

No.	Phenotype	Genes	Chromosome	Cytogenetics	OMIM
1	1q21.1 deletion/duplication syndrome (susceptibility locus for neurodevelopmental disorders)			1 1q21.1 deletion/duplication	612474
2	1p36 deletion; Monosomy 1p36 syndrome (Facial Clefting Anomalies, Generalized epilepsy with febrile seizures-3 plus (type 5), Cranial Suture Closure, Seizures)	DVL1, SKI, GABRD, MMP23, KCNAB2		1 1p36 deletion and/or duplication/triplication	607872
3	1q41q42 deletion	DISP1		1 1q41 deletion	
4	Autism/Schizophrenia 9 (SCZD9)	DISC1		1 1q42.2 deletion	
5	Alagille syndrome 2 (ALGS2)	NOTCH2		1 1p12-p11.2	610205
6	Bartter syndrome type 3	CLCNKB		1 1p36.13 deletion	607364
7	Bartter syndrome type 4 (infantile with sensorineural deafness)	CLCNKA, CLCNKB		1 1p36.13	602522
8	Bartter syndrome type 4 (infantile with sensorineural deafness)	BSND		1 1p32.3	602522
9	Down syndrome, susceptibility to	MTR		1 1q43	190685
10	Epilepsy, nocturnal frontal lobe (type 3)	CHRN2		1 1q21.3 deletion	
11	Fryns syndrome	DISP1 candidate		1 1q41 microdeletion/duplication	229850
12	Glucose transport defect / Epilepsy, autosomal dominant	SLC2A1 (GLUT1)		1 1p34.2 deletion	606777
13	Heterotaxy	LEFTY2 (EBAF)		1 1q42.12 deletion	
14	Macrocephaly/seizures	SYT14		1 1q32.2 deletion	
15	Maple syrup urine disease, classic/intermediate, type II	DBT		1 1p31	248610
16	Microcephaly/agenesis corpus callosum	AKT3		1 1q44 deletion	
17	Mitochondrial complex I deficiency; NADH-coenzyme Q reductase deficiency (nuclear-encoded)	NDUFS2		1 1q23	252010
18	Muscle-eye-brain disease	POMGNT1		1 1p34.1 deletion	
19	NFIA haploinsufficiency	NFIA		1 1p31.3 deletion	600727
20	Schwartz-Jampel syndrome, type1 (SJS1)	HSPG2		1 1p36.12 deletion	
21	Short stature, pituitary and cerebellar defects, & small sella turcica	LHX4		1 1q25.2 deletion	606606

22	Stickler syndrome, type II (STL2)	COL11A1	1	1p21.1 deletion	604841
23	Thrombocytopenia-absent radius syndrome (TAR)	HFE2, LIX1L, PIAS3, ANKRD35, ITGA10, RBM8A, PEX11B, POLR3GL, TXNIP, GNRR2	1	1q21.1 deletion	274000
24	Van der Woude syndrome (VWS)	IRF6	1	1q32.2 deletion	119300
25	2p15-p16.1 microdeletion		2	2p15-p16.1 deletion	
26	2q22.3q23.3/mental retardation, autosomal dominant 1	MBD5, EPC2, KIF5C	2	2q23.1 deletion	156200
27	2q32.2-2q33 microdeletion syndrome	SATB2	2	2q32.2-q33.1 deletion	612313
28	2q35/brachydactyly type A1	IHH	2	2q35	600726
29	2q37 deletion/brachydactyly-MR/obesity/Albright hereditary osteodystrophy	CENTG2, GPC1, GPR35, KIF1A (ATSV), STK25	2	2q37.3 deletion	600430
30	Autism susceptibility locus	SLC4A10	2	2q24.2	605556
31	Autism susceptibility locus	SLC25A12	2	2q31	603667
32	Autism susceptibility locus	DPP10	2	2q14.1 deletion	
33	Autism susceptibility locus	NRXN1	2	2p16.3 deletion	209850
34	Cleft palate, isolated (CPI)	SATB2	2	2q33.1 deletion	608148
35	Ectodermal dysplasia, anhidrotic	EDAR	2	2q13 deletion	
36	Epilepsy, juvenile myoclonic	CACNB4	2	2q22-q23 deletion	
37	Feingold syndrome	MYCN	2	2p24.3 deletion	164280
38	Heterotaxy, visceral 2	CFC1	2	2q21.1 deletion	
39	holoprosencephaly 2(HPE2)	SIX3	2	2p21 deletion	157170
40	Holoprosencephaly 6 (HPE6)		2	2q37.1q37.3 deletion	605934
41	Holoprosencephaly 9 (HPE9)/Pituitary anomalies with holoprosencephaly	GLI2	2	2q14.2 deletion	
42	Hypertension with CHD	BMPR2	2	2q33.1 deletion	
43	Hypotonia-Cystinuria syndrome/ with mitochondrial disease	PPM1B, SLC3A1, PREPL	2	2p21 deletion	
44	Mitochondrial complex I deficiency; NADH-coenzyme Q reductase deficiency (nuclear-encoded)	NDUFS1	2	2q33-q34	252010
45	Mowat-Wilson syndrome	ZEB2 (ZFHX1B)	2	2q22.3 deletion	235730

46	Nephronophthisis 1/Related Joubert Syndrome 4 (NPHP1)	NPHP1	2	2q13 homozygous deletion	609583
47	Nievergelt syndrome	AFF3 (LAF4)	2	2q11.2	
48	Noonan syndrome (NS4)	SOS1	2	2p22.1 deletion	610733
49	Orofacial cleft	SUMO1	2	2q32.2-q33 deletion	
50	Pseudovaginal perineoscrotal hypospadias	SRD5A2	2	2p23.1	607306
51	Seizures	DGKD	2	2q37.1 deletion	
52	Seizures, benign familial neonatal-infantile	SCN2A	2	2q23-q24.3 deletion	
53	Severe myoclonic epilepsy of infancy (SMEI) or Dravet syndrome; Generalized epilepsy with febrile seizures plus; GEFS+ type 2	SCN1A	2	2q24.3 deletion	604233
54	Split-hand/foot malformation 5 (SHFM5)	DLX1, DLX2	2	2q31.1 deletion	606708
55	Synpolydactyly (SPD1); HOXD deletion syndrome	HOXD13	2	2q31-q32 deletion	186000
56	Waardenburg syndrome, type 1	PAX3	2	2q36.1 deletion	193500
57	3p25 deletion	CNTN4	3	3p25.3 deletion	
58	3q29 microdeletion/duplication syndrome; Autism	PAK2, DLG1	3	3q29 deletion	609425
59	Ataxia	ITPR1	3	3p26.2 deletion	
60	Ataxia teleangiectasia	ATR	3	3q23 deletion	
61	Bartter with autosomal dominant	CASR	3	3q21.1 deletion	
62	Blepharophimosis, ptosis, and epicanthus inversus (BPES)	FOXL2	3	3q22.3 deletion	110100
63	Cerebral cavernous malformations 3	PDCD10	3	3q26.1	603285
64	Dandy-Walker malformation (DWM)	ZIC4, ZIC1	3	3q24 deletion	220200
65	Epilepsy, juvenile myoclonic	CLCN2	3	3q27.1 deletion	
66	Familial hypocalciuric hypercalcemia 1 (HHC1)	CASR	3	3q21.1	145980
67	Forebrain defects	TDGF1 (CRIPTO)	3	3p21.31 deletion	187395
68	Heterotaxy, visceral 4	ACVR2B	3	3p22.2 deletion	602730
69	Loeys-Dietz syndrome, type 2B (LDS2B)/Marfan syndrome, type II (MFS2)	TGFBR2	3	3p24.1 deletion	609192
70	Microphthalmia syndromic 3 (MCOPS3)	SOX2	3	3q26.33 deletion	206900
71	Neonatal severe primary hypoparathyroidism (NSHPT)	CASR	3	3q21.1	239200

72	Noonan syndrome 5 (NS5)	RAF1	3	3p25.1 deletion	
73	Speech delay, contractures, hypotonia and blepharophimosis	FOXP1	3	3p14.1-p13 deletion	
74	Split hand/foot malformation 4 (SHFM4)	TP63 (TP73L)	3	3q28 deletion	605289
75	Von Hippel-Lindau syndrome (VHL)	VHL	3	3p25.3 deletion	
76	Waardenburg syndrome, type IIA (WS2A)	MITF	3	3p14.1 deletion	193510
77	Achondroplasia; ACH	FGFR3	4	4p16.3	100800
78	Action myoclonus-renal failure syndrome (AMRF)	SCARB2	4	4q21.1 deletion	254900
79	Autism associated with interstitial deletion on chromosome 4q32-q34	GLRB, GRIA2, NPY1R, NPY5R, GLRA3 candidates	4	4q32.1 deletion	
80	Microtia	DRD5	4	4p16.1 deletion	
81	Partial anodontia, orofacial clefting/Witkop syndrome	MSX1	4	4p16.2 deletion	142983
82	Rieger syndrome, type 1 (RIEG1)	PITX2	4	4q25 deletion	180500
83	Wolf-Hirschhorn syndrome (WHS)	LETM1, WFS1, WHSC1, WHSC2, WHCR	4	4p16.3 deletion	194190
84	5q14.3 deletion	MEF2C	5	5q14.3	
85	5q21q31 deletion syndrome		5	5q21q31 deletion	
86	Chronic pancreatitis	SPINK1	5	5q32 deletion	
87	Corneal dystrophies	TGFBI	5	5q31.2	601692
88	Cornelia de Lange syndrome (CDLS)	NIPBL	5	5p13.2 deletion	122470
89	Cranio metaphyseal dysplasia autosomal dominant (CMDD)	ANKH	5	5p15.2 deletion	
90	Craniosynostosis (CRS2)	MSX2	5	5q35.2 deletion	
91	Cri-du-Chat syndrome (cry/speech/face critical regions)	TERT, EGR1	5	5p15.2 -p13.3 deletion	123450
92	Familial adenomatous polyposis/Gardner syndrome	APC	5	5q22.2	175100
93	Generalized epilepsy with febrile seizures plus (GEFS+) type 3/ Epilepsy, childhood absence (CAE)	GABRG2	5	5q34 deletion	604233
94	Holoprosencephaly and preaxial polydactyly	FBXW11	5	5q35.1 microduplication	605651
95	Juvenile myoclonic epilepsy	GABRA1	5	5q34 deletion	

96	Kartagener syndrome; ciliary dyskinesia, primary, 3	DNAH5	5	5p15.2 deletion	608644
97	Leukodystrophy, adult onset autosomal dominant (ADLD)	LMNB1	5	5q23.2 deletion	169500
98	Microcephaly, congenital heart disease	NPM1	5	5q35.1 deletion	
99	Mitochondrial complex I deficiency; NADH-coenzyme Q reductase deficiency (nuclear-encoded)	NDUFS6	5	5pter-p15.33	252010
100	Mitochondrial complex I deficiency; NADH-coenzyme Q reductase deficiency (nuclear-encoded)	NDUFS4	5	5q11.1	252010
101	Mitochondrial complex I deficiency; NADH-coenzyme Q reductase deficiency (nuclear-encoded)	NDUFAF2 (NDUFA12L)	5	5q12.1	252010
102	Monogenic audiogenic seizure susceptibility; Febrile convulsions, familial, 4	GPR98 (MASS1, VLGR1)	5	5q14.3 deletion	604352
103	Sotos syndrome	NSD1	5	5q35.3 deletion	117550
104	Tetralogy of Fallot / ASD secundum with atrioventricular conduction defects	NKX2-5 (CSX)	5	5q35.2 duplication	108900
105	Treacher-Collins-Franceschetti syndrome (TCOF)	TCOF1	5	5q33.1 deletion	154500
106	6q24.3q25.1(Diabetes mellitus, insulin-dependent, 5; IDDM5)	SUMO4	6	6q25.1	600320
107	Adrenal hyperplasia, congenital (CAH) due to 21-alpha hydroxylase deficiency	CYP21A2	6	6p21.32 deletion	119600
108	Arthropathy, progressive pseudorheumatoid, of childhood; (PPAC)	WISP3	6	6q21 deletion	208230
109	Autism/Joubert syndrome 3	AHI1	6	6q23.3	608629
110	Autism susceptibility locus (MRT6)	GRIK2	6	6q16.3	611092
111	Autism susceptibility locus	GLO1	6	6p21.3-p21.2 deletion	209850
112	Char syndrome	TFAP2B	6	6p12.3 deletion	
113	Cleft lip/palate	ESR1	6	6q25.1 deletion	
114	Cleidocranial dysplasia (CCD)	RUNX2	6	6p12.3 deletion	119600

115	Diabetes mellitus, transient neonatal 1; Transient neonatal diabetes with hypomethylation	PLAGL1 (ZAC)	6	6q24.2 paternal deletion	601410
116	Epilepsy, juvenile absence (JAE) juvenile myoclonic (EJM1)	PAQR8, EFHC1	6	6p12.2 deletion	
117	Epilepsy, progressive myoclonus	NHLRC1	6	6p22.3 deletion	
118	Iridogoniodysgenesis anomaly, Axenfeld-Rieger syndrome, 6p25.3 deletion syndrome	FOXC1 (FKHL7)	6	6p25.3 deletion	601631
119	Maple syrup urine disease, classic, type IB	BCKDHB	6	6q14	248611
120	Mitochondrial complex I deficiency; NADH-coenzyme Q reductase deficiency (nuclear-encoded)	NDUFAF4 (HRPAP20)	6	6q16.1	252010
121	Myoclonic epilepsy of Lafora	EPM2A	6	6q24.3 deletion	
122	Myoclonic epilepsy, juvenile/juvenile absence epilepsy	BRD2	6	6p21.32 deletion	
123	Prader-Willi syndrome-like/Obesity	SIM1	6	6q16.3 deletion	176270
124	Sacral/anorectal malformation syndrome	SYNJ2, SERAC1, GTF2H5, TULP4	6	6q25.3 deletion	
125	Vascular endothelial growth factor (VEGF)-related disorders	VEGFA	6	6p21.1 deletion	192240
126	7q11.23 duplication (WBS region)		7	7q11.23 duplication	609757
127	7q21.13q22.1 deletion/Ectrodactyly/Deafness	FZD1, CUX1	7	7q22.13q22.1 deletion	
128	Acheiropody/ and preaxial polydactyly	LMBR1	7	7q36.3 deletion	
129	Autism susceptibility locus (AUTS10)	EN2	7	7q36.3	611016
130	Autism susceptibility locus	CADPS2	7	7q31.32	609978
131	Autism susceptibility locus (AUTS9)	MET	7	7q31.2	611015
132	Autism susceptibility locus	NPTX2	7	7q22.1	
133	Autism susceptibility locus	DLX5	7	7q21.3 deletion	
134	Autism susceptibility locus	DPP6	7	7q36.2 deletion	
135	Cerebral cavernous malformations 1	KRIT1	7	7q11.2-q21	116860
136	Cerebral cavernous malformations 2	CCM2	7	7p13 deletion	603284
137	Currarino syndrome (Sacral agenesis)	MNX1 (HLXB9)	7	7q36.3 deletion	176450
138	Epilepsy	KCND2	7	7q31.31 deletion	

139	Greig cephalo-polysyndactyly syndrome (GCPS)	GLI3	7	7p14.1 deletion	175700
140	Hand-foot-uterus syndromes	HOXA13	7	7p15.2 deletion	
141	Holoprosencephaly 3 (HPE3)	SHH	7	7q36.3 deletion	142945
142	Lissencephaly/Autism /multifocal epilepsy	RELN	7	7q22.1 deletion	257320
143	Maple syrup urine disease, type III	DLD	7	7q31.1	238331
144	Saethre-Chotzen syndrome (SCS)	TWIST1	7	7p21.1 deletion	101400
145	Schizophrenia & epilepsy	CNTNAP2	7	7q36.1 deletion	
146	Speech delay/Autism	FOXP2	7	7q31.1 deletion	602081
147	Split hand/foot malformation with sensorineural hearing loss (SHFM1D)		7	7q21.2-q21.3 deletion	220600
148	Split-hand/foot malformation 1 (SHFM1)	SHFM1 (DSS1)	7	7q21.3	183600
149	Triphalangeal thumb Syndrome (Polydactyly, preaxial II; PPD2)	LMBR1 (the ZRS in intron 5)	7	7q36.3 duplication	174500
150	Williams-Beuren syndrome (WBS)	ELN, LIMK1, RFC2, CYLN2	7	7q11.23 deletion	194050
151	8p23.1 Inverted duplication/microdeletion	GATA4 candidate	8	8p22p23 inversion/duplication/deletion	
152	Atrial septal defect 2	GATA4	8	8p23.1 deletion	607941
153	Bipolar disorder	IMPA1	8	8q21.13 deletion	
154	Birk-Barel syndrome	KCNK9	8	8q24.3	
155	Branchiootorenal dysplasia syndrome (BOR)/Otofaciocervical (OFC)/Melnick-Fraser	EYA1	8	8q13.3 deletion	113650
156	Ceroid lipofuscinosis, neuronal 8, northern epilepsy variant	CLN8	8	8p23.3 deletion	
157	CHARGE syndrome	CHD7	8	8q12.2 deletion	214800
158	Diaphragmatic hernia 2, DIH2	SOX7, GATA4	8	8p23.1 deletion	222400
159	Epilepsy, Benign neonatal (EBN2)	KCNQ3	8	8q24.22 deletion	
160	Epilepsy, nocturnal frontal lobe (type 4)	CHRNA2	8	8p21.2 deletion	
161	Kabuki syndrome		8	8p23.1-p22 ??	147920
162	Langer-Giedion syndrome (LGS)/trichorhinophalangeal syndrome type II (TRPS2)	TRPS1, EXT1	8	8q23.3-q24.11 deletion	150230
163	Nablus mask-like facial syndrome		8	8q21.3-q22.1 microdeletion	608156
164	Tetralogy of Fallot/diaphragmatic hernia	ZFPM2 (FOG2)	8	8q23.1 deletion	

165	Trichorhinophalangeal syndrome I (TRPS1)	TRPS1	8	8q24.12 deletion	190350
166	9p deletion; Monosomy 9p syndrome	CER1 candidate	9	9p22.3-p23	158170
167	9q22.32-q22.33 Microdeletion	TGFBR1 candidate	9	9q22.33 deletion	
168	9q34.3 subtelomeric deletion syndrome	NOTCH1, EHMT1	9	9q34.3 deletion	610253
169	Autism susceptibility locus	CNTNAP3	9	9p13.1 deletion	
170	Basal cell nevus syndrome (BCNS) / Gorlin syndrome (GS)	PTCH1	9	9q22.32 deletion	109400
171	Ehlers-Danlos syndrome, type II	COL5A1	9	9q34.2-q34.3 deletion	130010
172	Holoprosencephaly 7 (HPE7)	PTCH1	9	9q22.32 missense gain-of-function mutations /duplication? duplications?	610828
173	Kartagener syndrome; ciliary dyskinesia, primary, 1	DNAI1	9	9p13.3 deletion	244400
174	Loeys-Dietz syndrome (LDS)	TGFBR1	9	9q22.33 deletion	609192
175	Nail-patella syndrome (NPS)	LMX1B	9	9q33.3 deletion	161200
176	Neuromotor development delay, cerebellar ataxia, and epilepsy	MLLT3 (AF9)	9	9p21.3 deletion	159558
177	Overgrowth syndrome	GABBR2 (GPR51), TGFBR1	9	9q22.33 interstitial microdeletion	607340
178	Ovotestes and male-to female XY sex-reversal, +/- adrenal failure	NR5A1 (SF1)	9	9q33.3 deletion	184757
179	Robinow syndrome/brachydactyly type B1 (BDB1)	ROR2	9	9q22.31 deletion	
180	Sex reversal, autosomal dominant 2 (SRA2)		9	9p24.3 deletion	154230
181	Telangiectasia, hereditary hemorrhagic, of rendu, osler, and weber; (HHT)	ENG	9	9q34.11 deletion	
182	Tuberous sclerosis 1	TSC1	9	9q34.13	191100
183	Walker-Warburg syndrome	POMT1	9	9q34.13 deletion	
184	10q22q23 deletion	NRG3, GRID1	10	10q22q23 deletion	
185	10q26 deletion syndrome		10	10q26 deletion	609625
186	Anterior segment mesenchymal dysgenesis (ASMD)/ Cataract	PITX3	10	10q24.32 deletion	
187	Autism susceptibility locus	JMJD1C (TRIP8), REEP3	10	10q21.3 deletion	
188	Bannayan-Riley-Ruvalcaba syndrome (BRRS)	PTEN	10	10q23.31 microdeletion	153480

189	Cleft lip/palate	FGFR2	10	10q26.13 deletion	
190	Cowden Disease (CD)	PTEN	10	10q23.31 microdeletion	158350
191	DiGeorge syndrome 2 (DGS2)	NEBL	10	10p14 deletion	601362
192	Epilepsy, lateral temporal lobe, autosomal dominant (ADLTE)	LGI1	10	10q23.33 deletion	
193	Hirschsprung disease	RET	10	10q12.11 deletion	
194	Hypoparathyroidism, sensorineural deafness, and renal disease (HDR)	GATA3	10	10p14 deletion/duplication	146255
195	Idiopathic dilated cardiomyopathy	NEBL candidate	10	10p12.31 deletion	605491
196	Juvenile polyposis syndrome (JPS), BMPR1A-related	BMPR1A	10	10q23.2 microdeletion	174900
197	Macrocephaly/autism syndrome	PTEN	10	10q23.31	605309
198	Papillorenal syndrome (Renal-coloboma syndrome)	PAX2	10	10q24.31 deletion	
199	Proteus/Proteus-like syndrome	PTEN	10	10q23.31	176920
200	Schizencephaly / multifocal epilepsy	EMX2	10	10q26.11 deletion	269160
201	Split-hand/foot malformation 3 (SHFM3)	FBXW4 (SHFM3)	10	10q24.32 deletion	600095
202	11q11–q13.3 duplication/multiple craniosynostoses, congenital heart defect	FGF4, FGF3	11	11q13.3 duplication	
203	Aniridia II (AN2)	PAX6	11	11p13 deletion	106210
204	Autism susceptibility locus	BDNF	11	11p14.1 deletion	
205	Autism susceptibility locus	NRXN2	11	11q13.1 deletion	
206	Bartter syndrome type 2	KCNJ1	11	11q24.3 deletion	241200
207	Beckwith-Wiedemann syndrome (BWS)	H19, IGF2, KCNQ1, CDKN1C	11	11p15.4/15.5 deletion/duplication	130650
208	CALC1, behavioral problems and autistic spectrum disorder	CALCA, CALCB	11	11p15.2 deletion	
209	Craniosynostosis	SOX6	11	11p15.2 deletion	
210	Jacobsen syndrome (JBS)/ Trigenocephaly 11q deletion		11	11q24.1 deletion (11q23.3-qter)	147791
211	Leukodystrophy due to mitochondrial complex 1 deficiency	NDUFV1	11	11q13.2 deletion	252010
212	Leukodystrophy with microdeletion 11q14.3		11	11q14.2q14.3 deletion	

213	Myoclonic epilepsy, neonatal, with suppression-burst pattern	SLC25A22	11	11p15.5 deletion	
214	Oto-dental syndrome (dental and inner-ear disease/ocular coloboma)	FGF3, FADD	11	11q13.3 deletion	
215	Potocki-Shaffer syndrome	ALX4, EXT2	11	11p11.2 deletion	601224
216	Smith-Lemli-Opitz syndrome (SLOS)	DHCR7	11	11q13.4 deletion	270400
217	Wilms tumor 1 (WT1)	WT1	11	11p13 deletion	194070
218	Wilms tumor -aniridia-genitourinary anomalies-mental retardation-and obesity syndrome (WAGRO)	BDNF, PAX6, WT1	11	11p13-p12 deletion	612469
219	Wilms tumor -aniridia genitourinary anomalies-mental retardation syndrome (WAGR)	PAX6, WT1	11	11p13 deletion	194072
220	12q14 microdeletion syndrome	HMGA2, GRIP1, LEMD3	12	12q14.3 deletion	
221	12q24.21-q24.23 microduplication	MED13L, NOS1, RFC5	12	12q24.21-q24.23 duplication	
222	Buschke-Ollendorff syndrome / Osteopoikilosis, short stature and mental retardation	LEMD3	12	12q14.3 deletion	166700
223	Chondrodysplasias (Stickler syndrome/Spondyloepiphyseal dysplasia)	COL2A1	12	12q13.11 deletion	108300
224	Episodic ataxia (EA1), partial epilepsy	KCNA1	12	12p13.32 deletion	
225	Holt-Oram syndrome	TBX5, TBX3	12	12q24.21 microdeletion	142900
226	Joubert syndrome 5	CEP290	12	12q21.32	610188
227	Leber congenital amaurosis X (LCAX)	CEP290	12	12q21.32	610142
228	Meckel syndrome, type 4	CEP290	12	12q21.32	611134
229	Noonan syndrome (NS1)	PTPN11	12	12q24.13 duplication	163950
230	Osler-Rendu-Weber syndrome 2; ORW2 (Hereditary hemorrhagic teleangiectasia type 2)	ACVRL1 (ALK1)	12	12q13.13 deletion	600376
231	Pallister-Killian syndrome 12p triplication		12	12p triplication	601803
232	Senior-Loken syndrome 6	CEP290	12	12q21.32	610189
233	Timothy syndrome	CACNA1C	12	12p13.33 deletion	
234	TSC2 angiomyolipomas, renal, modifier of	IFNG	12	12q14	191100
235	Ulnar-mammary syndrome	TBX3	12	12q24.21 deletion/duplication	181450

236	13q33q34 deletion/ genital malformation in males/Microcephaly/MR	EFNB2, ARHGEF7	13	13q33.3q34 deletion	
237	Autism susceptibility locus	NUFIP1	13	13q14.12 deletion	
238	Autism susceptibility locus	PCDH9	13	13q21.32 deletion	
239	Brachydactyly and other skeletal anomalies	GPC5, GPC6	13	13q31.3 deletion	
240	Hirschsprung disease (HSCR2)	EDNRB	13	13q22.3 deletion	600155
241	Holoprosencephaly 5 (HPE5)	ZIC2	13	13q32.3 deletion	609637
242	Pancreatic agenesis, congenital	PDX1 (IPF1)	13	13q12.2 deletion	260370
243	Retinoblastoma (RB1)	RB1	13	13q14.2 deletion	180200
244	Shah-Waardenburg syndrome	EDNRB	13	13q22.3 deletion	
245	Wilms tumor	BRCA2	13	13q12.3	194070
246	14q11.2 deletion syndrome	SUPT16H, CHD8	14	14q11.2 deletion	
247	14q12 deletion syndrome	FOXP1 (FOXP1B)	14	14q12 deletion	164874
248	14q22-q23 deletion	OTX2	14	14q22.1-q23.1 deletion	600037
249	Autism susceptibility locus	NRXN3	14	14q31.1 deletion	
250	Branchiootic syndrome-3 (BOS3)	SIX1	14	14q23.1 deletion	
251	Holoprosencephaly 8 (HPE8)		14	14q13.1-q13.2 microdeletion	609408
252	Microphthalmia with cataract 2 (MCOPCT2)	SIX6	14	14q23.1	212550
253	Microphthalmia syndromic 6 (MCOPS6)/Anophthalmia, pituitary hypoplasia, and ear anomalies	BMP4	14	14q22.2 deletion	
254	Oculopharyngeal muscular dystrophy	PABPN1	14	14q11.2	
255	Pituitary hypoplasia, Oculoauriculovertebral spectrum	SIX6	14	14q23.1(14q22.2-q22.3 del)?	
256	Tooth agenesis, selective 3 (STHAG3) Hypodontia/Oligodontia 3	PAX9	14	14q13.3 deletion	
257	15q11-q13 duplication syndrome, autistic features	GABRB3 candidate	15	15q11.2-q13.1 maternal duplication	608636
258	15q13.3 microdeletion syndrome	CHRNA7 candidate	15	15q13.1-q13.3 deletion	612001
259	15q21 microdeletion syndrome		15	15q21.1q21.2 deletion	
260	15q24 recurrent microdeletion syndrome		15	15q24.1-q24.3 deletion	
261	15q26 overgrowth syndrome	IGF1R	15	15q26.3 duplication	

262	15q26-qter deletion, Severe IUGR, developmental delay, postnatal growth retardation	IGF1R	15	15q26.3 deletion	612626
263	Angelman syndrome (AS)	UBE3A, AS-SRO	15	15q11.2-q12 deletion	105830
264	Angelman syndrome (AS) imprinting center	AS imprinting center	15	15q11.2 deletion	
265	Bartