-Baraitser syndrome, OMIM:601358;Coffin-siris syndrome;Blepharophimosis intellectual disability syndrome	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SMARCA4	Coffin-Siris syndrome 4, OMIM:614609	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SMARCB1	Coffin-Siris syndrome 3, OMIM:614608	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SMARCE1	Coffin-Siris syndrome 5, OMIM:616938	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SMC3	Cornelia de Lange syndrome 3, 610759;CDLS3	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SOS1	Fibromatosis, gingival, 135300Noonan syndrome 4, 610733;NOONAN SYNDROME 4	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SOX10	Waardenburg syndrome, type 4C, 613266Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584PCWH syndrome, 609136;PERIPHERAL DEMYELINATING NEUROPATHY, CENTRAL DYSMYELINATING LEUKODYSTROPHY, WAARDENBURG SYNDROME, AND HIRSCHSPRUNG DISEASE (PCWH)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SOX2	AEG SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SOX5	12P12.5 INTRAGENIC DELETIONS ASSOCIATED WITH INTELLECTUAL DISABILITY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SOX6	intellectual diability;ADHD;Craniosynostosis;Osteochondromas	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SPECC1L	Opitz GBBB syndrome, type II,145410;Intellectual disability;Autosomal dominant Opitz G/BBB syndrome	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SPRED1	Legius syndrome, 611431;LEGIUS SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SPTAN1	Developmental and epileptic encephalopathy 5, OMIM:613477;developmental and epileptic encephalopathy, 5, MONDO:0013277	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SRCAP	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, OMIM:619595;Floating-Harbor syndrome, OMIM:136140	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
STX1A	neurodevelopmental disorder, MONDO:0700092;intellectual disability, MONDO:0001071	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
STXBP1	Developmental and epileptic encephalopathy 4, OMIM:612164;developmental and epileptic encephalopathy, 4, MONDO:0012812	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SUFU	Joubert syndrome 32, OMIM:617757	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SUPT16H	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, OMIM:619480;Global developmental delay;Intellectual disability;Abnormality of the corpus callosum	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SYNGAP1	Mental retardation, autosomal dominant 5, 612621;EPILEPTIC ENCEPHALOPATHY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TBL1XR1	AUTISM	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TCF4	Pitt-Hopkins syndrome, 610954;PITT-HOPKINS SYNDROME (PTHS)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TCF7L2	Developmental disorders;Global developmental delay;Intellectual disability;Autism;Attention deficit hyperactivity disorder;Myopia;Abnormality of skeletal system	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TGIF1	HOLOPROSENCEPHALY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
THRA	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 6	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750Chilblain lupus, 610448Vasculopathy, retinal, with cerebral leukodystrophy, 192315{Systemic lupus erythematosus, susceptibility to}, 152700;AICARDI-GOUTIERES SYNDROME 1, DOMINANT AND RECESSIVE	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
TRIO	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, OMIM:617061;Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, OMIM:618825	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TRIP12	Mental retardation, autosomal dominant 49617752	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TRPM3	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, OMIM:620224	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TRRAP	Microcephaly;Seizures;Abnormal heart morphology;Autism;Developmental delay with or without dysmorphic facies and autism, 603015;Intellectual disability;Abnormality of the urinary system;Global developmental delay	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TSC1	Tuberous sclerosis-1, 191100Lymphangioleiomyomatosis, 606690;Focal cortical dysplasia, Taylor balloon cell type, 607341;TUBEROUS SCLEROSIS TYPE 1 (TSC1)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TSC2	Tuberous sclerosis-2, 613254Lymphangioleiomyomatosis, somatic, 606690;LYMPHANGIOLEIOMYOMATOSIS (LAM)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted

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TUBA1A	Lissencephaly 3, 611603;INTELLECTUAL DISABILITY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TUBB2B	Cortical dysplasia, complex, with other brain malformations 7, OMIM:610031	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TUBB3	CONGENITAL FIBROSIS OF THE EXTRAOCULAR MUSCLES	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TWIST1	SAETHRE-CHOTZEN SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
UBE3A	Angelman syndrome, 105830;ANGELMAN SYNDROME	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)
USP7	Hao-Fountain syndrome, 616863	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
VCP	neurodevelopmental disorder, MONDO:0700092;intellectual disability, MONDO:0001071	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
WDFY3	Microcephaly 18, primary, autosomal dominant, 617520	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
YY1	Gabriele-de Vries syndrome617557	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ZBTB18	intellectual disability with variable features; global developmental delay; Mental retardation, autosomal dominant 22, 612337	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZEB2	Mowat-Wilson syndrome, 235730;MOWAT-WILSON SYNDROME (MWIS)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ZFHX3	syndromic intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZFHX4	Developmental disorders;intellectual disability, dysmorphic features	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZIC1	Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736;?Craniosynostosis 6, 616602	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZIC2	Holoprosencephaly-5, 609637;HOLOPROSENCEPHALY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ZMYND11	INTELLECTUAL DISABILITY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ACTA2	Aortic aneurysm, familial thoracic 6, 611788;Multisystemic; smooth muscle dysfunction syndrome, 613834;Moyamoya disease 5, 614042	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ACVR1	Fibrodysplasia ossificans progressiva 135100	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AKT1	Cowden syndrome 6 OMIM:164730	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ALX4	Parietal foramina;Parietal foramina 2, (AD), 609597;Frontonasal dysplasia 2, (AR), 613451	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ANO5	Gnathodiaphyseal dysplasia, 166260; Muscular dystrophy, limb-girdle, type 2L, 611307; Miyoshi muscular dystrophy 3, 613319	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CACNB4	Episodic ataxia, type 5 613855; {Epilepsy, idiopathic generalized, susceptibility to, 9} 607682; {Epilepsy, juvenile myoclonic, susceptibility to, 6} 607682	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CASR	Hypocalciuric hypercalcemia, type I, 145980	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CD96	C syndrome, 211750	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CDC42BPB	CDC42BPB-related Neurodevelopmental Disorder;Central hypotonia;Global developmental delay;Intellectual disability;Seizures;Autistic behavior;Behavioral abnormality	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CDKN1C	IMAGE syndrome, 614732;ntrauterine Growth Restriction, Metaphyseal Dysplasia, Adrenal Hypoplasia Congenita, and Genital Anomalies	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CEP63	?Seckel syndrome 6 614728; Developmental dyslexia	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CHL1	verbal function and developmental delay	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHRM1	Neurodevelopmental delay;intellectual disability, MONDO:0001071;autism	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
DOCK8	Intellectual developmental disorder, autosomal dominant 2, OMIM:614113	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DPYSL2	intellectual disability, MONDO:0001071;Aplasia/Hypoplasia of the corpus callosum, HP:0007370	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DROSHA	Global developmental delay;Intellectual disability;Seizures;Cerebral white matter atrophy;Abnormality of the corpus callosum;Abnormality of movement;Stereotypic behavior;Abnormality of head or neck;Short foot	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EPB41L1	?Mental retardation, autosomal dominant 11 614257	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EPHA7	Global developmental delay;Intellectual disability, MONDO:0001071;Delayed speech and language development;Behavioral abnormality	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGF14	Spinocerebellar ataxia 27 609307	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGFR2	Antley-Bixler syndrome;Apert syndrome;Crouzon syndrome;Beare-Stevenson Cutis Gyrata syndrome	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GABBR1	Neurodevelopmental disorder with language delay and variable cognitive abnormalities, OMIM:620502	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
GATA6	Atrioventricular septal defect 5, 614474; Atrial septal defect 9, 614475; Pancreatic agenesis and congenital heart defects, 600001; Persistent truncus arteriosus, 217095; Tetralogy of Fallot, 187500	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GJB3	Erythrokeratodermia variabilis et progressiva, 133200;Deafness, autosomal dominant 2B, 612644;Deafness, autosomal recessive;Deafness, autosomal dominant, with peripheral neuropathy;Deafness, digenic, GJB2/GJB3, 220290	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GLI3	Greig cephalopolysyndactyly syndrome, OMIM:175700;Pallister-Hall syndrome, OMIM:146510	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
GNAI2	Syndromic intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
GNE	Sialuria, MIM#269921	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
HNF1B	Renal cysts and diabetes syndrome, 137920; Diabetes mellitus, noninsulin-dependent, 125853; {Renal cell carcinoma}, 144700	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
JMJD1C	Intellectual disability;Autism	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KCNA1	Episodic ataxia/myokymia syndrome, OMIM:160120	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KCNC3	Spinocerebellar ataxia 13, OMIM:605259;MONDO:0011529	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KIF5B	kyphomelic dysplasia, MONDO:0008881;intellectual disability, MONDO:0001071	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
LMNB2	Microcephaly 27, primary, autosomal dominant, OMIM:619180	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MAPK10	Lennox-Gastaut syndrome;LGS;Epileptic Encephalopathy;Epileptic Encephalopathy Lennox-Gastaut type	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MIR17HG	Feingold syndrome 2, 614326;FS2;Brachydactyly with short stature and microcephaly;Intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
POU1F1	Pituitary hormone deficiency, combined or isolated, 1, OMIM:613038	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PTH1R	Metaphyseal chondrodysplasia, Murk Jansen type, 156400; Chondrodysplasia, Blomstrand type, 215045; Eiken syndrome, 600002; Failure of tooth eruption, primary, 125350	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PTHLH	Humoral hypercalcemia of malignancy;Brachydactyly, type E2, 613382	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RAD51	Fanconi anemia, complementation group R, OMIM:617244	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
RYR2	Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, MIM: 115000; Ventricular tachycardia, catecholaminergic polymorphic, 1, MIM: 604772	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SALL1	Townes-Brocks branchiootorenal-like syndrome, 107480; Townes-Brocks syndrome 1, 107480; TBS	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SLC4A1	Ovalocytosis; Spherocytosis, type 4, 612653; [Malaria, resistance to], 611162; Renal tubular acidosis, distal, AD, 179800; Renal tubular acidosis, distal, AR, 611590; [Blood group, Diego], 110500; [Blood group, Waldner], 112010; [Blood group, Wright], 112050; [Blood group, Froese], 601551; [Blood group, Swann], 601550	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SLC5A7	Myasthenic syndrome, congenital, 20, presynaptic,CMS20, 617143	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SMAD3	Loeys-Dietz syndrome, type 3, 613795	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SOX9	Campomelic dysplasia with autosomal sex reversal, OMIM:114290;Acampomelic campomelic dysplasia, OMIM:114290;Campomelic dysplasia, OMIM:114290;campomelic dysplasia, MONDO:0007251	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SRGAP3	3p- syndrome, MIM:613792 (includes intellectual disability)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TAB2	Congenital heart defects, nonsyndromic, 2, OMIM:614980	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX1	22q11.2 deletion syndrome, Orphanet:567 (includes developmental delay); DiGeorge syndrome, 188400 (includes mild to moderate learning difficulties); Velocardiofacial syndrome, 192430 (includes learning disability and mental retardation)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
THRB	compromised intellectual development	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
TWIST2	Barber-Say syndrome, 209885 (includes mental retardation in some patients); Focal facial dermal dysplasia 3, Setleis type, 227260 (can include intellectual disability and developmental delay)	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ACAN	Spondyloepiphyseal dysplasia, Kimberley type, 608361;Spondyloepimetaphyseal dysplasia, aggrecan type, 612813;Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ADCY5	Dyskinesia, familial, with facial myokymia, 606703	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AFG3L2	Optic atrophy 12, OMIM:618977;Spinocerebellar ataxia 28, OMIM:610246;Spastic ataxia 5, autosomal recessive, OMIM:614487	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ANKH	Craniometaphyseal dysplasia, 123000Chondrocalcinosis 2, 118600;CRANIOMETAPHYSEAL DYSPLASIA JACKSON TYPE (CMDJ)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ARHGAP31	Adams-Oliver syndrome 1 100300	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ATL1	Spastic paraplegia 3A, autosomal dominant, 182600;Hereditary spastic paraplegia;Intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ATP2A2	Darier disease 124200	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BEAN1	Spinocerebellar ataxia 31 117210	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BFSP2	Cataract 12, multiple types, 611597	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BMPR1B	Acromesomelic dysplasia, Demirhan type, OMIM:609441;Brachydactyly, type A1, D, OMIM:616849;Brachydactyly, type A2, OMIM:112600	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CACNA1H	Hyperaldosteronism, familial, type IV 617027; {Epilepsy, childhood absence, susceptibility to, 6} 611942; {Epilepsy, idiopathic generalized, susceptibility to, 6} 611942	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CACNA1S	Hypokalemic periodic paralysis, type 1, 170400; {Malignant; hyperthermia susceptibility 5}, 601887; {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

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CACNG2	?Mental retardation, autosomal dominant 10 614256	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CDH15	Mental retardation, autosomal dominant 3, OMIM:612580	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CHRNA2	Epilepsy, nocturnal frontal lobe, type 4 610353	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHRNA4	Epilepsy, nocturnal frontal lobe, 1 600513 AD; {Nicotine addiction, susceptibility to} 188890	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHRNB2	Epilepsy, nocturnal frontal lobe, 3 605375	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CLCN5	Dent disease	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CLCN7	Osteopetrosis, autosomal recessive 4, OMIM:611490;Osteopetrosis, autosomal dominant 2, OMIM:166600	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL10A1	Metaphyseal chondrodysplasia, Schmid type, 156500	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL11A1	Stickler syndrome, type II, 604841;Marshall syndrome, 154780;{Lumbar disc herniation, susceptibility to}, 603932;Fibrochondrogenesis, 228520	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL11A2	Gene2Phenotype confirmed gene with ID HPO	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL1A1	Osteogenesis imperfecta, type I, 166200; OI type II, 166210; OI type III, 259420; OI type IV, 166220; Ehlers-Danlos syndrome, type I, 130000; Ehlers-Danlos syndrome, type VIIA, 130060; {Osteoporosis}, 166710; Caffey disease, 114000; [Bone mineral density variation QTL], 166710	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL2A1	Stickler syndrome, type I, 108300; Kniest dysplasia, 156550; Achondrogenesis, type II or hypochondrogenesis, 200610; SED congenita, 183900; SMED Strudwick type, 184250; Epiphyseal dysplasia, multiple, with myopia and deafness, 132450; Spondyloperipheral dysplasia, 271700; SED, Namaqualand type; Osteoarthritis with mild chondrodysplasia, 604864; Vitreoretinopathy with phalangeal epiphyseal dysplasia; Platyspondylic skeletal dysplasia, Torrance type, 151210; Otospondylomegaepiphyseal dysplasia, 215150; Avascular necrosis of the femoral head, 608805; Legg-Calve-Perthes disease, 150600; Stickler sydrome, type I, nonsyndromic ocular, 609508; Czech dysplasia, 609162	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL4A3	Alport syndrome, autosomal recessive, 203780;Hematuria, benign familial, 141200;Alport syndrome, autosomal dominant, 104200	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL4A4	Alport syndrome 2, autosomal recessive, OMIM:203780;Hematuria,familial benign, OMIM:141200	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL6A1	Bethlem myopathy 1, OMIM:158810;Ullrich congenital muscular dystrophy 1, OMIM:254090	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL6A3	Bethlem myopathy, OMIM:158810;Ullrich congenital muscular dystrophy, OMIM:254090	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL9A1	Stickler syndrome, type IV, OMIM:614134;Epiphyseal dysplasia, multiple, 6, OMIM:614135	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL9A2	Stickler syndrome, type V, OMIM:614284;Epiphyseal dysplasia, multiple, 2, OMIM:600204	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL9A3	Epiphyseal dysplasia, multiple, 3, with or without myopathy, OMIM:600969	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COMP	Pseudoachondroplasia, 177170;Epiphyseal dysplasia, multiple 1, 132400	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CP	[Hypoceruloplasminemia, hereditary], 604290; Cerebellar ataxia, 604290; Hemosiderosis, systemic, due to aceruloplasminemia, 604290	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CPA6	Epilepsy, familial temporal lobe, 5	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CRX	Cone-rod retinal dystrophy-2, 120970;Leber congenital; amaurosis 7, 613829	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRYBA1	Cataract 10, multiple types, 600881	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRYBA4	Cataract 23 610425	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRYBB2	Cataract 3, multiple types, 601547	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRYBB3	Cataract 22, OMIM:609741	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CRYGC	Cataract 2, multiple types 604307	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRYGD	Cataract 4, multiple types, 115700	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CSF1R	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DDX58	Singleton-Merten syndrome 2, MIM: 616298	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
DIP2B	Mental retardation, FRA12A type, 136630;MENTAL RETARDATION, FRA12A TYPE	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
DNM2	Charcot Marie Tooth disease, axonal, type 2M, 606482;Charcot Marie Tooth disease, dominant intermediate B, 606482	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
DNMT1	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant;Neuropathy, hereditary sensory, type IE	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DPP6	Intellectual developmental disorder, autosomal dominant 33, OMIM:616311	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
DSPP	Dentinogenesis imperfecta, Shields type II, 125490;Deafness, autosomal dominant 36, with dentinogenesis, 605594;Dentinogenesis imperfecta, Shields type III, 125500;Dentin dysplasia, type II, 125420	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EDNRA	Migraine, resistance to, 157300	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EDNRB	ABCD syndrome, 600501;ALBINISM, BLACK LOCK, CELL MIGRATION DISORDER OF THE NEUROCYTES OF THE GUT, AND DEAFNESS	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
EGR2	Gene2Phenotype confirmed gene with ID HPO	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EIF4G1	Parkinson disease 18, 614251	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ELN	Supravalvar aortic stenosis, 185500;Cutis laxa, AD, 123700	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EXT1	Gene2Phenotype confirmed gene with ID HPO	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EYA1	Branchiootorenal syndrome 1, with or without cataracts, 113650; Anterior segment anomalies with or without cataract, 113650; Branchiootic syndrome 1, 602588; ?Otofaciocervical syndrome, 166780	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FBN1	Marfan syndrome 154700;MASS syndrome 604308;Weill-Marchesani syndrome 2, dominant 608328;Marfan lipodystrophy syndrome 616914	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FBN2	Gene2Phenotype confirmed gene with ID HPO	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGF10	Aplasia of lacrimal and salivary glands, 180920;LADD syndrome, 149730	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGFR1	Hartsfield syndrome, OMIM:615465;Pfeiffer syndrome, OMIM:101600;Encephalocraniocutaneous lipomatosis, somatic mosaic, OMIM:613001	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGFR3	CATSHL syndrome 610474;Hypochondroplasia 146000;SADDAN 616482;Muenke syndrome 602849;Thanatophoric dysplasia, type I 187600	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
FKBP6	Williams-Beuren syndrome	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FLNB	Spondylocarpotarsal synostosis syndrome, 272460; Larsen syndrome, 150250; Atelosteogenesis, type I, 108720; Atelosteogenesis, type III, 108721; Boomerang dysplasia, 112310	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
FLT4	Lymphedema, hereditary, IA, 153100;Hemangioma, capillary; infantile, somatic, 602089	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXC1	Iridogoniodysgenesis, type 1, 601631;Rieger or Axenfeld; anomalies, 602482;Axenfeld-Rieger syndrome, type 3, 602482;Iris hypoplasia and glaucoma, 601631	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXC2	Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis; syndrome with renal disease and diabetes mellitus, 153400	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXE3	Anterior segment mesenchymal dysgenesis, 107250;Aphakia, congenital primary, 610256	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
FOXF1	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FTL	Neurodegeneration with brain iron accumulation 3 606159;Hyperferritinemia-cataract syndrome;L-ferritin deficiency, dominant and recessive	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GATA2	Immunodeficiency 21, 614172;Emberger syndrome, 614038;{Myelodysplastic syndrome, susceptibility to}, 614286;{Leukemia, acute; myeloid, susceptibility to}, 601626	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GATA4	Atrial septal defect 2, 607941;Ventricular septal defect 1, 614429;Atrioventricular septal defect 4, 614430;?Testicular anomalies with or without congenital heart disease, 615542;Tetralogy of; Fallot, 187500	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GDAP1	Charcot-Marie-Tooth disease, axonal, type 2K, OMIM:607831;Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, OMIM:607706;Charcot-Marie-Tooth disease, recessive intermediate, A, OMIM:608340;Charcot-Marie-Tooth disease, type 4A, OMIM:214400	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GDF5	Acromesomelic dysplasia, Hunter-Thompson type, 201250;Brachydactyly, type C, 113100;Chondrodysplasia, Grebe type, 200700;Du Pan syndrome, 228900;Brachydactyly, type A2, 112600;Symphalangism, proximal, 1B, 615298;Multiple synostoses syndrome 2, 610017;{Osteoarthritis-5}, 612400;Brachydactyly, type A1, C, 615072	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GDF6	Klippel-Feil syndrome 1, autosomal dominant, 118100;Microphthalmia, isolated 4, 613094;Microphthalmia with coloboma 6, digenic, 613703;Leber congenital amaurosis 17, 615360	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GJA1	Gene2Phenotype confirmed gene with ID HPO	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GJA3	Cataract 14, multiple types, 601885	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GJA8	Cataract 1, multiple types, 116200	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GJB2	Deafness, autosomal recessive 1A, 220290;Deafness, autosomal; dominant 3A, 601544;Vohwinkel syndrome, 124500;Keratoderma, palmoplantar, with deafness, 148350;Keratitis-ichthyosis-deafness; syndrome, 148210;Hystrix-like ichthyosis with deafness, 602540;Bart-Pumphrey syndrome, 149200	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GLMN	Glomuvenous malformations, 138000	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GLRA1	Hyperekplexia;developmental delay;infantile spasms and generalized tonic-clonic seizures	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GLUD1	Gene2Phenotype confirmed gene with ID HPO	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GPHN	Molybdenum cofactor deficiency C, OMIM:615501	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GRN	Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485; Aphasia, primary progressive, 607485; Ceroid lipofuscinosis, neuronal, 11, 614706	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GUCY2C	Diarrhea 6, 614616;Meconium ileus, 614665	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
HNF4A	MODY, type I, 125850; {Diabetes mellitus, noninsulin-dependent}, 125853; Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HOXA13	Hand-foot-uterus syndrome, 140000;Guttmacher syndrome, 176305	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HOXD13	Synpolydactyly, type II, 186000;Brachydactyly, type E, 113300;Brachydactyly, type D, 113200;Synpolydactyly with foot anomalies, 186000;Syndactyly, type V, 186300;Brachydactyly-syndactyly syndrome, 610713;?VACTERL association, 192350	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HSF4	Cataract 5, multiple types, OMIM:116800	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

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IFITM5	Osteogenesis imperfecta, type V, 610967	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
IHH	Acrocapitofemoral dysplasia, OMIM:607778;Brachydactyly, type A1, OMIM:112500	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
IRF6	van der Woude syndrome, 119300;Popliteal pterygium syndrome 1, 119500;Orofacial cleft 6, 608864	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
JAG1	Alagille syndrome 1, OMIM:118450	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KBTBD13	Nemaline myopathy 6, autosomal dominant, 609273	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCND3	Spinocerebellar ataxia 19, 607346	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNE1	Jervell and Lange-Nielsen syndrome 2, OMIM:612347;Long QT syndrome 5, OMIM:613695	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KIF22	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KIT	Piebaldism, 172800;Gastrointestinal stromal tumor, familial, 606764;Mast cell disease, 154800;Leukemia, acute myeloid, 601626;Germ cell tumors, 273300	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KLF1	Blood group--Lutheran inhibitor, 111150;[Hereditary; persistence of fetal hemoglobin], 613566;Dyserythropoietic anemia, congenital, type IV, 613673	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KRIT1	CEREBRAL CAVERNOUS MALFORMATIONS TYPE 1 (CCM1)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
LDB3	Myopathy, myofibrillar, 4, 609452; Cardiomyopathy, dilated 1C, 601493; Left ventricular noncompaction 3, with or without dilated cardiomyopathy, 601493	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LEMD3	Osteopoikilosis, 166700;Buschke-Ollendorff syndrome, 166700;Melorheostosis with osteopoikilosis, 155950	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LGI1	Epilepsy, familial temporal lobe, 1 600512;AUTOSOMAL DOMINANT PARTIAL EPILEPSY WITH AUDITORY FEATURES	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
LHX4	Pituitary hormone deficiency, combined, 4, 262700	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LITAF	Charcot-Marie-Tooth disease, type 1C, 601098	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LMNA	Emery-Dreifuss muscular dystrophy 2, AD, 181350; Cardiomyopathy, dilated, 1A, 115200; Lipodystrophy, familial partial, 2, 151660; Emery-Dreifuss muscular dystrophy 3, AR, 181350; Charcot-Marie-Tooth disease, type 2B1, 605588; Muscular dystrophy, congenital, 613205; Muscular dystrophy, limb-girdle, type 1B, 159001; Mandibuloacral dysplasia, 248370; Hutchinson-Gilford progeria, 176670; Restrictive dermopathy, lethal, 275210; Heart-hand syndrome, Slovenian type, 610140; Malouf syndrome, 212112	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
LMX1B	Nail-patella syndrome, 161200	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LRRK2	Parkinson disease 8, 607060	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MAFB	Duane retraction syndrome 3 (617041);Multicentric carpotarsal osteolysis syndrome (166300)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MAGI2	Epileptic encephalopathy;Infantile spasms	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MAP3K1	46XY sex reversal 6, 613762	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MAPT	Dementia, frontotemporal, with or without parkinsonism, 600274; Pick disease, 172700; Supranuclear palsy, progressive, 601104; Supranuclear palsy, progressive atypical, 260540; {Parkinson disease, susceptibility to}, 168600	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MATN3	Epiphyseal dysplasia, multiple, 5, 607078;{Osteoarthritis; susceptibility 2}, 140600;Spondyloepimetaphyseal dysplasia, 608728	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MITF	Waardenburg syndrome, type 2A, 193510; Waardenburg; syndrome/ocular albinism, digenic, 103470; Tietz albinism-deafness syndrome, 103500; {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MMP13	Spondyloepimetaphyseal dysplasia, Missouri type, 602111;Metaphyseal anadysplasia 1, 602111	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MNX1	Gene2Phenotype confirmed gene with ID HPO	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MPZ	Associated with Charcot-Marie-Tooth disease, dominant intermediate D (607791);Charcot-Marie-Tooth disease, type 1B	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MSX1	Ectodermal dysplasia 3, Witkop type, OMIM:189500;Orofacial cleft 5, OMIM:608874;Tooth agenesis, selective, 1, with or without orofacial cleft, OMIM:106600	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MSX2	Craniosynostosis, type 2, 604757;Parietal foramina 1, 168500;Parietal foramina with cleidocranial dysplasia, 168550	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MTMR14	{Centronuclear myopathy, autosomal, modifier of}, 160150;Autosomal dominant centronuclear myopathy	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYBPC1	Arthrogryposis, distal, type 1B 614335 AD;Lethal congenital contracture syndrome 4 614915 AR	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MYH3	Gene2Phenotype confirmed gene with ID HPO	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYH6	Cardiomyopathy, familial hypertrophic, 14, 613251;Atrial septal; defect 3, 614089;Cardiomyopathy, dilated, 1EE, 613252;{Sick sinus syndrome 3}, 614090	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYH8	Carney complex variant, 608837;Trismus-pseudocamptodactyly; syndrome, 158300	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYH9	May-Hegglin anomaly, 155100;Fechtner syndrome, 153640;Sebastian syndrome, 605249;Deafness, autosomal dominant 17, 603622;Epstein syndrome, 153650;Macrothrombocytopenia and progressive; sensorineural deafness, 600208	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYT1	Intellectual disability, MONDO:0001071	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NDN	Smith-Magenis-like syndrome	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, maternally imprinted (paternal allele expressed)
NDRG1	Charcot-Marie-Tooth disease, type 4D, 601455	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NEFL	Charcot Marie Tooth disease, type 2E, 607684;Charcot Marie Tooth disease, type 1F, 607734	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
NIPA1	Spastic paraplegia 6, autosomal dominant, 600363;Non Imprinted In Prader-Willi/Angelman Syndrome 1	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NKX2-5	Atrial septal defect 7, with or without AV conduction defects, 108900;Tetrology of Fallot, 187500;Hypothyroidism, congenital nongoitrous, 5, 225250;Ventricular septal defect 3, 614432;Hypoplastic left heart syndrome 2, 614435;Conotruncal heart malformations, variable, 217095	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NLRP3	CINCA syndrome, OMIM:607115	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NODAL	Heterotaxy, visceral, 5, 270100	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NOG	Symphalangism, proximal, 185800;Multiple synostoses syndrome 1, 186500;Tarsal-carpal coalition syndrome, 186570;Stapes ankylosis with broad thumb and toes, 184460;Brachydactyly, type B2, 611377	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NOTCH2	Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NR1I3	EHMT1-like Intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NR5A1	46XY sex reversal 3, 612965;Premature ovarian failure 7, 612964;Adrenocortical insufficiency;Spermatogenic failure 8, 613957	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NRXN2	autism spectrum disorder;intellectual disability	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NRXN3	Autism spectrum disorder	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PAX2	Gene2Phenotype confirmed gene with ID HPO	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PAX3	Waardenburg syndrome, type 1, 193500;Waardenburg syndrome, type; 3, 148820;Craniofacial-deafness-hand syndrome, 122880;Rhabdomyosarcoma 2, alveolar, 268220	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PAX6	Aniridia, Cerebellar Ataxia, And Mental Retardation;Aniridia, 106210Peters anomaly, 604229Cataract with late-onset corneal dystrophy, 106210Keratitis, 148190Foveal hyperplasia, 136520Morning glory disc anomaly, 120430Optic nerve hypoplasia, 165550Coloboma, ocular, 120200Coloboma of optic nerve, 120430Gillespie syndrome, 206700;KERATITIS HEREDITARY (KERH)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PAX9	Tooth agenesis, selective, 3, 604625	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PDCD10	CEREBRAL CAVERNOUS MALFORMATIONS TYPE 3 (CCM3)	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PDGFB	Meningioma, SIS-related, 607174; Dermatofibrosarcoma protuberans, 607907; Basal ganglia calcification, idiopathic, 5, 615483	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PDYN	Spinocerebellar ataxia 23, 610245	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PHOX2B	CENTRAL HYPOVENTILATION SYNDROME, CONGENITAL, WITH OR WITHOUT HIRSCHSPRUNG DISEASE	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PIEZO2	Gene2Phenotype confirmed gene with ID HPO	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PIK3R1	?Agammaglobulinemia 7, autosomal recessive, 615214; SHORT; syndrome, 269880; Immunodeficiency 36, 616005	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PITX2	Axenfeld-Rieger syndrome, type 1, 180500;Iridogoniodysgenesis, type 2, 137600;Ring dermoid of cornea, 180550;Peters anomaly, 604229	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PITX3	Anterior segment mesenchymal dysgenesis, 107250; Cataract 11, multiple types, 610623; Cataract 11, syndromic, 610623	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PMP22	Charcot-Marie-Tooth disease, type 1A, 118220; Dejerine-Sottas; disease, 145900; Neuropathy, recurrent, with pressure palsies, 162500; Charcot-Marie-Tooth disease, type 1E, 118300; Roussy-Levy syndrome, 180800; Neuropathy, inflammatory demyelinating, 139393	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PNKD	Paroxysmal nonkinesigenic dyskinesia, 118800	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
POLD1	{Colorectal cancer, susceptibility to, 10}, 612591;Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
POLR1D	Treacher Collins syndrome 2, OMIM:613717	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PRICKLE1	Epilepsy, progressive myoclonic 1B, OMIM:612437	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PRKAR1A	Acrodysostosis 1, with or without hormone resistance, OMIM:101800	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PRKCG	Spinocerebellar ataxia 14, 605361	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PSEN1	Alzheimer disease, type 3, 607822; Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822; Dementia, frontotemporal, 600274; Pick disease, 172700	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
QKI	INTELLECTUAL DISABILITY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
RANBP2	{Encephalopathy, acute, infection-induced, 3, susceptibility to}608033	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
RASA1	Parkes Weber syndrome, 608355;Capillary; malformation-arteriovenous malformation, 608354;Basal cell carcinoma, somatic, 605462	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
REEP1	Spastic paraplegia 31, autosomal dominant, 610250; ?Neuronopathy, distal hereditary motor, type VB, 614751	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RET	Gene2Phenotype both DD and IF gene with ID HPO	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

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RNF135	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ROR2	Brachydactyly, type B1, OMIM:113000 (AD);Robinow syndrome, autosomal recessive, OMIM:268310 (AR)	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
RPS19	Diamond-Blackfan anemia 1, 105650	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RTN2	Spastic paraplegia 12, autosomal dominant, 604805	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RUNX2	Cleidocranial dysplasia, 119600;Cleidocranial dysplasia, forme; fruste, with brachydactyly, 119600;Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600;Metaphyseal dysplasia with maxillary; hypoplasia with or without brachydactyly, 156510	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RYR3	EPILEPTIC ENCEPHALOPATHY	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SALL4	Duane-radial ray syndrome, 607323;IVIC syndrome, 147750	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCN4A	Hyperkalemic periodic paralysis, type 2, 170500; Paramyotonia; congenita, 168300; Myotonia congenita, atypical, acetazolamide-responsive, 608390; Myasthenic syndrome, acetazolamide-responsive, 614198; Hypokalemic periodic paralysis, type 2, 613345	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCN9A	Erythermalgia, primary, 133020; Paroxysmal extreme pain disorder, 167400, Insensitivity to pain, congenital, 243000; Febrile seizures, familial, 3B, 613863; Epilepsy, generalized, with febrile seizures plus, type 7, 613863; Small fiber neuropathy, 133020; {Dravet syndrome, modifier of}, 607208; HSAN2D, autosomal recessive, 243000	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SCRIB	8Q24.3 DELETION-LIKE	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SEMA3E	Severe Intellectual Disability with Cognitive Regression	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SETX	Ataxia-ocular apraxia-2, 606002; Amyotrophic lateral sclerosis; 4, juvenile, 602433	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SF3B4	Gene2Phenotype confirmed gene with ID HPO	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SGCE	Dystonia-11, myoclonic, 159900	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SHOX	Langer mesomelic dysplasia, OMIM:249700 (PR);Leri-Weill dyschondrosteosis, OMIM:127300 (PD);Short stature, idiopathic familial, OMIM:300582	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SIX1	Brachiootic syndrome 3, 608389;Deafness, autosomal dominant 23, 605192	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SIX5	Branchiootorenal syndrome 2, 610896	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SLC20A2	Basal ganglia calcification, idiopathic, 1; 13600;Idiopathic basal ganglia calcification, adult onset; Basal ganglia calcification, idiopathic, childhood onset	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SLC6A5	hyperekplexia (MIM:614618) and intellectual disability	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SLC9A9	{?Autism susceptibility 16}, 613410	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SNCA	Parkinson disease 4, 605543; Dementia, Lewy body, 127750; Parkinson disease 1, 168601	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SNX3	microcephaly, microphthalmia, ectrodactyly, prognathism (MMEP); mental retardation	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SOX17	Vesicoureteral reflux 3, 613674	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SPAST	Spastic paraplegia 4, autosomal dominant, 182601	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SPG7	Spastic paraplegia 7, autosomal recessive, OMIM:607259;hereditary spastic paraplegia 7, MONDO:0011803	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SPTLC2	Neuropathy, hereditary sensory and autonomic, type IC, 613640	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
STAT1	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892;Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796;Immunodeficiency 31C, autosomal dominant, 614162	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
STAT5B	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, OMIM:245590;Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, OMIM:618985	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
TARDBP	Amyotrophic lateral sclerosis 10, with or without FTD, 612069; Frontotemporal lobar degeneration, TARDBP-related, 612069	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX20	Atrial septal defect 4, 611363	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX3	Ulnar-mammary syndrome, 181450	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX4	Small patella syndrome, 147891	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX5	Holt-Oram syndrome, 142900	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TCOF1	Gene2Phenotype confirmed gene with ID HPO	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TEK	Venous malformations, multiple cutaneous and mucosal, 600195	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TFAP2A	Branchiooculofacial syndrome, 113620	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TFAP2B	Char syndrome, 169100	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGFB2	Loeys-Dietz syndrome, type 4, 614816	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGFB3	Arrhythmogenic right ventricular dysplasia 1, 107970;?Rienhoff syndrome, 615582	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGM6	Spinocerebellar ataxia 35, 613908	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
THAP1	DYSTONIA 6, TORSION	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TINF2	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TP63	ADULT syndrome, OMIM:103285;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, OMIM:604292;Hay-Wells syndrome, OMIM:106260;Limb-mammary syndrome, OMIM:603543;Orofacial cleft 8, OMIM:618149;Rapp-Hodgkin syndrome, OMIM:129400;Split-hand/foot malformation 4, OMIM:605289	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TRPS1	Trichorhinophalangeal syndrome, type I, 190350;Trichorhinophalangeal syndrome, type III, 190351	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TRPV4	Brachyolmia type 3, 113500;Spondylometaphyseal dysplasia, Kozlowski type, 184252;Metatropic dysplasia, 156530;Hereditary motor and sensory neuropathy, type IIc, 606071;Scapuloperoneal spinal; muscular atrophy, 181405;[Sodium serum level QTL 1], 613508;Parastremmatic dwarfism, 168400;SED, Maroteaux type, 184095;Spinal muscular atrophy, distal, congenital nonprogressive, 600175;Digital arthropathy-brachydactyly, familial, 606835	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TSHR	Hypothyroidism, congenital, nongoitrous, 1 275200; Thyroid; adenoma, hyperfunctioning, somatic; Hyperthyroidism, nonautoimmune, 609152; Thyroid carcinoma with thyrotoxicosis; Hyperthyroidism, familial; gestational, 603373	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
TTBK2	Spinocerebellar ataxia 11, 604432	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
VIP	Asperger syndrome	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
VPS35	Parkinson disease 17, 614203	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
WFS1	Wolfram syndrome 1, 222300;Wolfram-like syndrome, autosomal dominant, 614296	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
WNT4	MULLERIAN APLASIA AND HYPERANDROGENISM	Intellectual disability	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
WNT5A	Gene2Phenotype confirmed gene with ID HPO	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
WT1	Wilms tumor, type 1, 194070;Denys-Drash syndrome, 194080;Nephrotic syndrome, type 4, 256370;Frasier syndrome, 136680;Meacham syndrome, 608978;Mesothelioma, somatic, 156240	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
YAP1	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
YWHAE	neurodevelopmental disorder, MONDO:0700092	Intellectual disability	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ABCC9	CANTU SYNDROME HYPERTRICHOTIC OSTEOCHONDRODYSPLASIA 239850	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ACAN	SPONDYLOEPIMETAPHYSEAL DYSPLASIA AGGRECAN TYPE 612813;SPONDYLOEPIPHYSEAL DYSPLASIA TYPE KIMBERLEY 608361	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ACTA2	AORTIC ANEURYSM, FAMILIAL THORACIC 6 611788;MOYAMOYA DISEASE 5 614042	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ACTB	BARAITSER-WINTER SYNDROME;ACTB Haploinsufficiency syndtome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ACTC1	ACTC1-related distal arthrogryposis with congenital heart disease	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ACTG1	BARAITSER-WINTER SYNDROME	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ACVR1	FIBRODYSPLASIA OSSIFICANS PROGRESSIVA 135100	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ADAR	AICARDI-GOUTIERES SYNDROME ASSOCIATED WITH A TYPE I INTERFERON SIGNATURE 615010;DYSCHROMATOSIS SYMMETRICA HEREDITARIA 1 127400	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ADCY5	ADCY5-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ADNP	MENTAL RETARDATION, AUTOSOMAL DOMINANT, 28 615873	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AFF3	Skeletal dysplasia with severe neurological disease	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AGO1	AGO1-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AKT1	PROTEUS SYNDROME 176920	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AKT2	AKT2-related hypoinsulinemic hypoglycemia and hemihypertrophy, OMIM:240900	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AKT3	HEMIMEGALENCEPHALY AKT3 603387	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ALDH18A1	SPASTIC PARAPLEGIA 9, AUTOSOMAL DOMINANT 601162;CUTIS LAXA, AUTOSOMAL DOMINANT 3 616603;MENTAL RETARDATION-JOINT HYPERMOBILITY-SKIN LAXITY WITH OR WITHOUT METABOLIC ABNORMALITIES 612652	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ALX4	PARIETAL FORAMINA 2 609597;FRONTONASAL DYSPLASIA 2 613451	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ANKH	CHONDROCALCINOSIS 2 118600;CRANIOMETAPHYSEAL DYSPLASIA JACKSON TYPE 123000	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ANKRD11	KBG SYNDROME 148050	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ANKRD26	THROMBOCYTOPENIA 2 188000	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

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ANO5	LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 2L, OMIM:611307;GNATHODIAPHYSEAL DYSPLASIA, OMIM:166260	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ARHGAP31	ADAMS-OLIVER SYNDROME 1 100300	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ARID1A	COFFIN-SIRIS SYNDROME 135900	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ARID1B	MENTAL RETARDATION, AUTOSOMAL DOMINANT 12 135900;COFFIN SIRIS SYNDROME 135900	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ASXL1	BOHRING-OPITZ SYNDROME 605039	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ATL1	ATL1-associated hereditary spastic paraplegia	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ATN1	Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, OMIM:618494	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ATP1A2	ATP1A2-related epileptic encephalopathy;MIGRAINE, FAMILIAL HEMIPLEGIC, ATP1A2-related;Autosomal recessive ATP1A2-related neuronal migration disorder with epilepsy	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ATP1A3	RAPID-ONSET DYSTONIA-PARKINSONISM 128235;ALTERNATING HEMIPLEGIA OF CHILDHOOD 104290	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ATP6V0A1	ATP6V0A1-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AUTS2	SYNDROMIC INTELLECTUAL DISABILITY 612100	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BAP1	BAP1-associated neurodevelopmental syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BCL11A	INTELLECTUAL DISABILITY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BFSP2	CATARACT AUTOSOMAL DOMINANT BFSP2-RELATED 611597	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BHLHA9	SPLIT HAND AND FOOT MALFORMATION 220600;MESOAXIAL SYNOSTOTIC SYNDACTYLY WITH PHALANGEAL REDUCTION, MALIK-PERCIN TYPE 69432	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
BMP2	Short stature, palatal anomalies, congenital heart disease, and skeletal malformations	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BMP4	MICROPHTHALMIA, SYNDROMIC 6 607932;OROFACIAL CLEFT 11 600625	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BNC2	Congenital Lower Urinary Tract Obstruction	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BRAF	LEOPARD SYNDROME TYPE 3 613707;NOONAN SYNDROME TYPE 7 613706;CARDIOFACIOCUTANEOUS SYNDROME 115150	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BRSK2	Neurodevelopmental Disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CACNA1A	EPILEPTIC ENCEPHALOPATHY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CACNA1C	Timothy syndrome, OMIM:601005;Timothy syndrome, MONDO:0010979;CACNA1C-related disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CACNA1D	PRIMARY ALDOSTERONISM, SEIZURES, AND NEUROLOGIC ABNORMALITIES 615474;SINOATRIAL NODE DYSFUNCTION AND DEAFNESS 614896	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CACNA1E	Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesia	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CACNA1G	AUTOSOMAL RECESSIVE MENTAL RETARDATION;CACNA1G-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CAMTA1	CEREBELLAR ATAXIA, NONPROGRESSIVE, WITH MENTAL RETARDATION 614756	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CBFB	CBFB-related cleidocranial dysplasia	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CBL	NOONAN SYNDROME-LIKE DISORDER WITH OR WITHOUT JUVENILE MEYLOMONOCYTIC LEUKEMIA 613563	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CDH1	Blepharo-cheiro-dontic syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CDKN1C	BECKWITH-WIEDEMANN SYNDROME 130650;IMAGe Syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)
CDON	HOLOPROSENCEPHALY 11 614226	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CEP85L	CEP85L-associated posterior-predominant lissencephaly, OMIM:618873	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CFC1	CFC1-RELATED CONOTRUNCAL HEART MALFORMATIONS 319372	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHD2	EPILEPTIC ENCEPHALOPATHY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHD3	Macrocephaly and impaired speech and language	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHD7	CHARGE SYNDROME 214800;IDIOPATHIC HYPOGONADOTROPIC HYPOGONADISM 146110;KALLMANN SYNDROME TYPE 5 612370	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHD8	AUTISM 209850	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHRNA4	NOCTURNAL FRONTAL LOBE EPILEPSY TYPE 1 600513	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHRNB1	CHRNB1-related congenital myaesthenia, biallelic, OMIM:616314;CHRNB1-related congenital myaesthenia, monoallelic, OMIM:616313	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CHRNB2	CHRNB2-RELATED NOCTURNAL FRONTAL LOBE EPILEPSY, AUTOSOMAL DOMINANT 605375;NOCTURNAL FRONTAL LOBE EPILEPSY, AUTOSOMAL DOMINANT 117426	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CIC	CAPICUA, DROSOPHILA, HOMOLOG OF 612082	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CNNM2	CNNM2-related neurodevelopmental disorder with hypomagnesemia;autosomal recessive form	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CNOT3	CNOT3 syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COG4	COG4-CDG 319493;Saul-Wilson syndrome 618150	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL10A1	SCHMID TYPE METAPHYSEAL CHONDRODYSPLASIA 156500	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL11A1	STICKLER SYNDROME, TYPE II 604841;FIBROCHONDROGENESIS 228520	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL11A2	WEISSENBACHER-ZWEYMUELLER SYNDROME 184840;AUTOSOMAL RECESSIVE OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA 215150;DEAFNESS AUTOSOMAL DOMINANT TYPE 13 601868;STICKLER SYNDROME TYPE 3 184840;DEAFNESS AUTOSOMAL RECESSIVE TYPE 53 609706	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL1A1	CAFFEY DISEASE 114000;OSTEOGENESIS IMPERFECTA TYPE IIA 166210;OSTEOGENESIS IMPERFECTA TYPE III 259420;COL1A1/2-RELATED OSTEOGENESIS IMPERFECTA 166210;EHLERS-DANLOS SYNDROME, CLASSIC TYPE, COL1A1-RELATED 130060;OSTEOGENESIS IMPERFECTA TYPE I 166200;EHLERS-DANLOS SYNDROME TYPE VIIA 319158	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL2A1	ACHONDROGENESIS TYPE 2 200610;PLATYSPONDYLIC LETHAL SKELETAL DYSPLASIA TORRANCE TYPE 151210;SPONDYLOPERIPHERAL DYSPLASIA 271700;PRIMARY AVASCULAR NECROSIS OF FEMORAL HEAD 608805;SPONDYLOEPIMETAPHYSEAL DYSPLASIA STRUDWICK TYPE 184250;SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA 183900;RHEGMATOGENOUS RETINAL DETACHMENT AUTOSOMAL DOMINANT 609508;KNIEST DYSPLASIA 156550;STICKLER SYNDROME TYPE 1 NON-SYNDROMIC OCULAR 609508	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL4A1	PORENCEPHALY 1 175780	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL4A2	PORENCEPHALY 2 614483	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL4A3	ALPORT SYNDROME AUTOSOMAL RECESSIVE, OMIM:203780;ALPORT SYNDROME AUTOSOMAL DOMINANT, OMIM:104200	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL6A1	COL6A1 associated myopathy	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL6A2	COL6A2-related Ullrich congenital muscular dystrophy (monoallelic), OMIM:254090;COL6A2-related Ullrich congenital muscular dystrophy (biallelic), OMIM:254090	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL9A1	STICKLER SYNDROME TYPE 4 614134;MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 6 614135	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL9A2	MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 2 600204;STICKLER SYNDROME, TYPE V 614284	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL9A3	MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 3 600969;Stickler syndrome	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CREBBP	RUBINSTEIN-TAYBI SYNDROME TYPE 1 180849;CREBBP intellectual disability without typical RTS features	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRELD1	HETEROTAXY SYNDROME 207574	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRX	CRX-RELATED LEBER CONGENITAL AMAUROSIS LEBER CONGENITAL AMAUROSIS 7 613829	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRYAA	CATARACT, NUCLEAR 123580;CATARACT, AUTOSOMAL RECESSIVE CONGENITAL 1 604219	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CRYBA1	CATARACT CONGENITAL ZONULAR WITH SUTURAL OPACITIES 600881	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRYBB1	CATARACT 17, MULTIPLE TYPES, MONOALLELIC;CATARACT 17, MULTIPLE TYPES;CATARACT 17, MULTIPLE TYPES, BIALLELIC	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CRYBB2	CATARACT, CONGENITAL, CERULEAN TYPE, 2 601547;CATARACT, COPPOCK-LIKE 604307	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRYGC	CATARACT AUTOSOMAL DOMINANT 604219;CATARACT COPPOCK-LIKE 604307	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CTNNB1	MENTAL RETARDATION, AUTOSOMAL DOMINANT 19 615075	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CTNND1	Blepharo-cheiro-dontic syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CUL3	CUL3-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DEAF1	Autism, intellectual disability, basal ganglia dysfunction and epilepsy;MENTAL RETARDATION, AUTOSOMAL DOMINANT 24 615828	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
DHDDS	Epilepsy and intellectual disability	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DNM1	EPILEPTIC ENCEPHALOPATHY	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
DNM1L	DNM1L-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DNMT3A	Tatton-Brown Rahman syndrome (OVERGROWTH SYNDROME WITH INTELLECTUAL DISABILITY), 615879;Microcephalic primordial dwarfism	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DSP	DSP-related developmental disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DSPP	DENTINOGENESIS IMPERFECTA, SHIELDS TYPE II 125490;DEAFNESS AUTOSOMAL DOMINANT TYPE 39 WITH DENTINOGENESIS IMPERFECTA 1 605594	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DYNC1H1	SPINAL MUSCULAR ATROPHY, LOWER EXTREMITY-PREDOMINANT, AD 158600;SEVERE ID WITH NEURONAL MIGRATION DISORDER 600112	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DYRK1A	MENTAL RETARDATION AUTOSOMAL DOMINANT TYPE 7 614104	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EDAR	ECTODERMAL DYSPLASIA 10A, HYPOHIDROTIC/HAIR/NAIL TYPE, AUTOSOMAL DOMINANT;Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
EDN1	AURICULOCONDYLAR SYNDROME 602483	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
EDNRA	MANDIBULOFACIAL DYSOSTOSIS WITH ALOPECIA 616367	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

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EDNRB	ABCD SYNDROME, OMIM:600501	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EFTUD2	MANDIBULOFACIAL DYSOSTOSIS WITH MICROCEPHALY 610536	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EHMT1	Kleefstra syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ELN	ELN-RELATED CUTIS LAXA 314088;SUPRAVALVAR AORTIC STENOSIS 185500	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EP300	RUBINSTEIN-TAYBI SYNDROME TYPE 2 613684	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EXT1	HEREDITARY MULTIPLE EXOSTOSES TYPE 1 133700;TRICHO-RHINO-PHALANGEAL SYNDROME TYPE 2 150230	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EXT2	EXOSTOSES, MULTIPLE, TYPE 2 133701	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EYA1	BRANCHIOOTORENAL SYNDROME TYPE 1, OMIM:113650	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EZH2	WEAVER SYNDROME 2 614421	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FBN1	WEILL-MARCHESANI SYNDROME AUTOSOMAL DOMINANT, OMIM:608328;MARFAN SYNDROME, OMIM:154700;SHPRINTZEN-GOLDBERG CRANIOSYNOSTOSIS SYNDROME, OMIM:182212;Marfan Syndrome, biallelic, OMIM:154700	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
FBN2	Contractural arachnodactyly, congenital OMIM:121050;congenital contractural arachnodactyly MONDO:0007363	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FBXW11	SYNDROMIC INTELLECTUAL DISABILITY 612100	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FBXW7	FBXW7-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGF10	LADD SYNDROME 149730	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGF14	FGF14-related episodic ataxia	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGF9	MULTIPLE SYNOSTOSES SYNDROME TYPE 3 612961	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGFR1	Hypogonadotropic hypogonadism 2 with or without anosmia, OMIM:147950;Encephalocraniocutaneous lipomatosis, OMIM:613001;PFEIFFER SYNDROME, OMIM:101600;OSTEOGLOPHONIC DYSPLASIA, OMIM:166250;Hartsfield syndrome, OMIM:615465	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGFR2	BEARE-STEVENSON CUTIS GYRATA SYNDROME 123790;ANTLEY-BIXLER SYNDROME 207410;FAMILIAL SCAPHOCEPHALY SYNDROME 609579;JACKSON-WEISS SYNDROME 123150;APERT SYNDROME 101200;CROUZON SYNDROME 123500;LACRIMO-AURICULO-DENTO-DIGITAL SYNDROME 149730;ACROCEPHALOSYNDACTYLY TYPE V 101600	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGFR3	THANATOPHORIC DYSPLASIA TYPE 2 187601;THANATOPHORIC DYSPLASIA TYPE 1 187600;MUENKE SYNDROME 602849;ACHONDROPLASIA 100800;CROUZON SYNDROME WITH ACANTHOSIS NIGRICANS 612247;CAMPTODACTYLY TALL STATURE AND HEARING LOSS SYNDROME 610474;LACRIMO-AURICULO-DENTO-DIGITAL SYNDROME 149730;HYPOCHONDROPLASIA 146000	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FLNB	AUTOSOMAL DOMINANT LARSEN SYNDROME 150250;ATELOSTEOGENESIS TYPE 3 108721;ATELOSTEOGENESIS TYPE 1 108720;BOOMERANG DYSPLASIA 112310;SPONDYLOCARPOTARSAL SYNOSTOSIS SYNDROME 272460	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
FLT4	MILROY DISEASE 153100	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FN1	Spondylometaphyseal Dysplasia with Corner Fractures 184255	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXC1	IRIDOGONIODYSGENESIS ANOMALY 601631;AXENFELD-RIEGER SYNDROME TYPE 3 602482;PETERS ANOMALY 604229	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXC2	LYMPHEDEMA-DISTICHIASIS SYNDROME 153400;HEREDITARY LYMPHEDEMA II 241432	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXE3	CONGENITAL PRIMARY APHAKIA 610256;ANTERIOR SEGMENT MESENCHYMAL DYSGENESIS 107250	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
FOXF1	ALVEOLAR CAPILLARY DYSPLASIA WITH MISALIGNMENT OF PULMONARY VEINS 265380	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXG1	CONGENITAL VARIANT OF RETT SYNDROME 613454	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXL2	BLEPHAROPHIMOSIS, PTOSIS, AND EPICANTHUS INVERSUS SYNDROME 110100	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXP1	MENTAL RETARDATION WITH LANGUAGE IMPAIRMENT AND AUTISTIC FEATURES 613670	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXP2	SPEECH-LANGUAGE DISORDER 1 602081	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FTL	HEREDITARY HYPERFERRITINEMIA-CATARACT SYNDROME 600886	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GABBR1	GABBR1-associated neurodevelopmental disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GABRA1	JUVENILE MYOCLONIC EPILEPSY 611136;EPILEPTIC ENCEPHALOPATHY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GABRB3	EPILEPTIC ENCEPHALOPATHIES;CHILDHOOD ABSENCE EPILEPSY TYPE 5 612269	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GABRG2	EPILEPSY, GENERALIZED, WITH FEBRILE SEIZURES PLUS, TYPE 3 611277;GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, TYPE 3 611277	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GATA2	EMBERGER SYNDROME 614038	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GATA3	HYPOPARATHYROIDISM, SENSORINEURAL DEAFNESS, AND RENAL DISEASE, OMIM:146255	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GATA4	ATRIAL SEPTAL DEFECT TYPE 2 607941	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GATA6	PANCREATIC AGENESIS, DIAPHRAGMATIC HERNIA AND CONGENITAL HEART DEFECTS 600001;ATRIOVENTRICULAR SEPTAL DEFECT 5 614474;ATRIAL SEPTAL DEFECT 9 614475	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GCH1	DYSTONIA TYPE 5 128230;GTP CYCLOHYDROLASE 1 DEFICIENCY 233910	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GDF5	BRACHYDACTYLY TYPE C 113100;ACROMESOMELIC CHONDRODYSPLASIA GREBE TYPE 200700;ACROMESOMELIC CHONDRODYSPLASIA HUNTER-THOMPSON TYPE 201250;DU PAN SYNDROME 228900;SYMPHALANGISM PROXIMAL SYNDROME 185800;BRACHYDACTYLY TYPE A1 112500;BRACHYDACTYLY TYPE A2 112600;MULTIPLE SYNOSTOSES SYNDROME TYPE 2 610017	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GDF6	KLIPPEL-FEIL SYNDROME TYPE 1 118100;MICROPHTHALMIA ISOLATED TYPE 4 613094	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GFAP	ALEXANDER DISEASE 203450	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GJA1	HALLERMANN-STREIFF SYNDROME, OMIM:234100;SYNDACTYLY TYPE 3, OMIM:186100;AUTOSOMAL DOMINANT OCULODENTODIGITAL DYSPLASIA, OMIM:164200;HYPOPLASTIC LEFT HEART SYNDROME, OMIM:241550;AUTOSOMAL RECESSIVE OCULODENTODIGITAL DYSPLASIA, OMIM:257850	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GJA3	CATARACT ZONULAR PULVERULENT CATARACT TYPE 3 601885	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GJA8	CATARACT ZONULAR PULVERULENT TYPE 1 116200;CATARACT-MICROCORNEA SYNDROME 116150	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GJB3	DEAFNESS AUTOSOMAL DOMINANT TYPE 2B, OMIM:612644;ERYTHROKERATODERMIA VARIABILIS ET PROGRESSIVA, OMIM:133200;DEAFNESS, AUTOSOMAL RECESSIVE	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GJC2	SPASTIC PARAPLEGIA, 44 613206;LEUKODYSTROPHY, HYPOMYELINATING, 2 608804;LYMPHEDEMA, HEREDITARY, IC 613480	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GLI2	GLI2-RELATED HOLOPROSENCEPHALY 261768	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GLI3	PREAXIAL POLYDACTYLY TYPE IV 269157;GREIG CEPHALOPOLYSYNDACTYLY SYNDROME 175700;POSTAXIAL POLYDACTYLY TYPE A 174200;PALLISTER-HALL SYNDROME 146510	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GLMN	GLOMUVENOUS MALFORMATIONS 138000	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GLRA1	GLRA1-related hyperexplexia, biallelic, OMIM:149400;GLRA1-related hyperexplexia, monoallelic, OMIM:149400	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GLUD1	HYPERINSULINISM-HYPERAMMONEMIA SYNDROME 606762	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GNA14	Congenital vascular tumours	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GNAQ	Congenital Hemangioma	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GNAS	ALBRIGHT HEREDITARY OSTEODYSTROPHY, OMIM:103580;MCCUNE-ALBRIGHT SYNDROME, OMIM:174800;PSEUDOHYPOPARATHYROIDISM TYPE 1B, OMIM:603233	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GRIK2	MENTAL RETARDATION AUTOSOMAL RECESSIVE TYPE 6 611092	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant OMIM:614254;intellectual disability, autosomal dominant 8 MONDO:0013655	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GRIN2A	LANDAU-KLEFFNER SYNDROME 245570;EPILEPSY WITH NEURODEVELOPMENTAL DEFECTS 613971	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GRIN2B	EPILEPTIC ENCEPHALOPATHY;AUTISM 209850;MENTAL RETARDATION, AUTOSOMAL DOMINANT 6 613970	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GUCY2C	MECONIUM ILEUS;FAMILIAL DIARRHEA DIARRHEA 6 614616	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
HCN1	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 24 615871	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HDAC4	BRACHYDACTYLY-MENTAL RETARDATION SYNDROME 600430	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HESX1	SEPTOOPTIC DYSPLASIA 256657;HESX1-RELATED COMBINED PITUITARY HORMONE DEFICIENCY 319358	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
HK1	HK1-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HNF1B	RENAL CYSTS AND DIABETES SYNDROME 137920	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HNF4A	HNF4A-RELATED MATURITY-ONSET DIABETES OF THE YOUNG TYPE 1 125850;ATYPICAL DOMINANT FANCONI SYNDROME WITH MODY 315353	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HNRNPU	EPILEPTIC ENCEPHALOPATHY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HOXA13	HAND-FOOT-GENITAL SYNDROME 140000	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HOXD13	BRACHYDACTYLY-SYNDACTYLY SYNDROME 610713;VACTERL ASSOCIATION 192350;BRACHYDACTYLY TYPE E 113300;SYNPOLYDACTYLY 1 186000;SYNDACTYLY TYPE 5 186300;BRACHYDACTYLY TYPE D 113200	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

HPD	TYROSINEMIA TYPE 3 276710;HAWKINSINURIA 140350	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
HRAS	CONGENITAL MYOPATHY WITH EXCESS OF MUSCLE SPINDLES 218040;COSTELLO SYNDROME 218040	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HSF4	CATARACT ZONULAR HSF4-RELATED 116800;CATARACT MARNER TYPE 116800	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
IFIH1	AICARDI-GOUTIERES SYNDROME 7, OMIM:615846;SINGLETON-MERTEN SYNDROME, OMIM:182250	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
IFITM5	OSTEOGENESIS IMPERFECTA TYPE V 610967	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
IGF1R	INSULIN-LIKE GROWTH FACTOR I, RESISTANCE TO 270450	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
IGF2	BECKWITH-WIEDEMANN SYNDROME 130650;CHROMOSOME 11P15.5-RELATED RUSSELL-SILVER SYNDROME 180860	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, maternally imprinted (paternal allele expressed)
IHH	ACROCAPITOFEMORAL DYSPLASIA 607778;BRACHYDACTYLY, TYPE A1 112500	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
IRF6	VAN DER WOUDE SYNDROME 119300;POPLITEAL PTERYGIUM SYNDROME 119500	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ITPR1	Gillespie Syndrome, OMIM:206700;Gillespie Syndrome, monoallelic, OMIM:206700;SPINOCEREBELLAR ATAXIA 29, CONGENITAL NONPROGRESSIVE, OMIM:117360	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
JAG1	ALAGILLE SYNDROME 279357	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KANSL1	CHROMOSOME 17Q21.31 MICRODELETION SYNDROME 610443	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KAT6B	BLEPHAROPHIMOSIS/INTELLECTUAL DISABILITY PHENOTYPE WHICH IS NOONAN-LIKE;GENITOPATELLAR SYNDROME 606170	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KBTBD13	NEMALINE MYOPATHY 6 609273	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNC3	SPINOCEREBELLAR ATAXIA TYPE 13 605259	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCND2	KCND2-related neurodevelopmental disorder with or without seizures	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCND3	KCND3-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNJ11	DIABETES MELLITUS, KCNJ11-RELATED TRANSIENT NEONATAL 261090;FAMILIAL HYPERINSULINISM 3272	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KCNJ6	KEPPEN-LUBINSKY SYNDROME 614098	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNJ8	Cantu syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNMA1	GENERALIZED EPILEPSY AND PAROXYSMAL DYSKINESIA 609446	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KCNN3	ZIMMERMANN-LABAND SYNDROME	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNQ2	EPILEPTIC ENCEPHALOPATHY EARLY INFANTILE TYPE 7 613720;BENIGN NEONATAL EPILEPSY TYPE 1 121200	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNQ3	KCNQ3 syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KDM6B	AUTOSOMAL RECESSIVE MENTAL RETARDATION;KDM6B-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KIF11	AUTOSOMAL-DOMINANT MICROCEPHALY ASSOCIATED WITH LYMPHEDEMA AND/OR CHORIORETINOPATHY 152950	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KIF1A	NEUROPATHY, HEREDITARY SENSORY, TYPE IIC, 614213;NESCAV SYNDROME, 614255	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KIF22	SPONDYLOEPIMETAPHYSEAL DYSPLASIA WITH JOINT LAXITY, TYPE 2 603546	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KIF5A	KIF5A-associated severe neonatal myoclonus, OMIM:617235	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KIF5B	KIF5B-related disease	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KIT	HUMAN PIEBALDISM 172800	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KLF1	ANEMIA, DYSERYTHROPOIETIC CONGENITAL, TYPE IV 613673	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KLF7	KLF7-related developmental disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KMT2C	INTELLECTUAL DISABILITY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KMT2D	KABUKI SYNDROME 147920	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KMT2E	INTELLECTUAL DISABILITY;Neurodevelopmental disorder and Epilepsy 618512	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KRAS	CARDIOFACIOCUTANEOUS SYNDROME 115150;NOONAN SYNDROME TYPE 3 609942	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KRIT1	CEREBRAL CAVERNOUS MALFORMATIONS TYPE 1, OMIM:116860	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KRT74	HYPOTRICHOSIS SIMPLEX OF THE SCALP 2 613981	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LEMD3	MELORHEOSTOSIS 155950;BUSCHKE-OLLENDORFF SYNDROME 166700	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LHX4	LHX4-RELATED COMBINED PITUITARY HORMONE DEFICIENCY 290135	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LMNA	EMERY-DREIFUSS MUSCULAR DYSTROPHY TYPE 2, OMIM:181350;FAMILIAL PARTIAL LIPODYSTROPHY TYPE 2, OMIM:151660;CHARCOT-MARIE-TOOTH DISEASE TYPE 2B1, OMIM:605588;HUTCHINSON-GILFORD PROGERIA SYNDROME, OMIM:176670	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LMNB1	LMNB1-associated developmental disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LMNB2	LMNB2-related Primary Microcephaly	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LMX1B	NAIL-PATELLA SYNDROME 161200	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LRP5	VITREORETINOPATHY EXUDATIVE TYPE 4 601813;OSTEOPOROSIS-PSEUDOGLIOMA SYNDROME 259770;HIGH BONE MASS TRAIT 601884;OSTEOPETROSIS AUTOSOMAL DOMINANT TYPE 1 607634;ENDOSTEAL HYPEROSTOSIS WORTH TYPE 144750	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MAF	Ayme-Gripp syndrome: CATARACT, DEAFNESS, INTELLECTUAL DISABILITY, SEIZURES, AND A DOWN SYNDROME-LIKE FACIES;CATARACT PULVERULENT JUVENILE-ONSET MAF-RELATED 610202;CATARACT CONGENITAL CERULEAN TYPE 4 610202	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MAFB	MULTICENTRIC CARPOTARSAL OSTEOLYSIS SYNDROME, OMIM:166300;Duane Syndrome, Aberrant Extraocular Muscle Innervation, and Inner-Ear Defects	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MAGEL2	Schaaf-Yang syndrome, OMIM:615547	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, maternally imprinted (paternal allele expressed)
MAP2K1	CARDIOFACIOCUTANEOUS SYNDROME 115150	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MAP2K2	CARDIOFACIOCUTANEOUS SYNDROME 115150	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MAP3K1	46XY SEX REVERSAL 6 613762	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MATN3	MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 5 607078	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MBD5	EHMT1-LIKE INTELLECTUAL DISABILITY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MED13	MED13 - Neurodevelopment disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MED13L	INTELLECTUAL DISABILITY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MEF2C	MENTAL RETARDATION-STEREOTYPIC MOVEMENTS-EPILEPSY AND/OR CEREBRAL MALFORMATIONS 613443	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MEIS2	MEIS2-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MFN2	MFN2-related developmental disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MIR17HG	FEINGOLD SYNDROME 614326	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MITF	TIETZ SYNDROME, OMIM:103500;WAARDENBURG SYNDROME TYPE 2A, OMIM:193510;Coloboma, Osteopetrosis, Microphthalmia, Macrocephaly, Albinism, and Deafness, OMIM:617306	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MMP13	METAPHYSEAL ANADYSPLASIA TYPE 1 602111;SPONDYLOEPIMETAPHYSEAL DYSPLASIA MISSOURI TYPE 602111	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MNX1	CURRARINO SYNDROME 176450	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MPZ	NEUROPATHY, CONGENITAL HYPOMYELINATING, 2, OMIM:618184	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MSX1	CLEFT LIP +/- CLEFT PALATE 608874	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MSX2	ENLARGED PARIETAL FORAMINA/CRANIUM BIFIDUM 168550;CRANIOSYNOSTOSIS, TYPE 2 604757	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYBPC1	LETHAL CONGENITAL CONTRACTURE SYNDROME 4, OMIM:614915;MYBPC1-related arthrogryposis and myopathy	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MYCN	FEINGOLD SYNDROME TYPE 1 164280	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYH3	DISTAL ARTHROGRYPOSIS TYPE 2A, OMIM:193700;Recessive Spondylocarpotarsal Synostosis Syndrome	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MYH8	DISTAL ARTHROGRYPOSIS TYPE, OMIM:158300;CARNEY COMPLEX VARIANT, OMIM:608837	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYH9	DEAFNESS AUTOSOMAL DOMINANT TYPE 17 603622;SEBASTIAN SYNDROME 155100;MACROTHROMBOCYTOPENIA WITH PROGRESSIVE SENSORINEURAL DEAFNESS 155100;EPSTEIN SYNDROME 155100;MAY-HEGGLIN ANOMALY 155100;FECHTNER SYNDROME 155100	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYT1L	MYT1L syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NBEA	NBEA Neurodevelopment disorder with seizures	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NEDD4L	Periventricular nodular heterotopia with ID, cleft palate and 2.3 toe syndactyly	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NF1	WATSON SYNDROME 193520;NEUROFIBROMATOSIS TYPE 1 162200;NEUROFIBROMATOSIS-NOONAN SYNDROME 601321;FAMILIAL SPINAL NEUROFIBROMATOSIS 162210	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NFE2L2	NFE2L2-related leukoencephalopathy, immune deficiency and hypohomocysteinaemia, OMIM:617744	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NFIA	Macrocephaly with intellectual disability	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NFIX	MARSHALL-SMITH SYNDROME 602535;SOTOS-LIKE SYNDROME 614753	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NIPBL	CORNELIA DE LANGE SYNDROME TYPE 1 122470	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NKX2-1	CHOREOATHETOSIS, HYPOTHYROIDISM, AND NEONATAL RESPIRATORY DISTRESS 610978;BENIGN HEREDITARY CHOREA 118700	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

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NKX2-5	ATRIAL SEPTAL DEFECT WITH ATRIOVENTRICULAR CONDUCTION DEFECTS 108900;TETRALOGY OF FALLOT 187500;CONGENITAL HYPOTHYROIDISM NON-GOITROUS TYPE 5 225250	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NODAL	HETEROTAXY SYNDROME 207574	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NOG	TARSAL-CARPAL COALITION SYNDROME 186570;MULTIPLE SYNOSTOSES SYNDROME TYPE 1 186500;STAPES ANKYLOSIS WITH BROAD THUMB AND TOES 184460;BRACHYDACTYLY TYPE B2 611377;SYMPHALANGISM PROXIMAL SYNDROME 185800	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NOTCH1	LEFT VENTRICULAR OUTFLOW TRACT OBSTRUCTION 109730;ADAMS OLIVER SYNDROME	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NOTCH2	HAJDU-CHENEY SYNDROME 102500	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NPM1	Dyskeratosis Congenita	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NR2F1	BOSCH-BOONSTRA OPTIC ATROPHY SYNDROME 615722	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NR4A2	NR4A2-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NRAS	NOONAN SYNDROME TYPE 6 613224	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NRXN1	PITT HOPKINS 2;AUTISM 209850	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
NRXN2	AUTISM 209850	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NSD1	WEAVER SYNDROME 277590;BECKWITH-WIEDEMANN SYNDROME 130650;SOTOS SYNDROME 117550	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NTRK2	Epilepsy and intellectual disability	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ODC1	ODC1-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
OTX2	MICROPHTHALMIA SYNDROMIC TYPE 5 610125	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PAFAH1B1	SUBCORTICAL BAND HETEROTOPIA 607432;LISSENCEPHALY TYPE 1 607432	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PAX2	RENAL-COLOBOMA SYNDROME 120330	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PAX3	WAARDENBURG SYNDROME, TYPE 1 193500;CRANIOFACIAL-DEAFNESS-HAND SYNDROME 122880	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PAX6	COLOBOMA OF OPTIC NERVE 120430;ANIRIDIA 106210;FOVEAL HYPOPLASIA 136520;BILATERAL OPTIC NERVE HYPOPLASIA 165550;KERATITIS HEREDITARY 148190;PETERS ANOMALY 604229;ANIRIDIA CEREBELLAR ATAXIA AND MENTAL DEFICIENCY 206700	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PAX8	CONGENITAL HYPOTHYROIDISM NON-GOITROUS TYPE 2 218700	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PAX9	TOOTH AGENESIS, SELECTIVE, 3 604625	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PDCD10	CEREBRAL CAVERNOUS MALFORMATIONS TYPE 3 603285	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PDE10A	Childhood-Onset Chorea with Bilateral Striatal Lesions	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PDE4D	ACRODYSOSTOSIS 101800	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PHIP	Developmental delay, ID, obesity and dysmorphic features	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PHOX2B	CENTRAL HYPOVENTILATION SYNDROME, CONGENITAL, WITH OR WITHOUT HIRSCHSPRUNG DISEASE 209880;NEUROBLASTOMA WITH HIRSCHSPRUNG DISEASE 613013	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PIEZO2	ARTHROGRYPOSIS, DISTAL, TYPE 3 114300;Ataxia, dysmetria, contractures & scoliosis with normal cognition but loss of discriminative touch perception	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PIK3CA	HEMIMEGALENCEPHALY PIK3CA;CLOVES: CONGENITAL LIPOMATOUS OVERGROWTH, VASCULAR MALFORMATIONS, AND EPIDERMAL NEVI, OMIM:612918;MEGALENCEPHALY-CAPILLARY MALFORMATION-POLYMICROGYRIA SYNDROME, SOMATIC 3, OMIM:602501	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PIK3R1	SHORT SYNDROME 269880;AGAMMAGLOBULINEMIA 7, AUTOSOMAL RECESSIVE 615214	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PIK3R2	MEGALENCEPHALY-POLYMICROGYRIA-POLYDACTYLY-HYDROCEPHALUS SYNDROME 1 603387	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PIP5K1C	LETHAL CONGENITAL CONTRACTURE SYNDROME TYPE 3 611369	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PITX1	CONGENITAL CLUBFOOT 119800;HOMEOTIC ARM-TO-LEG TRANSFORMATION ASSOCIATED WITH GENOMIC REARRANGEMENTS AT THE PITX1 LOCUS	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PITX2	AXENFELD-RIEGER SYNDROME TYPE 1 180500;PETERS ANOMALY 604229;RING DERMOID OF CORNEA 180550;IRIDOGONIODYSGENESIS TYPE 2 137600	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PITX3	CATARACT AUTOSOMAL DOMINANT 604219;CATARACT POSTERIOR POLAR TYPE 4 610623;ANTERIOR SEGMENT MESENCHYMAL DYSGENESIS 107250	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PLCB4	AURICULOCONDYLAR SYNDROME 602483	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
POGZ	INTELLECTUAL DISABILITY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
POLD1	SUBCUTANEOUS LIPODYSTROPHY, DEAFNESS, MANDIBULAR HYPOPLASIA AND MALE HYPOGONADISM	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
POLR1D	TREACHER COLLINS SYNDROME TYPE 2 613717	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
POLR3B	Autosomal recessive mental retardation;Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism;POLR3B-related neurodevelopmental disorder	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PPM1D	PPM1D syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PPP2R1A	INTELLECTUAL DISABILITY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PRKACA	PRKACA-related Multiple Congenital Malformation Syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PRKAR1A	ACRODYSOSTOSIS 101800	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PRPF8	PRPF8-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PRRT2	AUTOSOMAL RECESSIVE MENTAL RETARDATION;BENIGN FAMILIAL INFANTILE EPILEPSY AND INFANTILE CONVULSIONS WITH CHOREOATHETOSIS SYNDROME 602066	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PRRX1	AGNATHIA-OTOCEPHALY COMPLEX biallelic;AGNATHIA-OTOCEPHALY COMPLEX monoallelic	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PTCH1	BASAL CELL NEVUS SYNDROME 109400;HOLOPROSENCEPHALY-7 610828	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PTEN	BANNAYAN-ZONANA SYNDROME 153480;COWDEN DISEASE 158350;MACROCEPHALY/AUTISM SYNDROME 605309;VACTERL ASSOCIATION WITH HYDROCEPHALUS 276950;PROTEUS SYNDROME 176920;LHERMITTE-DUCLOS DISEASE 158350	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PTH1R	JANSEN METAPHYSEAL CHONDRODYSPLASIA 156400;EIKEN SKELETAL DYSPLASIA 600002;PRIMARY FAILURE OF TOOTH ERUPTION 125350;CHONDRODYSPLASIA BLOMSTRAND TYPE 215045	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PTHLH	BRACHYDACTYLY, TYPE E2 613382;CLUBBING WITH SKELETAL DYSPLASIA INC ACROOSTEOLYSIS	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PTPN11	LEOPARD SYNDROME TYPE 1 151100;NOONAN SYNDROME 1 163950	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RAC1	Developmental Disorders with Diverse Phenotypes	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RAD21	COHESINOPATHY 614701	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RAF1	NOONAN SYNDROME 5 611553	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RAI1	SMITH-MAGENIS SYNDROME 182290	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RET	RENAL AGENESIS 191830;MULTIPLE ENDOCRINE NEOPLASIA IIB 162300	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ROR2	Brachydactyly, type B1, OMIM:113000 (AD);Robinow syndrome, autosomal recessive, OMIM:268310 (AR)	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
RORA	INTELLECTUAL DISABILITY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RPL11	Diamond-Blackfan anemia with cleft palate and abnormal thumbs	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RPS19	RPS19-RELATED DIAMOND-BLACKFAN ANEMIA 220176	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RPS26	DIAMOND-BLACKFAN ANEMIA 10, OMIM:613309	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RUNX2	CLEIDOCRANIAL DYSPLASIA 119600	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SALL1	TOWNES-BROCKS SYNDROME 107480	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SALL4	ACRO-RENAL-OCULAR SYNDROME 607323;DUANE-RADIAL RAY SYNDROME 607323	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SAMD9	MIRAGE - myelodysplasia, infection, restriction of growth, adrenal hypoplasia, genital phenotypes, enteropathy	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SATB2	NONSPECIFIC SEVERE ID;CLEFT PALATE ISOLATED 119540;SYNDROMAL PIERRE ROBIN SEQUENCE	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCN1A	SCN1A-RELATED SEIZURE DISORDERS 607208	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCN1B	EPILEPSY, GENERALIZED, WITH FEBRILE SEIZURES PLUS, TYPE 1 604233;BRUGADA SYNDROME 5 612838	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCN2A	NONSPECIFIC SEVERE ID;BENIGN FAMILIAL NEONATAL INFANTILE SEIZURES 248968;INFANTILE EPILEPTIC ENCEPHALOPATHY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCN3A	Focal epilepsy	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCN4A	HYPOKALEMIC PERIODIC PARALYSIS 613345;PARAMYOTONIA CONGENITA OF VON EULENBURG 168300;HYPERKALEMIC PERIODIC PARALYSIS TYPE 1 170500	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCN8A	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 13, 614558	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SETBP1	DEVELOPMENTAL AND EXPRESSIVE LANGUAGE DELAY;SCHINZEL-GIEDION MIDFACE RETRACTION SYNDROME 269150	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SETD2	SETD2-associated Overgrowth Syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SF3B4	ACROFACIAL DYSOSTOSIS 1, NAGER TYPE 154400	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SHANK2	SUSCEPTIBILITY TO AUTISM TYPE 17 613436	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SHANK3	PHELAN-MCDERMID SYNDROME 606232	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

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SHH	MICROPHTHALMIA ISOLATED WITH COLOBOMA TYPE 5 611638;TRIPHALANGEAL THUMB-POLYSYNDACTYLY SYNDROME 174500;SOLITARY MEDIAN MAXILLARY CENTRAL INCISOR 147250;HOLOPROSENCEPHALY TYPE 3 236100	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SHOC2	NOONAN-LIKE SYNDROME WITH LOOSE ANAGEN HAIR 607721	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SHOX	LERI-WEILL DYSCHONDROSTEOSIS 127300;LANGER MESOMELIC DYSPLASIA 249700	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SHROOM3	NEURAL TUBE DEFECT	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SIM1	Severe obesity with neurobehavioral features	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SIX1	BRANCHIOOTIC SYNDROME TYPE 3, OMIM:608389;Non-syndromic craniosynostosis	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SIX3	HOLOPROSENCEPHALY 609637	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SIX5	BRANCHIOOTORENAL SYNDROME TYPE 2 610896	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SKI	SHPRINTZEN-GOLDBERG CRANIOSYNOSTOSIS SYNDROME 182212	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SLC1A2	EPILEPTIC ENCEPHALOPATHY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SLC25A4	Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number;Fontaine progeroid syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SLC2A1	GLUT1 DEFICIENCY SYNDROME TYPE 2 612126;GLUT1 DEFICIENCY SYNDROME TYPE 1 606777	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SLC37A4	SLC37A4-related congenital disorder of glycosylation with liver dysfunction;Glycogen storage disease Ib, OMIM:232220	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SLC4A1	RENAL TUBULAR ACIDOSIS, DISTAL, AD 179800;RENAL TUBULAR ACIDOSIS, DISTAL, AR 611590	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SLC6A1	EPILEPSY WITH MYOCLONIC-ATONIC SEIZURES	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMAD3	SMAD3-RELATED LOEYS-DIETZ SYNDROME 319643	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, OMIM:175050;MYHRE SYNDROME, OMIM:139210	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMARCA2	COFFIN SIRIS 135900;NICOLAIDES-BARAITSER SYNDROME 601358	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMARCA4	COFFIN SIRIS 135900;RHABDOID TUMOR PREDISPOSITION SYNDROME 2 613325	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMARCB1	EHMT1-like SYNDROME;RHABDOID PREDISPOSITION SYNDROME 1 609322	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMARCE1	COFFIN SIRIS 135900	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMC3	CORNELIA DE LANGE SYNDROME TYPE 3 610759	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SOS1	NOONAN SYNDROME 4 610733	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SOX10	WAARDENBURG SYNDROME TYPE 2E 611584;KALLMANN SYNDROME WITH DEAFNESS;WAARDENBURG SYNDROME TYPE 4C 613266;YEMENITE DEAF-BLIND HYPOPIGMENTATION SYNDROME 601706;PERIPHERAL DEMYELINATING NEUROPATHY, CENTRAL DYSMYELINATING LEUKODYSTROPHY, WAARDENBURG SYNDROME, AND HIRSCHSPRUNG DISEASE 609136	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SOX17	VESICOURETERAL REFLUX TYPE 3 613674	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SOX2	AEG SYNDROME;MICROPHTHALMIA SYNDROMIC TYPE 3 206900	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SOX5	12P12.5 INTRAGENIC DELETIONS ASSOCIATED WITH INTELLECTUAL DISABILITY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SOX6	SOX6-related neurodevelopmental syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SOX9	PIERRE ROBIN SEQUENCE;CAMPOMELIC DYSPLASIA 114290	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SPAST	SPAST-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SPECC1L	FACIAL CLEFTING, OBLIQUE, 1 600251	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SPRED1	LEGIUS SYNDROME 611431	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SPTAN1	EPILEPTIC ENCEPHALOPATHY EARLY INFANTILE TYPE 5 613477	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SPTBN2	SCA14;Infantile ataxia with oculomotor and pyramidal signs	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SPTLC2	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE IC, OMIM:613640	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SRCAP	FLOATING-HARBOR SYNDROME 136140	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
STX1A	STX1A-associated neurodevelopmental disorder with epilepsy;STX1A-associated neurodevelopmental disorder without epilepsy	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
STXBP1	EPILEPTIC ENCEPHALOPATHY EARLY INFANTILE TYPE 4 612164;ANGELMAN/PITT HOPKINS SYNDROME-LIKE DISORDER	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SUFU	Joubert Syndrome with Cranio-facial and Skeletal Defects;SUFU-related Joubert and congenital ocular motor apraxia	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SYNGAP1	MENTAL RETARDATION AUTOSOMAL DOMINANT TYPE 5 612621;EPILEPTIC ENCEPHALOPATHY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TAB2	CONGENITAL HEART DISEASE, NONSYNDROMIC, 2 612863	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBL1XR1	Pierpont syndrome;Intellectual disability with autism spectrum disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX1	22Q11.2 DELETION SYNDROME 188400	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX20	ATRIAL SEPTAL DEFECT TYPE 4 611363	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX3	ULNAR-MAMMARY SYNDROME 181450	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX4	SMALL PATELLA SYNDROME 147891	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX5	HOLT-ORAM SYNDROME 142900	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TCF4	PITT-HOPKINS SYNDROME 610954	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TCF7L2	TCF7L2-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TCOF1	TREACHER COLLINS SYNDROME TYPE 1 154500	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TEK	VENOUS MALFORMATIONS, MULTIPLE CUTANEOUS AND MUCOSAL 600195	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TERC	Dyskeratosis congenita, autosomal dominant 1	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TFAP2A	BRANCHIOOCULOFACIAL SYNDROME 113620	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TFAP2B	CHAR SYNDROME 169100	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGFB1	Camurati-Engelmann disease, OMIM:131300	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGFB2	LOEYS-DIETZ SYNDROME, TYPE 4, OMIM:614816	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGFB3	LOEYS-DIETZ SYNDROME 615582	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGFBR1	LOEYS-DIETZ SYNDROME TYPE 2A 608967;LOEYS-DIETZ SYNDROME TYPE 1A 609192;AORTIC ANEURYSM FAMILIAL THORACIC TYPE 5 609192	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGFBR2	LOEYS-DIETZ SYNDROME;TGFBR2-RELATED LOEYS-DIETZ SYNDROME 249163	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGIF1	HOLOPROSENCEPHALY 609637	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
THAP1	DYSTONIA 6, TORSION 602629	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
THRA	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 6 614450	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TINF2	EXUDATIVE RETINOPATHY WITH BONE MARROW FAILURE	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TP63	ECTODERMAL DYSPLASIA RAPP-HODGKIN TYPE 129400;NON-SYNDROMIC OROFACIAL CLEFT TYPE 8 129400;SPLIT-HAND/FOOT MALFORMATION TYPE 4 605289;ANKYLOBLEPHARON-ECTODERMAL DEFECTS-CLEFT LIP/PALATE 106260;ECTRODACTYLY-ECTODERMAL DYSPLASIA-CLEFT LIP/PALATE SYNDROME TYPE 3 604292;LIMB-MAMMARY SYNDROME 603543;ACRO-DERMATO-UNGUAL-LACRIMAL-TOOTH SYNDROME 103285	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TPM2	ARTHROGRYPOSIS, DISTAL, TYPE 1 108120	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TPM3	Nemaline/Cap myopathy	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TRIO	INTELLECTUAL DISABILITY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TRIP12	TRIP12-related intellectual disability with/without autism spectrum disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TRPM3	TRPM3-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TRPS1	TRICHO-RHINO-PHALANGEAL SYNDROME TYPE 1 190350	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TRPV3	OLMSTED SYNDROME 614594	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TRPV4	METATROPIC DYSPLASIA 156530;SPONDYLOMETAPHYSEAL DYSPLASIA, KOZLOWSKI TYPE 184252	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TRRAP	Autism and Syndromic Intellectual Disability	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TSC1	TUBEROUS SCLEROSIS TYPE 1 191100	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TSC2	TUBEROUS SCLEROSIS TYPE 2 613254;LYMPHANGIOLEIOMYOMATOSIS 606690	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TSHR	HYPERTHYROIDISM, FAMILIAL GESTATIONAL 603373;HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 1 275200	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
TUBA1A	LISSENCEPHALY TYPE 3 611603;INTELLECTUAL DISABILITY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TUBB2B	POLYMICROGYRIA ASYMMETRIC 610031	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TUBB3	CONGENITAL FIBROSIS OF THE EXTRAOCULAR MUSCLES 600638;CORTICAL DYSPLASIA, COMPLEX, WITH OTHER BRAIN MALFORMATIONS 1 614039	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TWIST1	CRANIOSYNOSTOSIS, TYPE 1 123100;SAETHRE-CHOTZEN SYNDROME 101400	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TWIST2	SETLEIS SYNDROME, OMIM:227260;ABLEPHARON MACROSTOMIA SYNDROME, OMIM:200110	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal

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UBE3A	ANGELMAN SYNDROME 105830	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)
USP7	USP7-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
WDFY3	Primary Microcephaly or macrocephaly with developmental delay	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
WFS1	Wolfram-like syndrome, autosomal dominant, OMIM:614296;WOLFRAM SYNDROME 1, OMIM:222300	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
WNT4	MULLERIAN APLASIA AND HYPERANDROGENISM 158330;SERKAL SYNDROME 611812	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
WNT5A	WNT5A-RELATED ROBINOW SYNDROME, AUTOSOMAL DOMINANT 180700	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
WT1	DENYS-DRASH SYNDROME 194080;FRASIER SYNDROME FRASIER SYNDROME FRASIER SYNDROME 136680	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
YAP1	COLOBOMA, OCULAR, WITH OR WITHOUT HEARING IMPAIRMENT, CLEFT LIP/PALATE, AND/OR MENTAL RETARDATION 120433	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
YWHAE	YWHAE-related developmental delay, seizures, hypotonia and brain abnormalities	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
YY1	INTELLECTUAL DISABILITY, OMIM:616579	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZBTB18	ZBTB18 syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZEB2	MOWAT-WILSON SYNDROME 235730	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZFHX3	ZFHX3-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZFHX4	ZFHX4-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZIC1	CRANIOSYNOSTOSIS 6 616602	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZIC2	HOLOPROSENCEPHALY 609637	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZMYND11	INTELLECTUAL DISABILITY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZNF750	SEBORRHEA-LIKE DERMATITIS WITH PSORIASIFORM ELEMENTS 610227	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ABCB6	MICROPHTHALMIA, ISOLATED, WITH COLOBOMA 7 614497	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ACVR2B	HETEROTAXY SYNDROME 207574	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ANK2	ANK2-related neurodevelopmental disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AXIN1	CAUDAL DUPLICATION ANOMALY, OMIM:607864	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CACNA1H	EPILEPSY, CHILDHOOD ABSENCE, SUSCEPTIBILITY TO, 6 611942	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CACNB4	JUVENILE MYOCLONIC EPILEPSY, OMIM:611136	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CD96	C SYNDROME 211750	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CDC42BPB	CDC42BPB-related Neurodevelopmental Disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CDH15	MENTAL RETARDATION AUTOSOMAL DOMINANT TYPE 3 612580	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHRM1	CHRM1-associated intellectual disability	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHRNA2	CHRNA2-RELATED NOCTURNAL FRONTAL LOBE EPILEPSY, AUTOSOMAL DOMINANT, OMIM:291607	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COMP	MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 1, OMIM:132400	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRKL	Bladder exstrophy plus	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRYAB	MYOPATHY, MYOFIBRILLAR, FATAL INFANTILE HYPERTONIC, ALPHA-B CRYSTALLIN-RELATED 613869;CATARACT POSTERIOR POLAR TYPE 2 613763	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CRYBA4	MICROPHTHALMIA ISOLATED WITH CATARACT TYPE 4, OMIM:610426	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRYGD	Cataract 2, multiple types, OMIM:115700	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CSDE1	CSDE1-associated intellectual disability and autism	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DDX58	SINGLETON-MERTEN SYNDROME 182250	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DHX16	Intellectual Disability, Central Nervous System anomalies and Seizures	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DIP2B	MENTAL RETARDATION, FRA12A TYPE 136630	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DISP1	HOLOPROSENCEPHALY, OMIM:609637	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DLG5	DLG5-associated developmental disorder (biallelic);DLG5-associated developmental disorder (monoallelic)	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
DLL1	INTELLECTUAL DISABILITY 616579	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DMPK	DYSTROPHIA MYOTONICA TYPE 1, OMIM:160900	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EMX2	FAMILIAL SCHIZENCEPHALY, EMX2-RELATED 269160	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EPB41L1	INTELLECTUAL DISABILITY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FBXW4	SPLIT-HAND/FOOT MALFORMATION TYPE 3 246560	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GABRG1	GABRG1-associated epileptic encephalopathy	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GDF3	MICROPHTHALMIA ISOLATED TYPE 7 613704;KLIPPEL-FEIL SYNDROME TYPE 3 613702;MICROPHTHALMIA ISOLATED WITH COLOBOMA TYPE 6 613703	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GNE	GNE-associated congenital myopathy;GNE-associated sialuria, OMIM:269921	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
HOXA11	Radioulnar Synostosis with Amegakaryocytic Thrombocytopenia	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ITGA6	EPIDERMOLYSIS BULLOSA WITH PYLORIC ATRESIA 226730	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
JMJD1C	JMJD1C-related neurodevelopmental disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KANK1	CEREBRAL PALSY SPASTIC QUADRIPLEGIC TYPE 2 612900	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNA1	KCNA1-related epileptic encephalopathy, biallelic;KCNA1-related epileptic encephalopathy, monoallelic	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KCNK9	BIRK-BAREL SYNDROME, OMIM:612292	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)
KIRREL3	MENTAL RETARDATION AUTOSOMAL DOMINANT TYPE 4 612581	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KITLG	WAARDENBURG SYNDROME TYPE 2	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LDB3	MYOPATHY MYOFIBRILLAR TYPE 4, OMIM:609452	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LEFTY2	HETEROTAXY SYNDROME 207574	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LRP6	Tooth Agenesis	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MAGI2	EARLY ONSET EPILEPTIC ENCEPHALOPATHY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MAPK10	EPILEPTIC ENCEPHALOPATHY LENNOX-GASTAUT TYPE 606369	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MIR184	EDICT SYNDROME 614303;KERATOCONUS WITH CATARACT	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYH6	CARDIOMYOPATHY DILATED TYPE 1EE 613252;CARDIOMYOPATHY FAMILIAL HYPERTROPHIC TYPE 14 613251;ATRIAL SEPTAL DEFECT TYPE 3 614089	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYOC	PRIMARY CONGENITAL GLAUCOMA TYPE 3A 231300	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYT1	OAVS/Goldenhar syndrome	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NCKAP1	NCKAP1-related Neurodevelopmental Disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NOTCH3	INFANTILE MYOFIBROMATOSIS 615293	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NR1I3	EHMT1-LIKE INTELLECTUAL DISABILITY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NRXN3	AUTISM 209850	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PLCG2	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, OMIM:614878;Familial cold autoinflammatory syndrome 3, OMIM:614468	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
QKI	INTELLECTUAL DISABILITY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RAD51	MIRROR MOVEMENTS 2 614508	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RANBP2	ACUTE NECROTIZING ENCEPHALOPATHY 1, SUSCEPTIBILITY TO 285648	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RASA1	PARKES WEBER SYNDROME 608355;CAPILLARY MALFORMATION-ARTERIOVENOUS MALFORMATION 608354	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RBFOX1	RBFOX1-related neurodevelopmental disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RNF135	MACROCEPHALY, MACROSOMIA, FACIAL DYSMORPHISM SYNDROME 614192	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RRM1	RRM1-related mitochondrial DNA depletion/deletions syndrome	DDG2P	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
RYR2	RYR2-related Catecholaminergic polymorphic ventricular tachycardia and intellectual disability	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RYR3	EPILEPTIC ENCEPHALOPATHY	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCRIB	8Q24.3 DELETION-LIKE	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SH3BP2	Cherubism, OMIM:118400	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SIX6	MICROPHTHALMIA, ISOLATED, WITH CATARACT 2 212550	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SLC9A9	SUSCEPTIBILITY TO AUTISM TYPE 16 613410	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMAD2	CONGENITAL HEART DISEASE	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMAD6	Non-syndromic craniosynostosis	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SNX3	MICROPHTHALMIA SYNDROMIC TYPE 8 601349	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

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SRGAP3	SLIT-ROBO RHO GTPASE-ACTIVATING PROTEIN 3 606525	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
STIM1	TUBULAR-AGGREGATE MYOPATHY 160565	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SUMO1	CLEFT LIP +/- CLEFT PALATE 608874	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SUPT16H	SUPT16H-related neurodevelopmental disorder	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TLL1	ATRIAL SEPTAL DEFECT TYPE 6 613087	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TMEM114	CONGENITAL AND JUVENILE CATARACT 611579	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TSHZ1	AURAL ATRESIA, CONGENITAL 607842	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
VANGL1	NEURAL TUBE DEFECTS 182940	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
VCP	VCP-related developmental disorder (monoallelic)	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
VIP	ASPERGER	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZEB1	CORNEAL DYSTROPHY FUCHS ENDOTHELIAL TYPE 6 613270;POSTERIOR POLYMORPHOUS CORNEAL DYSTROPHY TYPE 3 609141	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZFPM2	TETRALOGY OF FALLOT 187500;DIAPHRAGMATIC HERNIA 3 610187;46,XY GONADAL DYSGENESIS	DDG2P	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ABCC9	Hypertrichotic osteochondrodysplasia 23985 (Cantu syndrome);Hypertrichotic osteochondrodysplasia239850	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ACAN	Osteochondritis dissecans, short stature, and early-onset osteoarthritis 165800;Spondyloepimetaphyseal dysplasia, aggrecan type 61283;Spondyloepiphyseal dysplasia, Kimberley type 608361	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ACVR1	Fibrodysplasia ossificans progressiva 135100	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
AFF3	KINSSHIP syndrome, OMIM:619297	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ALPL	hypophosphatasia; skeletal dysplasias;skeletal dysplasias;Osteogenesis Imperfecta and Decreased Bone Density	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ALX4	Frontonasal dysplasia 2 613451	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ANKH	Chondrocalcinosis 2 118600;Craniometaphyseal dysplasia 123000	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ANKRD11	KBG syndrome 148050	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ANO5	Gnathodiaphyseal dysplasia OMIM:166260;gnathodiaphyseal dysplasia MONDO:0008151;Osteogenesis Imperfecta and Decreased Bone Density;skeletal dysplasias;skeletal dysplasias;Disproportionate Short Stature	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ARHGAP31	Adams-Oliver syndrome 1 100300	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ASXL1	Bohring-Opitz syndrome 605039	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
BMP2	short stature, facial dysmorphism and skeletal anomalies with or without cardiac aomalies 617877.;Brachydactyly, type A2 112600;{HFE hemochromatosis, modifier of} 235200	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
BMPR1B	Acromesomelic dysplasia, Demirhan type, OMIM:609441;Brachydactyly, type A1, D, OMIM:616849;Brachydactyly, type A2, OMIM:112600	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CASR	Hypocalcemia, autosomal dominant 601198;Hypocalciuric hypercalcemia, type I 145980;Hyperparathyroidism, neonatal 239200;Hypocalcemia, autosomal dominant, with Bartter syndrome 601198	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CBFB	Cleidocranial dysplasia 2, OMIM:620099;cleidocranial dysplasia 2, MONDO:0859307	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CDKN1C	IMAGE syndrome 614732	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)
CLCN7	Osteopetrosis, autosomal recessive 4, OMIM:611490;Osteopetrosis, autosomal dominant 2, OMIM:166600	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COG4	Saul-Wilson syndrome, OMIM:618150;microcephalic osteodysplastic dysplasia, Saul-Wilson type, MONDO:0019407	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL10A1	Metaphyseal chondrodysplasia, Schmid type 156500	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
COL11A1	Stickler syndrome, type II, OMIM:604841;Marshall syndrome, OMIM:154780;Fibrochondrogenesis 1, OMIM:228520	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL11A2	Stickler syndrome, type III 184840;Otospondylomegaepiphyseal dysplasia 215150;Fibrochondrogenesis 2 614524?;Fibrochondrogenesis 2 614524;Weissenbacher-Zweymuller syndrome 277610	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL1A1	Osteogenesis imperfecta, type I 166200;Caffey disease 114000;Osteogenesis imperfecta, type III 259420;Osteogenesis imperfecta, type II 166210;Ehlers-Danlos syndrome, type VIIA 130060;Ehlers-Danlos syndrome, classic 130000;Osteogenesis imperfecta, type IV 166220	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL1A2	Ehlers-Danlos syndrome, cardiac valvular form, OMIM:225320;Ehlers-Danlos syndrome, type VIIB, OMIM:130060;Osteogenesis imperfecta, type II, OMIM:166210;Osteogenesis imperfecta, type III, OMIM:259420;Osteogenesis imperfecta, type IV, OMIM:166220	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL2A1	Epiphyseal dysplasia, multiple, with myopia and deafness 132450;Spondyloepiphyseal dysplasia, Stanescu type 616583;Stickler sydrome, type I, nonsyndromic ocular 609508;Achondrogenesis, type II or hypochondrogenesis 200610;Kniest dysplasia 156550;Legg-Calve-Perthes disease 150600;Otospondylomegaepiphyseal dysplasia 215150;Stickler syndrome, type I 108300;SMED Strudwick type 184250;Spondyloperipheral dysplasia 271700;Platyspondylic skeletal dysplasia, Torrance type 151210;Czech dysplasia 609162;SED congenita 183900;Osteoarthritis with mild chondrodysplasia 604864;Avascular necrosis of the femoral head 608805	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL9A1	Stickler syndrome, type IV, OMIM:614134;Epiphyseal dysplasia, multiple, 6, OMIM:614135	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL9A2	Stickler syndrome, type V, OMIM:614284;Epiphyseal dysplasia, multiple, 2, OMIM:600204	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL9A3	Epiphyseal dysplasia, multiple, 3, with or without myopathy, OMIM:600969	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
COMP	Epiphyseal dysplasia, multiple, 1 132400;Pseudoachondroplasia 177170	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CREBBP	Rubinstein-Taybi syndrome 180849;Rubinstein-Taybi syndrome180849	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
DLX3	Amelogenesis imperfecta, type IV 104510;Trichodontoosseous syndrome 190320	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
DLX5	?Split-hand/foot malformation 1 with sensorineural hearing loss, OMIM:220600;Split-hand/foot malformation 1, OMIM:183600	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
DNMT3A	Tatton-Brown-Rahman syndrome 615879	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type 610536	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ENPP1	Hypophosphatemic rickets, autosomal recessive, 2 613312;Arterial calcification, generalized, of infancy, 1 208000;Cole disease 615522	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
EXT1	trichorhinophalangeal syndrome type 2 -150230;Exostoses, multiple, type 1133700;Exostoses, multiple, type 13370	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
EXT2	Exostoses, multiple, type 2, OMIM:133701	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
EZH2	Weaver syndrome	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FBN1	Geleophysic dysplasia 2 614185;Stiff skin syndrome 184900;Marfan syndrome 154700;Acromicric dysplasia 102370;Weill-Marchesani syndrome 2, dominant 608328	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FBN2	Contractural arachnodactyly, congenital OMIM:121050;congenital contractural arachnodactyly MONDO:0007363	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
FBXW11	Neurodevelopmental, jaw, eye, and digital syndrome, OMIM:618914;neurodevelopmental, jaw, eye, and digital syndrome, MONDO:0030057	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGF10	LADD syndrome 149730	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FGF23	Hypophosphatemic rickets, autosomal dominant 193100;Osteomalacia, tumor-induced;Tumoral calcinosis, hyperphosphatemic, familial 211900	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FGF9	Multiple synostoses syndrome 3, OMIM:612961;multiple synostoses syndrome 3, MONDO:0013064	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FGFR1	Hypogonadotropic hypogonadism 2 with or without anosmia 147950;Encephalocraniocutaneous lipomatosis, somatic mosaism 613001;Pfeiffer syndrome 101600;Trigonocephaly 1 190440;Hartsfield syndrome 615465;Jackson-Weiss syndrome 123150;Osteoglophonic dysplasia 166250	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FGFR2	Beare-Stevenson cutis gyrata syndrome 123790;Craniosynostosis, nonspecific Crouzon syndrome 123500;Craniofacial-skeletal-dermatologic dysplasia 101600;Pfeiffer syndrome 101600;Gastric cancer, somatic 613659;Jackson-Weiss syndrome 123150;LADD syndrome 149730;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis 207410;Apert syndrome 101200;Bent bone dysplasia syndrome 614592	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGFR3	Thanatophoric dysplasia, type I 187600;Muenke syndrome 602849;CATSHL syndrome 610474;SADDAN 616482;Thanatophoric dysplasia, type II 187601;Achondroplasia 100800;LADD syndrome 149730;Hypochondroplasia 146000;Crouzon syndrome with acanthosis nigricans 612247	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
FLNB	Spondylocarpotarsal synostosis syndrome 272460;Atelosteogenesis, type III 108721;Boomerang dysplasia 112310;Atelosteogenesis, type I 108720;Larsen syndrome 150250	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
FN1	Spondylometaphyseal dysplasia, corner fracture type 184255	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
GDF5	Brachydactyly, type C 113100;Acromesomelic dysplasia, Hunter-Thompson type 201250;Du Pan syndrome 228900;{Osteoarthritis-5} 612400;Chondrodysplasia, Grebe type 200700;Brachydactyly, type A2 112600;Brachydactyly, type A1, C 615072;Symphalangism, proximal, 1B 615298;Multiple synostoses syndrome 2 610017	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GDF6	Klippel-Feil syndrome 1, autosomal dominant 118100;Multiple synostoses syndrome type 4 - 617898.	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
GHR	Laron dwarfism, OMIM:262500;Growth hormone insensitivity, partial, OMIM:604271;Increased responsiveness to growth hormone, OMIM:604271	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GJA1	Craniometaphyseal dysplasia, autosomal recessive, OMIM:218400;Oculodentodigital dysplasia, OMIM:164200;Oculodentodigital dysplasia, autosomal recessive, OMIM:257850;Syndactyly, type III, OMIM:186100	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GLI3	Greig cephalopolysyndactyly syndrome, OMIM:175700;Pallister-Hall syndrome, OMIM:146510;Polydactyly, postaxial, types A1 and B, OMIM:174200;Polydactyly, preaxial, type IV, OMIM:174700	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted

PrenatalAutism - elenco dei geni e delle malattie genetiche ad insorgenza de novo investigate

GNAS	Pseudohypoparathyroidism Ia, OMIM:103580;pseudohypoparathyroidism type 1A, MONDO:0007078;Pseudohypoparathyroidism Ib, OMIM:603233;pseudohypoparathyroidism type 1B, MONDO:0011301;Pseudohypoparathyroidism Ic, OMIM:612462;pseudohypoparathyroidism type 1C, MONDO:0012911;McCune-Albright syndrome, somatic, mosaic, OMIM:174800;panostotic fibrous dysplasia, MONDO:0043168;Osseous heteroplasia, progressive, OMIM:166350;ACTH-independent macronodular adrenal hyperplasia. OMIM:219080;ACTH-independent macronodular adrenal hyperplasia 1, MONDO:0020735;Pseudopseudohypoparathyroidism, OMIM:612463;pseudopseudohypoparathyroidism, MONDO:0012912	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HOXA13	Hand-foot-genital syndrome 140000;Hand-foot-uterus syndrome 140000;Guttmacher syndrome 176305	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
HOXD13	Syndactyly, type V 186300;Brachydactyly-syndactyly syndrome 610713;Brachydactyly, type E 113300;Synpolydactyly 1 186000;Brachydactyly, type D 113200	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
HPGD	Craniooosteoarthropathy 259100;Digital clubbing, isolated congenital 119900;Hypertrophic osteoarthropathy, primary, autosomal recessive 1 259100	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
IDH1	Metaphyseal chondromatosis with D-2-hydroxyglutaric aciduria 614875;Maffucci syndrome 614569;Ollier disease/ Dyschondroplasia 166000	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
IFIH1	Singleton-Merten syndrome 1, OMIM:182250	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
IFITM5	Osteogenesis imperfecta, type V 610967	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
IHH	Acrocapitofemoral dysplasia, OMIM:607778;Brachydactyly, type A1, OMIM:112500	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KAT6B	Genitopatellar syndrome 606170;SBBYSS syndrome 603736;GTPTS,Ohdo	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KIF22	Spondyloepimetaphyseal dysplasia with joint laxity, type 2 603546	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KMT2D	Kabuki syndrome 1 - 147920	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LEMD3	Melorheostosis with osteopoikilosis 155950 IC;Osteopoikilosis 166700;Buschke-Ollendorff syndrome 166700	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
LMBR1	Laurin-Sandrow syndrome 135750;Polydactyly, preaxial type II 174500;Hypoplastic or aplastic tibia with polydactyly 188740;Triphalangeal thumb, type I 174500;Triphalangeal thumb-polysyndactyly syndrome 174500;Syndactyly, type IV 186200;Acheiropody 200500	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
LMNA	Emery-Dreifuss muscular dystrophy 2, 181350;Heart-hand syndrome, Slovenian type 610140;Charcot-Marie-Tooth disease, type 2B1 605588;Emery-Dreifuss muscular dystrophy 3, 616516;Cardiomyopathy, dilated, 1A 115200;Hutchinson-Gilford progeria 176670;Mandibuloacral dysplasia 248370;Lipodystrophy, familial partial, 2 151660;Restrictive dermopathy, lethal 275210;Foundation Trust)Mandibuloacral dysplasia 248370;616516;Muscular dystrophy, congenital 613205;Muscular dystrophy, limb-girdle, type 1B 159001;Malouf syndrome 212112	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
LMX1B	Nail-patella syndrome161200;Nail-patella syndrome 161200	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
LRP4	Sclerosteosis 2 614305;Cenani-Lenz syndactyly syndrome 212780	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
LRP5	Exudative vitreoretinopathy 4 601813;[Bone mineral density variability 1] 601884;Osteopetrosis, autosomal dominant 1 607634;Osteosclerosis 144750;van Buchem disease, type 2 607636;Osteoporosis-pseudoglioma syndrome 259770;Hyperostosis, endosteal 144750;{Osteoporosis} 166710	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
LTBP3	Geleophysic dysplasia 3, OMIM:617809;Dental anomalies and short stature, OMIM:610216;geleophysic dysplasia 3, MONDO:0054722,	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MAFB	Multicentric carpotarsal osteolysis syndrome 166300	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MATN3	MED;multiple epiphyseal dysplasia;Multiple Epiphyseal Dysplasia, Dominant;Disproportionate Short Stature;Epiphyseal dysplasia, multiple, 5, 607078; {Osteoarthritis susceptibility 2}, 140600;Spondyloepimetaphyseal dysplasia, 608728; Spondyloepimetaphyseal dysplasia, 608728	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MGP	Keutel syndrome, OMIM:245150;spondyloepiphyseal dysplasia, MONDO:0016761	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MMP13	Metaphyseal anadysplasia 1 602111;Spondyloepimetaphyseal dysplasia, Missouri type 602111;Metaphyseal dysplasia, Spahr type - 250400	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MNX1	Currarino syndrome 176450	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MSX2	Craniosynostosis, type 2 604757;Parietal foramina with cleidocranial dysplasia 168550;Parietal foramina 1 168500	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MYCN	Feingold syndrome (Microcephaly-oculo-digito-esophageal-duodenal) 164280	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MYH3	Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, OMIM:178110;Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, OMIM:618469;contractures, pterygia, and spondylocarpotarsal fusion syndrome 1A, MONDO:0008338	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
NF1	Neurofibromatosis, type 1 162200;Neurofibromatosis, type 1162200;Neurofibromatosis, familial spinal 162210;Neurofibromatosis-Noonan syndrome601321;Neurofibromatosis, familial spinal162210;Neurofibromatosis-Noonan syndrome 601321	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NFIX	Marshall-Smith syndrome 602535;Sotos syndrome 2 614753	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NIPBL	Cornelia de Lange syndrome 1 122470	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NLRP3	CINCA syndrome, OMIM:607115	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NOG	Symphalangism, proximal, 1A 185800;Brachydactyly, type B2 611377;Tarsal-carpal coalition syndrome 186570;Stapes ankylosis with broad thumb and toes 184460;Multiple synostoses syndrome 1 186500	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NOTCH1	Limb, scalp and skull defects;AOS;Combination of aplasia cutis congenita of the scalp vertex and terminal transverse limb defects (e.g., amputations, syndactyly, brachydactyly, or oligodactyly);Adams-Oliver syndrome 5, 616028	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NOTCH2	Hajdu-Cheney (Serpentine fibula polycystic kidney) syndrome 102500;Alagille syndrome 2 610205	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NPR2	Acromesomelic dysplasia, Maroteaux type 602875;Short stature with nonspecific skeletal abnormalities 616255;Epiphyseal chondrodysplasia, Miura type 615923	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
NSD1	Sotos syndrome 1 117550	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PAX3	Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 1, 193500;Waardenburg syndrome, type 3, 148820	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PDE4D	Acrodysostosis 2, with or without hormone resistance 614613;Acrodysostosis 2, with or without hormone resistance614613	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PIK3R1	SHORT syndrome 269880	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PITX1	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly 119800;Liebenberg syndrome 186550	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
POLR1D	Treacher Collins syndrome 2 613717	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PRKAR1A	Acrodysostosis 1, with or without hormone resistance, OMIM:101800	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PTH1R	Failure of tooth eruption, primary 125350;Eiken syndrome 600002;Metaphyseal chondrodysplasia, Murk Jansen type 156400;Chondrodysplasia, Blomstrand type 215045	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PTHLH	Brachydactyly, type E2 613382;Brachydactyly, type E2613382	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PTPN11	Metachondromatosis 156250;LEOPARD syndrome 1 151100;Noonan syndrome 1 163950;LEOPARD syndrome 1151100	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ROR2	Brachydactyly, type B1, OMIM:113000 (AD);Robinow syndrome, autosomal recessive, OMIM:268310 (AR)	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
RUNX2	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly 156510;Cleidocranial dysplasia, forme fruste, with brachydactyly 119600;Cleidocranial dysplasia, forme fruste, dental anomalies only 119600;Cleidocranial dysplasia 119600	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SALL1	Townes Brocks syndrome (Renal-Ear-Anal-Radial syndrome) 107480	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SALL4	Okihiro (Duane-radial ray) syndrome 607323;IVIC syndrome 147750	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SETD2	Luscan-Lumish syndrome616831;Luscan-Lumish syndrome 616831	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SF3B4	Acrofacial dysostosis 1, Nager type 154400	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SH3BP2	Cherubism, OMIM:118400	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SHOX	Langer mesomelic dysplasia, OMIM:249700 (PR);Leri-Weill dyschondrosteosis, OMIM:127300 (PD);Short stature, idiopathic familial, OMIM:300582;Dorsolateral bowed, short radii;Bowing and curving of radius;Radioulnar shortening	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SKI	Shprintzen-Goldberg syndrome 182212	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SLC34A1	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMAD3	Loeys-Dietz syndrome 3 613795	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SMAD4	Myhre syndrome139210;Myhre syndrome 139210	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SMAD6	{Radioulnar synostosis, nonsyndromic}, OMIM:179300	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SMC3	Cornelia de Lange syndrome 3 610759	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SOST	Craniodiaphyseal dysplasia, autosomal dominant 122860;Van Buchem disease 239100;Sclerosteosis 1 269500	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SOX9	Campomelic dysplasia with autosomal sex reversal 114290;Acampomelic campomelic dysplasia 114290;Campomelic dysplasia 114290	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TBX3	Ulnar-mammary syndrome 181450	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TBX4	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, OMIM:147891	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TBX5	Holt-Oram syndrome 142900	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TBX6	Spondylocostal dysostosis 5122600;Spondylocostal dysostosis 5 122600	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TCOF1	Treacher Collins syndrome 1 154500	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TERT	Dyskeratosis congenita, autosomal dominant 2, OMIM:613989;Dyskeratosis congenita, autosomal recessive 4, OMIM:613989	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
TGFB1	Camurati-Engelmann disease, OMIM:131300	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TGFB2	Loeys-Dietz syndrome 4 614816	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TGFBR2	Loeys-Dietz syndrome 2 610168	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted

PrenatalAutism - elenco dei geni e delle malattie genetiche ad insorgenza de novo investigate

TNFRSF11A	Osteolysis, familial expansile 174810;Osteopetrosis, autosomal recessive 7 612301;Paget disease of bone 2, early-onset 602080	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
TP63	ADULT syndrome, OMIM:103285;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, OMIM:604292;Limb-mammary syndrome, OMIM:603543;Split-hand/foot malformation 4, OMIM:605289	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TRPS1	Trichorhinophalangeal syndrome, type III 190351;Trichorhinophalangeal syndrome, type I 190350	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TRPV4	Brachyolmia type 3 113500;Hereditary motor and sensory neuropathy, type IIc 606071;Digital arthropathy-brachydactyly, familial 606835;SED, Maroteaux type 184095;Parastremmatic dwarfism 168400;Metatropic dysplasia 156530;Scapuloperoneal spinal muscular atrophy 181405;Spinal muscular atrophy, distal, congenital nonprogressive 600175;Spondylometaphyseal dysplasia, Kozlowski type 184252	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TWIST1	Robinow-Sorauf syndrome 180750;Saethre-Chotzen syndrome 101400;Craniosynostosis, type 1 123100;Saethre-Chotzen syndrome with eyelid anomalies 101400	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
WNT5A	Robinow syndrome, autosomal dominant 1 180700	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
YY1	Gabriele-de Vries syndrome617557;Gabriele-de Vries syndrome 617557	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
DROSHA	Global developmental delay;Intellectual disability;Seizures;Cerebral white matter atrophy;Abnormality of the corpus callosum;Abnormality of movement;Stereotypic behavior;Abnormality of head or neck;Short foot	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FBLN1	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses 608180	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
HDAC4	Albright hereditary osteodystrophy type 3;Brachydactyly-intellectual disability;Albright hereditary osteodystrophy type 3, Albright hereditary osteodystrophy-like syndrome, Brachydactyly-intellectual disability, Del(2)(q37) 600430;Albright hereditary osteodystrophy-like syndrome;Del(2)(q37) 600430	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KIF5B	kyphomelic dysplasia, MONDO:0008881	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MIR17HG	FS2;Brachydactyly with short stature and microcephaly;Microcephaly-oculo-digito-esophageal-duodenal syndrome;Feingold syndrome 2, 614326	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PFN1	Paget s disease of bone;bone Paget disease MONDO:0005382	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PLEKHM1	Osteopetrosis, autosomal recessive 6 - 611497;Osteopetrosis, autosomal recessive 6 611497;Osteopetrosis, autosomal dominant 3 - 618107	Skeletal dysplasia	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
RAD21	Cornelia de Lange syndrome 4, OMIM:614701	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
DSPP	Dentinogenesis imperfecta, Shields type II, 125490; Deafness, autosomal dominant 36, with dentinogenesis, 605594; Dentinogenesis imperfecta, Shields type III, 125500; Dentin dysplasia, type II, 125420 -3	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FOXC1	Anterior segment dysgenesis 3, multiple subtypes, MIM: 601631; Axenfeld-Rieger syndrome, type 3, MIM: 602482	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
GDF3	Klippel-Feil anomaly with laryngeal malformation - 613702	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
HOXA11	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1 605432	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MTAP	Diaphyseal medullary stenosis with malignant fibrous histiocytoma 112250	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SHH	Preaxial polydactyly type 1 (PPD1)	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SMARCE1	Coffin-Siris syndrome 5, OMIM:616938	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SULF1	Mesomelia-synostoses syndrome600383;Mesomelia-synostoses syndrome 600383	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
THPO	Thrombocythemia 1 187950 (rare presentation with congenital limb defects)	Skeletal dysplasia	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ABCC9	CANTU SYNDROME HYPERTRICHOTIC OSTEOCHONDRODYSPLASIA	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ACAN	SPONDYLOEPIPHYSEAL DYSPLASIA TYPE KIMBERLEY;SPONDYLOEPIMETAPHYSEAL DYSPLASIA AGGRECAN TYPE	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ACTA1	Nemaline myopathy 3, autosomal dominant or recessive, OMIM:161800	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ACTA2	AORTIC ANEURYSM, FAMILIAL THORACIC 6;MOYAMOYA DISEASE 5	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ACTB	BARAITSER-WINTER SYNDROME;ACTB Haploinsufficiency syndtome	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ACTC1	Atrial septal defect 5 612794	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ACTG1	BARAITSER-WINTER SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ACVR2B	Heterotaxy;Dextrocardia;Double outlet right ventricle;Transposition of the great arteries;Gut malrotation;polysplenia;right-sided spleen;asplenia	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ACVRL1	Telangiectasia, hereditary hemorrhagic, type 2, OMIM:600376	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ADAR	Aicardi-Goutieres syndrome 6, OMIM:615010;Dyschromatosis symmetrica hereditaria, OMIM:127400	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ADNP	MENTAL RETARDATION, AUTOSOMAL DOMINANT, 28	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AFF3	KINSSHIP syndrome, OMIM:619297	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
AKT1	PROTEUS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AKT2	Hypoinsulinemic hypoglycemia with hemihypertrophy, OMIM:240900;Hypoinsulinemic hypoglycemia and body hemihypertrophy, MONDO:0009416	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, OMIM:615937	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ALDH18A1	CUTIS LAXA, AUTOSOMAL DOMINANT 3;SPASTIC PARAPLEGIA 9, AUTOSOMAL DOMINANT;MENTAL RETARDATION-JOINT HYPERMOBILITY-SKIN LAXITY WITH OR WITHOUT METABOLIC ABNORMALITIES	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ALPL	HYPOPHOSPHATASIA	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ALX4	FRONTONASAL DYSPLASIA 2;PARIETAL FORAMINA 2	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ANKH	CHONDROCALCINOSIS 2;CRANIOMETAPHYSEAL DYSPLASIA JACKSON TYPE	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ANKRD11	KBG SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ARHGAP31	ADAMS-OLIVER SYNDROME 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ARID1A	COFFIN-SIRIS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ARID1B	MENTAL RETARDATION, AUTOSOMAL DOMINANT 12;COFFIN SIRIS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ASXL1	BOHRING-OPITZ SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ATN1	Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, OMIM:618494	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
BCL11A	INTELLECTUAL DISABILITY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BFSP2	CATARACT AUTOSOMAL DOMINANT BFSP2-RELATED	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BMP2	Short stature, palatal anomalies, congenital heart disease, and skeletal malformations;Brachydactyly, type A2 112600	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BMP4	MICROPHTHALMIA, SYNDROMIC 6;OROFACIAL CLEFT 11	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BNC2	Lower urinary tract obstruction, congenital, 618612	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BRAF	CARDIOFACIOCUTANEOUS SYNDROME;LEOPARD SYNDROME TYPE 3;NOONAN SYNDROME TYPE 7	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CACNA1C	Timothy syndrome, OMIM:601005;Timothy syndrome, MONDO:0010979;Long QT syndrome 8, OMIM:618447;long qt syndrome 8, MONDO:0032756;Brugada syndrome 3, OMIM:611875;Brugada syndrome 3, MONDO:0012742;CACNA1C-related disorder	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CACNA1E	Developmental and Epileptic Encephalopathy with Contractures Macrocephaly and Dyskinesias;Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesia	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CACNA1G	Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CASR	Hypocalcemia, autosomal dominant, 601198;Hypocalciuric hypercalcemia, type I, 145980;Hyperparathyroidism, neonatal, 239200;Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CBL	NOONAN SYNDROME-LIKE DISORDER WITH OR WITHOUT JUVENILE MEYLOMONOCYTIC LEUKEMIA	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CDH1	Blepharo-cheiro-dontic syndrome	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CDKN1C	BECKWITH-WIEDEMANN SYNDROME;IMAGe Syndrome	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)
CDON	HOLOPROSENCEPHALY 11	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CEP85L	Lissencephaly 10, posterior predominant, OMIM:618873	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CFC1	CFC1-RELATED CONOTRUNCAL HEART MALFORMATIONS;Heterotaxy, visceral, 2, autosomal, 605376	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHD7	KALLMANN SYNDROME TYPE 5;IDIOPATHIC HYPOGONADOTROPIC HYPOGONADISM;CHARGE SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHRNB1	Myasthenic syndrome, congenital, 2A, slow-channel, 616313;?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CHRNE	Myasthenic syndrome, congenital, 4A, slow-channel, OMIM:605809;Myasthenic syndrome, congenital, 4B, fast-channel, OMIM:616324;Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, OMIM:608931	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CLCN7	CLCN7-RELATED OSTEOPETROSIS;Osteopetrosis, autosomal recessive 4, OMIM:611490;Osteopetrosis, autosomal dominant 2, OMIM:166600;Hypopigmentation, organomegaly, and delayed myelination and development, OMIM:618541	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CNOT3	CNOT3 syndrome;Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL10A1	SCHMID TYPE METAPHYSEAL CHONDRODYSPLASIA	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL11A1	FIBROCHONDROGENESIS;STICKLER SYNDROME, TYPE II	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL11A2	WEISSENBACHER-ZWEYMUELLER SYNDROME;DEAFNESS AUTOSOMAL RECESSIVE TYPE 53;AUTOSOMAL RECESSIVE OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA;DEAFNESS AUTOSOMAL DOMINANT TYPE 13;STICKLER SYNDROME TYPE 3	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal

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COL12A1	Bethlem myopathy 2, 616471;?Ullrich congenital muscular dystrophy 2, 616470	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL1A1	COL1A1/2-RELATED OSTEOGENESIS IMPERFECTA;EHLERS-DANLOS SYNDROME, CLASSIC TYPE, COL1A1-RELATED;OSTEOGENESIS IMPERFECTA TYPE III;EHLERS-DANLOS SYNDROME TYPE VIIA;OSTEOGENESIS IMPERFECTA TYPE IIA;CAFFEY DISEASE;OSTEOGENESIS IMPERFECTA TYPE I	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL1A2	Ehlers-Danlos syndrome;Osteogenesis imperfecta	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL2A1	SPONDYLOEPIMETAPHYSEAL DYSPLASIA STRUDWICK TYPE;SPONDYLOPERIPHERAL DYSPLASIA;STICKLER SYNDROME TYPE 1 NON-SYNDROMIC OCULAR;PLATYSPONDYLIC LETHAL SKELETAL DYSPLASIA TORRANCE TYPE;ACHONDROGENESIS TYPE 2;RHEGMATOGENOUS RETINAL DETACHMENT AUTOSOMAL DOMINANT;SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA;KNIEST DYSPLASIA	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL3A1	HP:0002126;HP:0001883;HP:0006496	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL4A1	PORENCEPHALY 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL4A2	PORENCEPHALY 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL6A1	Bethlem myopathy 1, OMIM:158810;Ullrich congenital muscular dystrophy 1, OMIM:254090	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL6A2	Ullrich congenital muscular dystrophy 1 254090;Bethlem myopathy 1 158810	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL6A3	Bethlem myopathy, OMIM:158810;Ullrich congenital muscular dystrophy, OMIM:254090	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL9A1	STICKLER SYNDROME TYPE 4;MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 6	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL9A2	MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 2;STICKLER SYNDROME, TYPE V	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CREBBP	CREBBP intellectual disability without typical RTS features;RUBINSTEIN-TAYBI SYNDROME TYPE 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRYAA	CATARACT, AUTOSOMAL RECESSIVE CONGENITAL 1;CATARACT, NUCLEAR	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CRYBA1	CATARACT CONGENITAL ZONULAR WITH SUTURAL OPACITIES	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRYBA4	CATARACT ZONULAR TYPE 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRYBB1	CATARACT 17, MULTIPLE TYPES;CATARACT 17, MULTIPLE TYPES, MONOALLELIC;CATARACT, CONGENITAL NUCLEAR, AUTOSOMAL RECESSIVE 3	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CRYBB2	CATARACT, COPPOCK-LIKE;CATARACT, CONGENITAL, CERULEAN TYPE, 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRYBB3	Cataract 22, OMIM:609741	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
CRYGC	CATARACT AUTOSOMAL DOMINANT	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRYGD	CATARACT AUTOSOMAL DOMINANT;CATARACT CONGENITAL CERULEAN TYPE 3	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CTNNB1	MENTAL RETARDATION, AUTOSOMAL DOMINANT 19	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CTNND1	Blepharo-cheiro-dontic syndrome	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CYP11A1	Adrenal insufficiency, congenital, wth 46XY sex reversal, partial or complete, OMIM:613743	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
DCC	Mirror movements 1 and/or agenesis of the corpus callosum, OMIM:157600;Gaze palsy, familial horizontal, with progressive scoliosis, 2, OMIM:617542	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
DISP1	Holoprosencephaly	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DLL1	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, OMIM:618709	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
DLX5	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600;Split-hand/foot malformation 1, 183600	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
DNM1L	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
DNM2	Lethal congenital contracture syndrome 5, 615368	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
DNMT3A	OVERGROWTH SYNDROME WITH INTELLECTUAL DISABILITY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DSP	Epidermolysis bullosa, lethal acantholytic, OMIM:609638 (AR);Skin fragility-woolly hair syndrome, OMIM:607655 (AR);Keratosis palmoplantaris striata II, OMIM:612908 (AD);Cardiomyopathy, dilated, with woolly hair and keratoderma, OMIM:605676 (AR);Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, OMIM:615821 (AD);Arrhythmogenic right ventricular dysplasia 8, OMIM:607450 (AD)	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
DYNC1H1	SEVERE ID WITH NEURONAL MIGRATION DISORDER;SPINAL MUSCULAR ATROPHY, LOWER EXTREMITY-PREDOMINANT, AD	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DYRK1A	MENTAL RETARDATION AUTOSOMAL DOMINANT TYPE 7	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EDNRA	MANDIBULOFACIAL DYSOSTOSIS WITH ALOPECIA	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EFTUD2	MANDIBULOFACIAL DYSOSTOSIS WITH MICROCEPHALY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EHMT1	9Q SUBTELOMERIC DELETION SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ELN	ELN-RELATED CUTIS LAXA;SUPRAVALVAR AORTIC STENOSIS	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EMX2	Schizencephaly, 269160	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EP300	RUBINSTEIN-TAYBI SYNDROME TYPE 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EXT1	HEREDITARY MULTIPLE EXOSTOSES TYPE 1;TRICHO-RHINO-PHALANGEAL SYNDROME TYPE 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EXT2	EXOSTOSES, MULTIPLE, TYPE 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EYA1	BRANCHIOOTORENAL SYNDROME TYPE 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EZH2	WEAVER SYNDROME 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FBN1	SHPRINTZEN-GOLDBERG CRANIOSYNOSTOSIS SYNDROME;MARFAN SYNDROME;MASS SYNDROME/OVERLAP CONNECTIVE TISSUE DISEASE	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FBN2	Contractural arachnodactyly, congenital OMIM:121050;congenital contractural arachnodactyly MONDO:0007363	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
FGF10	LADD SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGF8	Hypogonadotropic hypogonadism 6 with or without anosmia 612702	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGFR1	IDIOPATHIC HYPOGONADOTROPIC HYPOGONADISM;OSTEOGLOPHONIC DYSPLASIA;PFEIFFER SYNDROME;KALLMANN SYNDROME TYPE 2;Hartsfield syndrome;Encephalocraniocutaneous lipomatosis	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGFR2	BEARE-STEVENSON CUTIS GYRATA SYNDROME;LACRIMO-AURICULO-DENTO-DIGITAL SYNDROME;JACKSON-WEISS SYNDROME;ACROCEPHALOSYNDACTYLY TYPE V;FAMILIAL SCAPHOCEPHALY SYNDROME;ANTLEY-BIXLER SYNDROME;CROUZON SYNDROME;APERT SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGFR3	LACRIMO-AURICULO-DENTO-DIGITAL SYNDROME;MUENKE SYNDROME;ACHONDROPLASIA;CROUZON SYNDROME WITH ACANTHOSIS NIGRICANS;CAMPTODACTYLY TALL STATURE AND HEARING LOSS SYNDROME;HYPOCHONDROPLASIA;THANATOPHORIC DYSPLASIA TYPE 2;THANATOPHORIC DYSPLASIA TYPE 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FLNB	ATELOSTEOGENESIS TYPE 1;BOOMERANG DYSPLASIA;SPONDYLOCARPOTARSAL SYNOSTOSIS SYNDROME;AUTOSOMAL DOMINANT LARSEN SYNDROME;ATELOSTEOGENESIS TYPE 3	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
FLNC	Arthrogryposis, MONDO:0008779	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FLT4	MILROY DISEASE	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXC1	IRIDOGONIODYSGENESIS ANOMALY;AXENFELD-RIEGER SYNDROME TYPE 3;PETERS ANOMALY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXC2	LYMPHEDEMA-DISTICHIASIS SYNDROME;HEREDITARY LYMPHEDEMA II	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXE3	Anterior segment dysgenesis 2, multiple subtypes, OMIM:610256;Cataract 34, multiple types, OMIM:612968;{Aortic aneurysm, familial thoracic 11, susceptibility to}, OMIM:617349 CONGENITAL PRIMARY APHAKIA;ANTERIOR SEGMENT MESENCHYMAL DYSGENESIS	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
FOXF1	ALVEOLAR CAPILLARY DYSPLASIA WITH MISALIGNMENT OF PULMONARY VEINS	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXG1	Rett Syndrome, congenital variant OMIM:613454;Rett syndrome, congenital variant MONDO:0013270	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FTL	HEREDITARY HYPERFERRITINEMIA-CATARACT SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GATA2	EMBERGER SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GATA3	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, OMIM:146255;Hypoparathyroidism-deafness-renal disease syndrome, MONDO:0007797	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GATA4	ATRIAL SEPTAL DEFECT TYPE 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GATA6	ATRIAL SEPTAL DEFECT 9;PANCREATIC AGENESIS, DIAPHRAGMATIC HERNIA AND CONGENITAL HEART DEFECTS;ATRIOVENTRICULAR SEPTAL DEFECT 5	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GDF1	Congenital heart defects, multiple types, 6, OMIM:613854;Right atrial isomerism (Ivemark), OMIM:208530	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GDF5	BRACHYDACTYLY TYPE C;SYMPHALANGISM PROXIMAL SYNDROME;DU PAN SYNDROME;BRACHYDACTYLY TYPE A1;ACROMESOMELIC CHONDRODYSPLASIA HUNTER-THOMPSON TYPE;ACROMESOMELIC CHONDRODYSPLASIA GREBE TYPE;BRACHYDACTYLY TYPE A2;MULTIPLE SYNOSTOSES SYNDROME TYPE 2	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GDF6	MICROPHTHALMIA ISOLATED TYPE 4;KLIPPEL-FEIL SYNDROME TYPE 1;Syndromic CAKUT	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GFAP	ALEXANDER DISEASE	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GJA1	AUTOSOMAL DOMINANT OCULODENTODIGITAL DYSPLASIA;HYPOPLASTIC LEFT HEART SYNDROME;AUTOSOMAL RECESSIVE OCULODENTODIGITAL DYSPLASIA;HALLERMANN-STREIFF SYNDROME	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GJA3	CATARACT ZONULAR PULVERULENT CATARACT TYPE 3	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GJA8	CATARACT-MICROCORNEA SYNDROME;CATARACT ZONULAR PULVERULENT TYPE 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GJC2	LYMPHEDEMA, HEREDITARY, IC;LEUKODYSTROPHY, HYPOMYELINATING, 2;SPASTIC PARAPLEGIA, 44	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GLI2	GLI2-RELATED HOLOPROSENCEPHALY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

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GLI3	Greig cephalopolysyndactyly syndrome, OMIM:175700;Pallister-Hall syndrome, OMIM:146510;Polydactyly, postaxial, types A1 and B, OMIM:174200;Polydactyly, preaxial, type IV, OMIM:174700	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GNAS	Pseudohypoparathyroidism Ia, OMIM:103580;pseudohypoparathyroidism type 1A, MONDO:0007078;Pseudohypoparathyroidism Ib, OMIM:603233;pseudohypoparathyroidism type 1B, MONDO:0011301;Pseudohypoparathyroidism Ic, OMIM:612462;pseudohypoparathyroidism type 1C, MONDO:0012911;McCune-Albright syndrome, somatic, mosaic, OMIM:174800;panostotic fibrous dysplasia, MONDO:0043168;Osseous heteroplasia, progressive, OMIM:166350;progressive osseous heteroplasia, MONDO:0008153;ACTH-independent macronodular adrenal hyperplasia. OMIM:219080;ACTH-independent macronodular adrenal hyperplasia 1, MONDO:0020735;Pseudopseudohypoparathyroidism, OMIM:612463;pseudopseudohypoparathyroidism, MONDO:0012912	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant OMIM:614254;intellectual disability, autosomal dominant 8 MONDO:0013655;Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive OMIM:617820;neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive MONDO:0060629	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GRIN2B	Intellectual developmental disorder, autosomal dominant 6, with or without seizures, OMIM:613970;Developmental and epileptic encephalopathy 27, OMIM:616139	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GUCY2C	MECONIUM ILEUS;FAMILIAL DIARRHEA DIARRHEA 6	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
HESX1	Septooptic dysplasia, OMIM:182230;Septooptic dysplasia, MONDO:0008428	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
HK1	Hemolytic anemia due to hexokinase deficiency, OMIM:235700;Neurodevelopmental disorder with visual defects and brain anomalies, OMIM:618547	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
HMGA2	Silver-Russell syndrome 5, OMIM:618908;Silver-Russell syndrome 5, MONDO:0020795	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HNF1B	RENAL CYSTS AND DIABETES SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HNF4A	ATYPICAL DOMINANT FANCONI SYNDROME WITH MODY;HNF4A-RELATED MATURITY-ONSET DIABETES OF THE YOUNG TYPE 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HOXA13	HAND-FOOT-GENITAL SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HOXD13	SYNPOLYDACTYLY 1;BRACHYDACTYLY TYPE D;BRACHYDACTYLY-SYNDACTYLY SYNDROME;BRACHYDACTYLY TYPE E;VACTERL ASSOCIATION;SYNDACTYLY TYPE 5	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HRAS	COSTELLO SYNDROME;CONGENITAL MYOPATHY WITH EXCESS OF MUSCLE SPINDLES	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HSF4	Cataract 5, multiple types, OMIM:116800	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
IDH1	Metaphyseal chondromatosis with D-2-hydroxyglutaric aciduria 614875;Maffucci syndrome 614569;Ollier disease/ Dyschondroplasia 166000	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
IFIH1	Aicardi-Goutieres syndrome 7, OMIM:615846;Singleton-Merten syndrome 1, OMIM:182250	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
IFITM5	OSTEOGENESIS IMPERFECTA TYPE V	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
IGF1R	Insulin-like growth factor I, resistance to, OMIM:270450	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
IGF2	CHROMOSOME 11P15.5-RELATED RUSSELL-SILVER SYNDROME;BECKWITH-WIEDEMANN SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, maternally imprinted (paternal allele expressed)
IHH	Acrocapitofemoral dysplasia, OMIM:607778;Brachydactyly, type A1, OMIM:112500	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
IRF6	POPLITEAL PTERYGIUM SYNDROME;VAN DER WOUDE SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
JAG1	ALAGILLE SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KANSL1	CHROMOSOME 17Q21.31 MICRODELETION SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KAT6B	BLEPHAROPHIMOSIS/INTELLECTUAL DISABILITY PHENOTYPE WHICH IS NOONAN-LIKE;GENITOPATELLAR SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNJ2	Andersen syndrome, OMIM:170390;Andersen-Tawil syndrome, MONDO:0008222	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KIF11	AUTOSOMAL-DOMINANT MICROCEPHALY ASSOCIATED WITH LYMPHEDEMA AND/OR CHORIORETINOPATHY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KIF1A	NEUROPATHY, HEREDITARY SENSORY, TYPE IIC, 614213;NESCAV SYNDROME, 614255	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KIF22	SPONDYLOEPIMETAPHYSEAL DYSPLASIA WITH JOINT LAXITY, TYPE 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KLF1	Dyserythropoietic anemia, congenital, type IV, OMIM:613673;Hydrops Fetalis	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KMT2C	INTELLECTUAL DISABILITY;Kleefstra syndrome 2 617768	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KMT2D	KABUKI SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KRAS	NOONAN SYNDROME TYPE 3;CARDIOFACIOCUTANEOUS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KRIT1	CEREBRAL CAVERNOUS MALFORMATIONS TYPE 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LHX4	LHX4-RELATED COMBINED PITUITARY HORMONE DEFICIENCY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LMBR1	Triphalangeal thumb-polysyndactyly syndrome 174500;Laurin-Sandrow syndrome 135750;Hypoplastic or aplastic tibia with polydactyly 188740;Syndactyly, type IV 186200;Triphalangeal thumb, type I 174500;Polydactyly, preaxial type II 174500;Acheiropody 200500	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
LMNA	HUTCHINSON-GILFORD PROGERIA SYNDROME;MUSCULAR DYSTROPHY CONGENITAL LMNA-RELATED;HEART-HAND SYNDROME SLOVENIAN TYPE;MANDIBULOACRAL DYSPLASIA WITH TYPE A LIPODYSTROPHY;EMERY-DREIFUSS MUSCULAR DYSTROPHY TYPE 2;CARDIOMYOPATHY DILATED WITH HYPERGONADOTROPIC HYPOGONADISM;LETHAL TIGHT SKIN CONTRACTURE SYNDROME;CHARCOT-MARIE-TOOTH DISEASE TYPE 2B1;FAMILIAL PARTIAL LIPODYSTROPHY TYPE 2;LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 1B;CARDIOMYOPATHY DILATED TYPE 1A	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
LMNB1	Microcephaly 26, primary, autosomal dominant, OMIM:619179	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
LMNB2	Microcephaly 27, primary, autosomal dominant, OMIM:619180	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
LMX1B	NAIL-PATELLA SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LTBP3	PLATYSPONDYLY WITH AMELOGENESIS IMPERFECTA	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MAF	CATARACT, DEAFNESS, INTELLECTUAL DISABILITY, SEIZURES, AND A DOWN SYNDROME-LIKE FACIES;CATARACT CONGENITAL CERULEAN TYPE 4;CATARACT PULVERULENT JUVENILE-ONSET MAF-RELATED	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MAFB	Duane Syndrome, Aberrant Extraocular Muscle Innervation, and Inner-Ear Defects;MULTICENTRIC CARPOTARSAL OSTEOLYSIS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MAGEL2	Schaaf-Yang syndrome;ARTHROGRYPOSIS MULTIPLEX CONGENITA;Schaaf-Yang syndrome, 615547	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, maternally imprinted (paternal allele expressed)
MAP2K1	CARDIOFACIOCUTANEOUS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MAP2K2	CARDIOFACIOCUTANEOUS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MAP3K1	46XY SEX REVERSAL 6	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MATN3	MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 5	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MED13L	Impaired intellectual development and distinctive facial features with or without cardiac defects, OMIM:616789	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MEF2C	MENTAL RETARDATION-STEREOTYPIC MOVEMENTS-EPILEPSY AND/OR CEREBRAL MALFORMATIONS	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MEIS2	Cleft palate, cardiac defects, and mental retardation, OMIM:600987;Cardiac malformation, cleft lip/palate, microcephaly, and digital anomalies, MONDO:0010970	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MMP13	METAPHYSEAL ANADYSPLASIA TYPE 1;SPONDYLOEPIMETAPHYSEAL DYSPLASIA MISSOURI TYPE	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MNX1	CURRARINO SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MSX1	Orofacial cleft 5, OMIM:608874	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MSX2	ENLARGED PARIETAL FORAMINA/CRANIUM BIFIDUM;CRANIOSYNOSTOSIS, TYPE 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYBPC1	Lethal congenital contracture syndrome 4 614915;Arthrogryposis, distal, type 1B 614335	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MYCN	FEINGOLD SYNDROME TYPE 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYH2	Proximal myopathy and ophthalmoplegia, OMIM:605637;Myopathy, proximal, and ophthalmoplegia, MONDO:0011577	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MYH3	DISTAL ARTHROGRYPOSIS TYPE 2B;DISTAL ARTHROGRYPOSIS TYPE 2A	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYH6	ATRIAL SEPTAL DEFECT TYPE 3;CARDIOMYOPATHY DILATED TYPE 1EE;CARDIOMYOPATHY FAMILIAL HYPERTROPHIC TYPE 14	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MYH7	Laing distal myopathy, OMIM:160500;Laing early-onset distal myopathy, MONDO:0008050;Cardiomyopathy, hypertrophic, 1, OMIM:192600;Hypertrophic cardiomyopathy 1, MONDO:0008647;Cardiomyopathy, dilated, 1S, OMIM:613426;Dilated cardiomyopathy 1S, MONDO:0013262;Left ventricular noncompaction 5, OMIM:613426	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYH8	DISTAL ARTHROGRYPOSIS TYPE;CARNEY COMPLEX VARIANT	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYH9	SEBASTIAN SYNDROME;DEAFNESS AUTOSOMAL DOMINANT TYPE 17;EPSTEIN SYNDROME;FECHTNER SYNDROME;MACROTHROMBOCYTOPENIA WITH PROGRESSIVE SENSORINEURAL DEAFNESS;MAY-HEGGLIN ANOMALY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYOCD	Megabladder, congenital, OMIM:618719;Megabladder, congenital, MONDO:0032879	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
MYT1	Oculo-auriculo-vertebral spectrum (OAVS);OAVS/Goldenhar syndrome	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NEDD4L	Periventricular nodular heterotopia 7, OMIM:617201;Periventricular nodular heterotopia 7, MONDO:0014966	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NF1	NEUROFIBROMATOSIS-NOONAN SYNDROME;FAMILIAL SPINAL NEUROFIBROMATOSIS;NEUROFIBROMATOSIS TYPE 1;WATSON SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NFIA	Brain malformations with or without urinary tract defects, OMIM:613735	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NFIX	MARSHALL-SMITH SYNDROME;SOTOS-LIKE SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NIPBL	CORNELIA DE LANGE SYNDROME TYPE 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NKX2-5	ATRIAL SEPTAL DEFECT WITH ATRIOVENTRICULAR CONDUCTION DEFECTS;TETRALOGY OF FALLOT;CONGENITAL HYPOTHYROIDISM NON-GOITROUS TYPE 5	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

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NODAL	HETEROTAXY SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NOG	BRACHYDACTYLY TYPE B2;SYMPHALANGISM PROXIMAL SYNDROME;MULTIPLE SYNOSTOSES SYNDROME TYPE 1;TARSAL-CARPAL COALITION SYNDROME;STAPES ANKYLOSIS WITH BROAD THUMB AND TOES	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NOTCH1	ADAMS OLIVER SYNDROME;LEFT VENTRICULAR OUTFLOW TRACT OBSTRUCTION	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NOTCH2	HAJDU-CHENEY SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NR5A1	46XY SEX REVERSAL 3;SPERMATOGENIC FAILURE 8	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NRAS	NOONAN SYNDROME TYPE 6	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NSD1	WEAVER SYNDROME;BECKWITH-WIEDEMANN SYNDROME;SOTOS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
OTX2	MICROPHTHALMIA SYNDROMIC TYPE 5	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PAFAH1B1	Lissencephaly 1, OMIM:607432;Subcortical laminar heterotopia, OMIM:607432	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PAX2	RENAL-COLOBOMA SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PAX3	WAARDENBURG SYNDROME, TYPE 1;CRANIOFACIAL-DEAFNESS-HAND SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PAX6	COLOBOMA OF OPTIC NERVE;ANIRIDIA;BILATERAL OPTIC NERVE HYPOPLASIA;ANIRIDIA CEREBELLAR ATAXIA AND MENTAL DEFICIENCY;KERATITIS HEREDITARY;PETERS ANOMALY;FOVEAL HYPOPLASIA	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PAX8	CONGENITAL HYPOTHYROIDISM NON-GOITROUS TYPE 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PDCD10	CEREBRAL CAVERNOUS MALFORMATIONS TYPE 3	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PDE4D	ACRODYSOSTOSIS	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PEX6	Heimler syndrome 2, OMIM:616617;Peroxisome biogenesis disorder 4A (Zellweger), OMIM:614862;Peroxisome biogenesis disorder 4B, OMIM:614863	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PHIP	Developmental delay, ID, obesity and dysmorphic features	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PHOX2B	NEUROBLASTOMA WITH HIRSCHSPRUNG DISEASE;CENTRAL HYPOVENTILATION SYNDROME, CONGENITAL, WITH OR WITHOUT HIRSCHSPRUNG DISEASE	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PIEZO2	ARTHROGRYPOSIS, DISTAL, TYPE 3;Ataxia, dysmetria, contractures & scoliosis with normal cognition but loss of discriminative touch perception	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PIK3CA	MEGALENCEPHALY-CAPILLARY MALFORMATION-POLYMICROGYRIA SYNDROME, SOMATIC 3;CLOVES: CONGENITAL LIPOMATOUS OVERGROWTH, VASCULAR MALFORMATIONS, AND EPIDERMAL NEVI;HEMIMEGALENCEPHALY PIK3CA	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PIK3R1	AGAMMAGLOBULINEMIA 7, AUTOSOMAL RECESSIVE;SHORT SYNDROME	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, OMIM:603387	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PITX1	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, OMIM:119800;Clubfoot, MONDO:0007342;Liebenberg syndrome, OMIM:186550;Brachydactyly-elbow wrist dysplasia syndrome, MONDO:0008520	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PITX2	RING DERMOID OF CORNEA;IRIDOGONIODYSGENESIS TYPE 2;AXENFELD-RIEGER SYNDROME TYPE 1;PETERS ANOMALY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PITX3	ANTERIOR SEGMENT MESENCHYMAL DYSGENESIS;CATARACT POSTERIOR POLAR TYPE 4;CATARACT AUTOSOMAL DOMINANT	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PKD1	Polycystic kidney disease, 173900;Autosomal recessive polycystic kidney disease (ARPKD);Autosomal dominant polycystic kidney disease (ADPKD)	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PKD2	Polycystic kidney disease 613095	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PLCB4	AURICULOCONDYLAR SYNDROME	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PLEC	Muscular dystrophy, limb-girdle, autosomal recessive 17, OMIM:613723;Epidermolysis bullosa simplex 5A, Ogna type, OMIM:131950;Epidermolysis bullosa simplex 5C, with pyloric atresia, OMIM:612138;Epidermolysis bullosa simplex with muscular dystrophy, OMIM:226670	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
POGZ	INTELLECTUAL DISABILITY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
POLG2	Mitochondrial DNA depletion syndrome syndrome 16 (hepatic type), OMIM:618528;Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), OMIM:619425;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, OMIM:610131	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
POLR1D	TREACHER COLLINS SYNDROME TYPE 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PPP2R1A	INTELLECTUAL DISABILITY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PRKACA	Cardioacrofacial dysplasia 1, OMIM:619142	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PRKAG2	Cardiomyopathy, hypertrophic 6, OMIM:600858;Hypertrophic cardiomyopathy 6, MONDO:0010946;Glycogen storage disease of heart, lethal congenital, OMIM:261740;Lethal congenital glycogen storage disease of heart, MONDO:0009867	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PRKAR1A	ACRODYSOSTOSIS	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PRRX1	Agnathia-otocephaly complex, OMIM:202650	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PTCH1	Holoprosencephaly 7, OMIM:610828	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PTH1R	CHONDRODYSPLASIA BLOMSTRAND TYPE;EIKEN SKELETAL DYSPLASIA;JANSEN METAPHYSEAL CHONDRODYSPLASIA;PRIMARY FAILURE OF TOOTH ERUPTION	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PTHLH	BRACHYDACTYLY, TYPE E2;CLUBBING WITH SKELETAL DYSPLASIA INC ACROOSTEOLYSIS	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PTPN11	LEOPARD SYNDROME TYPE 1;NOONAN SYNDROME 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RAC1	Developmental Disorders with Diverse Phenotypes	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RAD21	COHESINOPATHY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RAD51	Fanconi anaemia, complementation group R, OMIM:617244	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
RAF1	NOONAN SYNDROME 5	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RAI1	SMITH-MAGENIS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RASA1	PARKES WEBER SYNDROME;CAPILLARY MALFORMATION-ARTERIOVENOUS MALFORMATION	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RBP4	Microphthalmia, isolated, with coloboma 10, OMIM:616428	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
RET	RENAL AGENESIS;MULTIPLE ENDOCRINE NEOPLASIA IIB	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ROBO1	Tetralogy of Fallot and septal defects;Neurooculorenal syndrome, OMIM:620305	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ROR2	Brachydactyly, type B1, OMIM:113000 (AD);Robinow syndrome, autosomal recessive, OMIM:268310 (AR)	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
RPL11	Diamond-Blackfan anemia 7 612562;Diamond-Blackfan anemia with cleft palate and abnormal thumbs	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RPL35A	Diamond-Blackfan anemia 5, OMIM:612528;Diamond-Blackfan anemia 5, MONDO:0012925	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RPL5	Diamond-Blackfan anemia 6 612561	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RPS10	Diamond-Blackfan anemia 9 613308	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RPS17	Diamond-Blackfan anemia 4 612527	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RPS19	RPS19-RELATED DIAMOND-BLACKFAN ANEMIA	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RPS24	Diamond-blackfan anemia 3, OMIM:610629;Diamond-Blackfan anemia 3, MONDO:0012529	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RPS26	Diamond-Blackfan anemia 10 613309	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RPS7	Diamond-Blackfan anemia 8, OMIM:612563;Diamond-Blackfan anemia 8, MONDO:0012939	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
RUNX2	CLEIDOCRANIAL DYSPLASIA	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SALL1	TOWNES-BROCKS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SALL4	DUANE-RADIAL RAY SYNDROME;ACRO-RENAL-OCULAR SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SAMD9	MIRAGE - myelodysplasia, infection, restriction of growth, adrenal hypoplasia, genital phenotypes, enteropathy	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SATB2	NONSPECIFIC SEVERE ID;SYNDROMAL PIERRE ROBIN SEQUENCE;CLEFT PALATE ISOLATED	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCN1A	Dravet syndrome, OMIM:607208;Arthrogryposis multiplex congenita	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCN2A	NONSPECIFIC SEVERE ID;INFANTILE EPILEPTIC ENCEPHALOPATHY;BENIGN FAMILIAL NEONATAL INFANTILE SEIZURES	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCN3A	Epileptic encephalopathy, early infantile, 62, OMIM:617938;Epilepsy, familial focal, with variable foci 4, OMIM:617935;Intellectual disability;Malformations of cortical development	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SCN4A	PARAMYOTONIA CONGENITA OF VON EULENBURG;HYPERKALEMIC PERIODIC PARALYSIS TYPE 1;HYPOKALEMIC PERIODIC PARALYSIS	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCN5A	{Sudden infant death syndrome, susceptibility to}, OMIM:272120;Long QT syndrome 3, OMIM:603830	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SETBP1	DEVELOPMENTAL AND EXPRESSIVE LANGUAGE DELAY;SCHINZEL-GIEDION MIDFACE RETRACTION SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SETD2	microcephaly;profound intellectual disability;congenital anomalies;dysmorphic facial features	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SF3B4	ACROFACIAL DYSOSTOSIS 1, NAGER TYPE	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SHANK3	Phelan-McDermid syndrome, OMIM:606232;Phelan-McDermid syndrome, MONDO:0011652	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SHH	HOLOPROSENCEPHALY TYPE 3;TRIPHALANGEAL THUMB-POLYSYNDACTYLY SYNDROME;MICROPHTHALMIA ISOLATED WITH COLOBOMA TYPE 5;SOLITARY MEDIAN MAXILLARY CENTRAL INCISOR	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SHOC2	NOONAN-LIKE SYNDROME WITH LOOSE ANAGEN HAIR	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SHOX	Langer mesomelic dysplasia, OMIM:249700 (PR);Leri-Weill dyschondrosteosis, OMIM:127300 (PD);Short stature, idiopathic familial, OMIM:300582	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SIX3	HOLOPROSENCEPHALY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

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SIX5	BRANCHIOOTORENAL SYNDROME TYPE 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SKI	SHPRINTZEN-GOLDBERG CRANIOSYNOSTOSIS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SLC12A6	Charcot-Marie-Tooth disease, axonal, type 2II, OMIM:620068;Agenesis of the corpus callosum with peripheral neuropathy, OMIM:218000	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SMAD2	Loeys-Dietz syndrome 6, OMIM:619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, OMIM:619657	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SMAD3	SMAD3-RELATED LOEYS-DIETZ SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMAD4	MYHRE SYNDROME;JUVENILE POLYPOSIS/HEREDITARY HEMORRHAGIC TELANGIECTASIA SYNDROME;JUVENILE POLYPOSIS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMARCA2	NICOLAIDES-BARAITSER SYNDROME;COFFIN SIRIS	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMARCA4	RHABDOID TUMOR PREDISPOSITION SYNDROME 2;COFFIN SIRIS	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMARCB1	RHABDOID PREDISPOSITION SYNDROME 1;?COFFIN-SIRIS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMARCE1	Coffin-Siris syndrome 5, OMIM:616938	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMC3	CORNELIA DE LANGE SYNDROME TYPE 3	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SOS1	NOONAN SYNDROME 4	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SOST	SOST-Related Sclerosing Bone Dysplasias 122860;Sclerosteosis 1, 269500;Craniodiaphyseal dysplasia, autosomal dominant, 122860	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SOX10	PERIPHERAL DEMYELINATING NEUROPATHY, CENTRAL DYSMYELINATING LEUKODYSTROPHY, WAARDENBURG SYNDROME, AND HIRSCHSPRUNG DISEASE;KALLMANN SYNDROME WITH DEAFNESS;YEMENITE DEAF-BLIND HYPOPIGMENTATION SYNDROME;WAARDENBURG SYNDROME TYPE 4C;WAARDENBURG SYNDROME TYPE 2E	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SOX17	VESICOURETERAL REFLUX TYPE 3	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SOX18	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, OMIM:137940;Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, MONDO:0019073;Hypotrichosis-lymphedema-telangiectasia syndrome, OMIM:607823;Hypotrichosis-lymphedema-telangiectasia syndrome, MONDO:0011914	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SOX2	AEG SYNDROME;MICROPHTHALMIA SYNDROMIC TYPE 3	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SOX6	Tolchin-Le Caignec syndrome, OMIM:618971;Tolchin-Le Caignec syndrome, MONDO:0033544	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SOX9	PIERRE ROBIN SEQUENCE;CAMPOMELIC DYSPLASIA	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SPECC1L	?Facial clefting, oblique, 1, OMIM:600251;Tessier number 4 facial cleft, MONDO:0010850;Hypertelorism, Teebi type, OMIM:145420;Hypertelorism, Teebi type, MONDO:0007780;Opitz GBBB syndrome, type II, OMIM:145410;Autosomal dominant Opitz G/BBB syndrome, MONDO:0007779	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SPRED1	LEGIUS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SPTB	Elliptocytosis-3, OMIM:617948;Anemia, neonatal hemolytic, fatal or near-fatal, OMIM:617948;Spherocytosis, type 2, OMIM:616649	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SRCAP	FLOATING-HARBOR SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SUFU	Joubert syndrome 32, OMIM:617757;Joubert Syndrome with Cranio-facial and Skeletal Defects	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
TAB2	Congenital heart defects, nonsyndromic, 2, OMIM:614980	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBL1XR1	Intellectual disability with autism spectrum disorder;Pierpont syndrome	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX1	22Q11.2 DELETION SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX20	ATRIAL SEPTAL DEFECT TYPE 4	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX3	ULNAR-MAMMARY SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX4	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, OMIM:147891	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX5	HOLT-ORAM SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TBX6	Spondylocostal dysostosis 5 122600	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TCF4	PITT-HOPKINS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TCOF1	TREACHER COLLINS SYNDROME TYPE 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TFAP2A	BRANCHIOOCULOFACIAL SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TFAP2B	CHAR SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGFB2	LOEYS-DIETZ SYNDROME, TYPE 4	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGFB3	LOEYS-DIETZ SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGFBR1	LOEYS-DIETZ SYNDROME TYPE 2A;LOEYS-DIETZ SYNDROME TYPE 1A;AORTIC ANEURYSM FAMILIAL THORACIC TYPE 5	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGFBR2	LOEYS-DIETZ SYNDROME;TGFBR2-RELATED LOEYS-DIETZ SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGIF1	Holoprosencephaly 4 142946;HOLOPROSENCEPHALY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
THRA	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 6	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TINF2	EXUDATIVE RETINOPATHY WITH BONE MARROW FAILURE	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TNNI2	Arthrogryposis multiplex congenita, distal, type 2B 601680	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TNNT3	Arthrogryposis, distal, type 2B2, OMIM:618435;Arthrogryposis, distal, type 2B2, MONDO:0032750	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TP63	ADULT syndrome, OMIM:103285;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, OMIM:604292;Hay-Wells syndrome, OMIM:106260;Limb-mammary syndrome, OMIM:603543;Orofacial cleft 8, OMIM:618149;Rapp-Hodgkin syndrome, OMIM:129400;Split-hand/foot malformation 4, OMIM:605289	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TPM2	ARTHROGRYPOSIS, DISTAL, TYPE 1;Arthrogryposis multiplex congenita, distal, type 1, 108120;Arthrogryposis, distal, type 2B, 601680	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TPM3	Congenital fiber-type disproportion myopathy 255310	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
TRIP12	TRIP12-related intellectual disability with/without autism spectrum disorder	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TRPS1	TRICHO-RHINO-PHALANGEAL SYNDROME TYPE 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TRPV4	METATROPIC DYSPLASIA;SPONDYLOMETAPHYSEAL DYSPLASIA, KOZLOWSKI TYPE	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TRRAP	Developmental delay with or without dysmorphic facies and autism, OMIM:618454;multiple congenital anomalies	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TSC1	TUBEROUS SCLEROSIS TYPE 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TSC2	LYMPHANGIOLEIOMYOMATOSIS;TUBEROUS SCLEROSIS TYPE 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TUBA1A	INTELLECTUAL DISABILITY;LISSENCEPHALY TYPE 3	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TUBB2B	Cortical dysplasia, complex, with other brain malformations 7, OMIM:610031	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TUBB3	CORTICAL DYSPLASIA, COMPLEX, WITH OTHER BRAIN MALFORMATIONS 1;CONGENITAL FIBROSIS OF THE EXTRAOCULAR MUSCLES	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TWIST1	SAETHRE-CHOTZEN SYNDROME;CRANIOSYNOSTOSIS, TYPE 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TWIST2	Ablepharon-macrostomia syndrome, 200110;Barber-Say syndrome, 209885	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
WNT5A	WNT5A-RELATED ROBINOW SYNDROME, AUTOSOMAL DOMINANT	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
WT1	FRASIER SYNDROME FRASIER SYNDROME FRASIER SYNDROME;DENYS-DRASH SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
YY1	INTELLECTUAL DISABILITY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZBTB18	ZBTB18 syndrome	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZEB2	MOWAT-WILSON SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZIC1	CRANIOSYNOSTOSIS 6	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZIC2	HOLOPROSENCEPHALY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ACO2	INFANTILE CEREBELLAR-RETINAL DEGENERATION	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ACVR1	Fibrodysplasia ossificans progressiva, OMIM:135100	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ANKRD26	THROMBOCYTOPENIA 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ATP1A3	Developmental and epileptic encephalopathy 99, OMIM:619606;Polymicrogyria	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CACNA1A	Developmental and epileptic encephalopathy 42, OMIM:617106	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CACNA1D	Primary aldosteronism, seizures, and neurologic abnormalities, OMIM:615474	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CAMTA1	CEREBELLAR ATAXIA, NONPROGRESSIVE, WITH MENTAL RETARDATION	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CD96	C SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CELSR1	Lymphatic malformation 9, OMIM:619319	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHD3	Apraxia of speech	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHD8	AUTISM	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHRNB2	CHRNB2-RELATED NOCTURNAL FRONTAL LOBE EPILEPSY, AUTOSOMAL DOMINANT;NOCTURNAL FRONTAL LOBE EPILEPSY, AUTOSOMAL DOMINANT	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CITED2	Atrial septal defect 8, OMIM:614433;Ventricular septal defect 2, OMIM:614431;Congenital heart disease	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CRELD1	HETEROTAXY SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
DHDDS	Epilepsy and intellectual disability	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

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DNM1	EPILEPTIC ENCEPHALOPATHY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EDN1	AURICULOCONDYLAR SYNDROME	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
EDN3	Central hypoventilation syndrome, congenital, OMIM:209880;Waardenburg syndrome, type 4B, OMIM:613265;{Hirschsprung disease, susceptibility to, 4}, OMIM:613712	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ENG	Telangiectasia, hereditary hemorrhagic, type 1, OMIM:187300	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FBXW11	Neurodevelopmental, jaw, eye, and digital syndrome, OMIM:618914	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FGF9	Multiple synostoses syndrome 3, OMIM:612961	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FN1	Spondylometaphyseal Dysplasia with Corner Fractures	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXL2	BLEPHAROPHIMOSIS, PTOSIS, AND EPICANTHUS INVERSUS SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXP2	Speech-language disorder-1, OMIM:602081;Structural abnormalities of basal ganglia	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GABRA1	EPILEPTIC ENCEPHALOPATHY;JUVENILE MYOCLONIC EPILEPSY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GABRG2	EPILEPSY, GENERALIZED, WITH FEBRILE SEIZURES PLUS, TYPE 3;GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, TYPE 3	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GLMN	Glomulovenous malformations, OMIM:138000	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
GNA14	Congenital vascular tumours	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GNAQ	Congenital Hemangioma	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HPD	TYROSINEMIA TYPE 3;HAWKINSINURIA	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KCNC3	SPINOCEREBELLAR ATAXIA TYPE 13	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNJ6	KEPPEN-LUBINSKY SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNJ8	Cantu syndrome	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KCNK9	Birk-Barel syndrome, OMIM:612292	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)
KRT74	HYPOTRICHOSIS SIMPLEX OF THE SCALP 2	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MIR17HG	FEINGOLD SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MPZ	Hypomyelinating neuropathy, congenital, 2, OMIM:618184	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NEXN	Cardiomyopathy, dilated, 1CC, OMIM:613122;Cardiomyopathy, hypertrophic, 20, OMIM:613876	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
NID1	Hydrocephalus with or without seizures;Dandy-Walker malformation and occipital cephalocele	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NTRK2	Epilepsy and intellectual disability	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ORAI1	Myopathy, tubular aggregate, 2, OMIM:615883	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PDE10A	Childhood-Onset Chorea with Bilateral Striatal Lesions	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
POLD1	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, OMIM:615381	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
RORA	INTELLECTUAL DISABILITY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SHANK2	SUSCEPTIBILITY TO AUTISM TYPE 17	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SHROOM3	NEURAL TUBE DEFECT	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SLC1A2	EPILEPTIC ENCEPHALOPATHY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SLC25A4	Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SLC4A1	Ovalocytosis, SA type, OMIM:166900	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SOX5	12P12.5 INTRAGENIC DELETIONS ASSOCIATED WITH INTELLECTUAL DISABILITY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SPTAN1	Developmental and epileptic encephalopathy 5, OMIM:613477;developmental and epileptic encephalopathy, 5, MONDO:0013277	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
STAT3	Autoimmune disease, multisystem, infantile-onset, 1, OMIM:615952;Hyper-IgE recurrent infection syndrome, OMIM:147060	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
STIM1	Myopathy, tubular aggregate, OMIM:160565;Immunodeficiency 10, OMIM:612783;Stormorken syndrome, OMIM:185070	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SYT2	Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, OMIM:619461;Myasthenic syndrome, congenital, 7, presynaptic, OMIM:616040	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
TBC1D1	CAKUT	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TLL1	Atrial septal defect 6, OMIM:613087	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TRIO	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, OMIM:617061	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TRPM7	Cardiac arrhythmia, stillbirth	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TRPV3	OLMSTED SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
WNT4	SERKAL SYNDROME;MULLERIAN APLASIA AND HYPERANDROGENISM	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
YAP1	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, OMIM:120433	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ZFPM2	Diaphragmatic hernia 3, OMIM:610187;Tetralogy of Fallot, OMIM:187500	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ZMYND11	INTELLECTUAL DISABILITY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ZNF750	SEBORRHEA-LIKE DERMATITIS WITH PSORIASIFORM ELEMENTS	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ABCC8	Hyperinsulinemic hypoglycemia, familial 256450	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ANO5	MIYOSHI MUSCULAR DYSTROPHY TYPE 3;GNATHODIAPHYSEAL DYSPLASIA	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
AUTS2	Intellectual developmental disorder, autosomal dominant 26, OMIM:615834	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CHD2	EPILEPTIC ENCEPHALOPATHY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHRNA4	NOCTURNAL FRONTAL LOBE EPILEPSY TYPE 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL4A3	ALPORT SYNDROME AUTOSOMAL RECESSIVE;ALPORT SYNDROME AUTOSOMAL DOMINANT	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COL5A1	Ehlers-Danlos syndrome, classic type 130000	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL5A2	Ehlers-Danlos syndrome, classic type 130000	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
COL9A3	Stickler syndrome, type VI, OMIM:620022;Epiphyseal dysplasia, multiple, 3, with or without myopathy, OMIM:600969	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
COMP	ARE THE CAUSE OF PSEUDOACHONDROPLASIA;MULTIPLE EPIPHYSEAL DYSPLASIA TYPE 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CRX	CRX-RELATED LEBER CONGENITAL AMAUROSIS LEBER CONGENITAL AMAUROSIS 7	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CYP19A1	Aromatase deficiency, OMIM:613546;Aromatase excess syndrome, OMIM:139300	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
DICER1	GLOW syndrome, somatic mosaic, OMIM:618272;Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors , OMIM:138800;Pleuropulmonary blastoma, OMIM:601200	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
DSPP	DENTINOGENESIS IMPERFECTA, SHIELDS TYPE II;DEAFNESS AUTOSOMAL DOMINANT TYPE 39 WITH DENTINOGENESIS IMPERFECTA 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXP1	MENTAL RETARDATION WITH LANGUAGE IMPAIRMENT AND AUTISTIC FEATURES	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GABRB3	EPILEPTIC ENCEPHALOPATHIES;CHILDHOOD ABSENCE EPILEPSY TYPE 5	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GCH1	GTP CYCLOHYDROLASE 1 DEFICIENCY;DYSTONIA TYPE 5	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
GJB2	VOHWINKEL SYNDROME;ICHTHYOSIS HYSTRIX-LIKE WITH DEAFNESS SYNDROME;PALMOPLANTAR KERATODERMA WITH DEAFNESS;BART-PUMPHREY SYNDROME;DEAFNESS AUTOSOMAL RECESSIVE TYPE 1A	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GLUD1	HYPERINSULINISM-HYPERAMMONEMIA SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GRIN2A	EPILEPSY WITH NEURODEVELOPMENTAL DEFECTS;LANDAU-KLEFFNER SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
H19	Wilms tumor 2 194071;Beckwith-Wiedemann syndrome 130650;Silver-Russell syndrome 180860	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HCN1	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 24	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HDAC4	BRACHYDACTYLY-MENTAL RETARDATION SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HNRNPU	EPILEPTIC ENCEPHALOPATHY	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
HOXA2	Microtia with or without hearing impairment (AD), OMIM:612290	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ITPR1	Spinocerebellar ataxia 29, congenital nonprogressive, OMIM:117360;Spinocerebellar ataxia 15, OMIM:606658;Gillespie syndrome, OMIM:206700	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KBTBD13	NEMALINE MYOPATHY 6	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNJ11	DIABETES MELLITUS, KCNJ11-RELATED TRANSIENT NEONATAL;FAMILIAL HYPERINSULINISM	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KCNQ1	Long QT syndrome 1, OMIM:192500	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNQ2	BENIGN NEONATAL EPILEPSY TYPE 1;EPILEPTIC ENCEPHALOPATHY EARLY INFANTILE TYPE 7	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KCNQ3	KCNQ3 syndrome	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KIT	HUMAN PIEBALDISM	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
KMT2E	INTELLECTUAL DISABILITY;O'Donnell-Luria-Rodan syndrome, 618512	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LDB3	CARDIOMYOPATHY DILATED TYPE 1C;MYOPATHY MYOFIBRILLAR TYPE 4;LEFT VENTRICULAR NON-COMPACTION TYPE 3	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
LEMD3	MELORHEOSTOSIS;BUSCHKE-OLLENDORFF SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MYT1L	MYT1L syndrome	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

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NKX2-1	BENIGN HEREDITARY CHOREA;CHOREOATHETOSIS, HYPOTHYROIDISM, AND NEONATAL RESPIRATORY DISTRESS	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NLRP3	CINCA syndrome, OMIM:607115	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NR2F1	BOSCH-BOONSTRA OPTIC ATROPHY SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NRXN2	AUTISM	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NSMF	Hypogonadotropic hypogonadism 9 with or without anosmia, OMIM:614838	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PAX9	TOOTH AGENESIS, SELECTIVE, 3	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PMP22	Neuropathy, recurrent, with pressure palsies 162500;Charcot-Marie-Tooth disease, type 1A 118220;Charcot-Marie-Tooth disease, type 1E 118300;Dejerine-Sottas disease 145900;Neuropathy, inflammatory demyelinating 139393;Roussy-Levy syndrome 180800	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PPM1D	PPM1D syndrome	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PROK2	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PROKR2	Hypogonadotropic hypogonadism 3 with or without anosmia 244200	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PRRT2	BENIGN FAMILIAL INFANTILE EPILEPSY AND INFANTILE CONVULSIONS WITH CHOREOATHETOSIS SYNDROME;AUTOSOMAL RECESSIVE MENTAL RETARDATION	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PTEN	MACROCEPHALY/AUTISM SYNDROME;BANNAYAN-ZONANA SYNDROME;PROTEUS SYNDROME;VACTERL ASSOCIATION WITH HYDROCEPHALUS;COWDEN DISEASE;LHERMITTE-DUCLOS DISEASE	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCN1B	EPILEPSY, GENERALIZED, WITH FEBRILE SEIZURES PLUS, TYPE 1;BRUGADA SYNDROME 5	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCN8A	COGNITIVE IMPAIRMENT WITH OR WITHOUT CEREBELLAR ATAXIA;EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 13	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCNN1A	Pseudohypoaldosteronism, type I, OMIM:264350	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SIM1	Severe obesity with neurobehavioral features	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SIX1	BRANCHIOOTIC SYNDROME TYPE 3;DEAFNESS AUTOSOMAL DOMINANT TYPE 23	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SLC2A1	GLUT1 DEFICIENCY SYNDROME TYPE 2;GLUT1 DEFICIENCY SYNDROME TYPE 1	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SLC6A1	EPILEPSY WITH MYOCLONIC-ATONIC SEIZURES	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SMAD6	{Craniosynostosis 7, susceptibility to}, OMIM:617439;Aortic valve disease 2, OMIM:614823;{Radioulnar synostosis, nonsyndromic}, OMIM:179300	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SPTA1	Elliptocytosis-2, OMIM:130600;Spherocytosis, type 3, OMIM:270970;Hydrops fetalis;Congenital anaemia	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
SPTLC2	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE IC	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
STAT1	STAT1 DEFICIENCY COMPLETE;MENDELIAN SUSCEPTIBILITY TO MYCOBACTERIAL DISEASE;FAMILIAL CANDIDIASIS TYPE 7	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
STXBP1	EPILEPTIC ENCEPHALOPATHY EARLY INFANTILE TYPE 4;ANGELMAN/PITT HOPKINS SYNDROME-LIKE DISORDER	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SYNGAP1	EPILEPTIC ENCEPHALOPATHY;MENTAL RETARDATION AUTOSOMAL DOMINANT TYPE 5	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TEK	VENOUS MALFORMATIONS, MULTIPLE CUTANEOUS AND MUCOSAL	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGFB1	Camurati-Engelmann disease, OMIM:131300	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
THAP1	DYSTONIA 6, TORSION	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TNXB	Ehlers-Danlos syndrome due to tenascin X deficiency 606408;Vesicoureteral reflux 8 615963	Fetal anomalies	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
UBE3A	ANGELMAN SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)
WDR11	KALLMANN SYNDROME	Fetal anomalies	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BRAF	LEOPARD syndrome 3 613707;Cardiofaciocutaneous syndrome 115150;Noonan syndrome 7 613706	RASopathies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia 613563	RASopathies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
HRAS	Costello syndrome, 218040	RASopathies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KRAS	Noonan syndrome 3 609942;Cardiofaciocutaneous syndrome 2 615278	RASopathies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279;LEOPARD syndrome;?Noonan syndrome	RASopathies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MAP2K2	Cardiofaciocutaneous syndrome 4 615280	RASopathies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NF1	Neurofibromatosis-Noonan syndrome 601321;Neurofibromatosis, type 1 162200	RASopathies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NRAS	Noonan syndrome 6 613224;Cardio-Facio-cutanenous syndrome	RASopathies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PTPN11	LEOPARD syndrome 1 151100;Noonan syndrome 1 163950	RASopathies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
RAF1	LEOPARD syndrome 2 611554;Noonan syndrome 5 611553	RASopathies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SHOC2	Noonan syndrome-like with loose anagen hair 1, 607721	RASopathies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SOS1	Noonan syndrome 4 610733	RASopathies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SPRED1	Legius syndrome 611431	RASopathies	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FGFR1	Jackson-Weiss syndrome OMIM:123150;Osteoglophonic dysplasia OMIM:166250;Pfeiffer syndrome OMIM:101600;Trigonocephaly 1 OMIM:190440	Common craniosynostosis syndromes	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGFR2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis OMIM:207410;Apert syndrome OMIM:101200;Beare-Stevenson cutis gyrata syndrome OMIM:123790;Pfeiffer syndrome OMIM:101600;Craniofacial-skeletal-dermatologic dysplasia OMIM:101600;Crouzon syndrome OMIM:123500;Jackson-Weiss syndrome OMIM:123150;Saethre-Chotzen syndrome OMIM:101400;Scaphocephaly, maxillary retrusion, and mental retardation OMIM:609579	Common craniosynostosis syndromes	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGFR3	Muenke syndrome OMIM:602849;Crouzon syndrome with acanthosis nigricans OMIM:612247;Thanatophoric dysplasia, type I OMIM:187600;Thanatophoric dysplasia, type II OMIM:187601	Common craniosynostosis syndromes	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TWIST1	Craniosynostosis 1 OMIM:123100;Saethre-Chotzen syndrome with or without eyelid anomalies OMIM:101400	Common craniosynostosis syndromes	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ACTB	Baraitser-Winter syndrome 1, OMIM:243310;Baraitser-Winter syndrome 1, MONDO:0009470	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ACTG1	Baraitser-Winter syndrome 2, OMIM:614583	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ALPL	hypophosphatasia	Rare syndromic craniosynostosis or isolated multisuture synostosis	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ALX4	Parietal foramina;Parietal foramina 2, (AD), 609597;Frontonasal dysplasia 2, (AR), 613451	Rare syndromic craniosynostosis or isolated multisuture synostosis	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
ARID1B	Coffin-Siris syndrome 1, OMIM:135900	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ASXL1	Bohring-Opitz syndrome, 605039;Metopic synostosis frequently associated with Bohring-Opitz syndrome	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
BRAF	cardiofaciocutaneous syndrome type 115150;Noonan syndrome type 7 613706	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted

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CHD7	CHARGE syndrome, OMIM:214800;CHARGE syndrome, MONDO:0008965	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FBN1	Marfan syndrome, OMIM:154700;Marfan lipodystrophy syndrome, OMIM:616914;craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FGF9	Multiple synostoses syndrome 3, OMIM:612961	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FGFR1	Craniosynostosis	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGFR2	Crouzon syndrome, 123500; Jackson-Weiss syndrome, 123150; Beare-Stevenson cutis gyrata syndrome, 123790; Pfeiffer syndrome, 101600; Apert syndrome, 101200; Saethre-Chotzen; Craniosynostosis, nonspecific syndrome, 101400; Gastric cance;Craniosynostosis	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FGFR3	Muenke syndrome;Crouzon syndrome with acanthosis nigricans	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
GLI3	Greig cephalopolysyndactyly syndrome, OMIM:175700	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
GNAS	103580;pseudohypoparathyroidism type 1a 103580;pseudohypoparathyroidism type 1a	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, maternally imprinted (paternal allele expressed)
IHH	Syndactyly, type 1, with or without craniosynostosis, OMIM:185900;Chr2q35dup syndrome	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
JAG1	Alagille syndrome 1, OMIM:118450	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KAT6B	KAT6B-related disorders;Genitopatellar syndrome, OMIM:606170	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KMT2D	Kabuki syndrome 1, OMIM:147920	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KRAS	cardiofaciocutaneous syndrome type 2 615278;Noonan syndrome type 3 609942;615278;609942	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MSX2	Craniosynostosis, type 2, 604757;Parietal foramina 1, 168500;Parietal foramina with cleidocranial dysplasia, 168550;Craniosynostosis;MSX2-related craniosynostosis	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NFIA	Metopic synostosis, hydrocephalus, thin corpus callosum, mild developmental delay, autism, macrocephaly	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NFIX	Malan syndrome, OMIM:614753;Marshall-Smith syndrome, OMIM:602535;craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PRRX1	Agnathia-otocephaly complex, OMIM:202650;craniosynostosis, MONDO:0015469;craniosynostosis, various combinations of sutures	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PTCH1	metopic craniosynostosis	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PTPN11	Noonan syndrome type 1 163950;leopard syndrome 151100	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
RUNX2	Craniosynostosis;156510	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SIX1	sagittal synostosis, multi-suture synostosis	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted

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SKI	Shprintzen-Goldberg syndrome 182212;182212	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SMAD6	metopic synostosis;sagittal synostosis;{Craniosynostosis 7, susceptibility to} 617439	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SOX6	Tolchin-Le Caignec syndrome, OMIM:618971;craniosynostosis	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SPECC1L	Opitz GBBB syndrome, type II 145410	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
STAT3	Hyper IgE recurrent infection syndrome 147060;147060	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TFAP2B	Char syndrome 169100	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
TGFBR1	Loeys-Dietz syndrome 1 609192;609192	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TGFBR2	610168;Loeys-Dietz syndrome 2 610168	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TWIST1	Saethre-Chotzen syndrome, 101400; Saethre-Chotzen syndrome with eyelid anomalies, 101400; Craniosynostosis, type 1, 123100; Robinow-Sorauf syndrome, 180750	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ZEB2	Mowat-Wilson syndrome 235730;235730	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ZIC1	?Craniosynostosis 6, 616602;Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ANKH	Craniometaphyseal dysplasia, OMIM:123000;craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AXIN2	Oligodontia-colorectal cancer syndrome, OMIM:608615;craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FGF10	craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FREM1	Trigonocephaly 2, OMIM:614485	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SMAD3	Loeys-Dietz syndrome 3, OMIM:613795	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
ABCC9	hypertrichotic osteochondrodysplasia, Cantu syndrome 239850	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
ACVRL1	craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
AXIN1	craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CACNA1E	craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
CHD3	craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

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COL11A1	craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
EHMT1	craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXP1	craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
FOXP2	craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
GLI2	craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
IFRD1	craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
MED13L	Impaired intellectual development and distinctive facial features with or without cardiac defects, OMIM:616789;craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
NTRK2	Obesity, hyperphagia, and developmental delay, OMIM:613886;craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
PITX2	Anterior segment dysgenesis 4, MIM: 137600; Axenfeld-Rieger syndrome, type 1; 180500; Ring dermoid of cornea, MIM: 180550	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SCN8A	craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SIX2	six2-related frontonasal dysplasia, MONDO:0044628;craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SMAD2	Loeys-Dietz syndrome 6, OMIM:619656	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SP7	craniosynostosis, MONDO:0015469	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
SRCAP	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, MIM: 619595; Floating-Harbor syndrome, MIM: 136140	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
TWIST2	Ablepharon-macrostomia syndrome;Barber-Say syndrome;Focal facial dermal dysplasia 3, Setleis type	Rare syndromic craniosynostosis or isolated multisuture synostosis	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
BRAF	LEOPARD Syndrome;Noonan Syndrome;Cardiofaciocutaneous Syndrome;LEOPARD syndrome 3;Cardio-facio-cutaneous syndrome	Fetal hydrops	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;NSLL;Noonan syndrome-like disorder associated with JMML;Fetal hydrops (in some patients)	Fetal hydrops	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FLT4	Lymphedema, hereditary, IA, 153100;Hereditary lymphoedema type 1a;fetal hydrops	Fetal hydrops	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
FOXC2	Lymphedema-distichiasis syndrome, 153400;fetal hydrops	Fetal hydrops	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
HRAS	Costello syndrome	Fetal hydrops	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KLF1	Dyserythropoietic anemia, congenital, type IV, OMIM:613673;Hydrops Fetalis	Fetal hydrops	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
KMT2D	Kabuki syndrome	Fetal hydrops	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
KRAS	Noonan syndrome 3;Noonan syndrome;Cardiofaciocutaneous syndrome 2;Cardiofaciocutaneous Syndrome;Cardio-Facio-Cutaneous syndrome;CFC syndrome	Fetal hydrops	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MAP2K1	Cardiofaciocutaneous syndrome 3;Cardiofaciocutaneous Syndrome;Cardio-Facio-Cutaneous syndrome;CFC syndrome;LEOPARD syndrome;?Noonan syndrome	Fetal hydrops	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MAP2K2	Cardiofaciocutaneous syndrome 4;Cardio-Facio-Cutaneous syndrome type 4;Cardiofaciocutaneous Syndrome;Cardio-Facio-Cutaneous syndrome;CFC syndrome	Fetal hydrops	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
NRAS	Noonan syndrome;Noonan syndrome 6;Cardio-Facio-cutanenous syndrome;CFC Syndrome	Fetal hydrops	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
PEX6	Peroxisome biogenesis disorder 4A (Zellweger), OMIM:614862;Peroxisome biogenesis disorder 4B, OMIM:614863	Fetal hydrops	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
PTPN11	LEOPARD syndrome;LEOPARD syndrome 1;Noonan syndrome;Noonan syndrome 1	Fetal hydrops	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
RAF1	Noonan syndrome;Noonan syndrome 5;LEOPARD syndrome;LEOPARD syndrome 2	Fetal hydrops	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SHOC2	Noonan-like syndrome with loose anagen hair	Fetal hydrops	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
SOS1	Noonan syndrome;Noonan syndrome 4	Fetal hydrops	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
MECP2	Rett syndrome, 312750Mental retardation, X-linked, syndromic 13, 300055Rett syndrome, preserved speech variant, 312750Encephalopathy, neonatal severe, 300673{Autism susceptibility, X-linked 3}, 300496Angelman syndrome, 105830Mental retardation, X-linked syndromic, Lubs type, 300260;RETT SYNDROME (RTT)[	Intellectual disability	X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease
MECP2	MENTAL RETARDATION SYNDROMIC X-LINKED LUBS TYPE 300260;MENTAL RETARDATION SYNDROMIC X-LINKED TYPE 13 300055;CHROMOSOME XQ28 DUPLICATION SYNDROME 300815;ENCEPHALOPATHY NEONATAL SEVERE DUE TO MECP2 MUTATIONS 300673;RETT SYNDROME (RTT)[312750	DDG2P	X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease
MECP2	ENCEPHALOPATHY NEONATAL SEVERE DUE TO MECP2 MUTATIONS;MENTAL RETARDATION SYNDROMIC X-LINKED TYPE 13;MENTAL RETARDATION SYNDROMIC X-LINKED LUBS TYPE;CHROMOSOME XQ28 DUPLICATION SYNDROME;RETT SYNDROME (RTT)[	Fetal anomalies	X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease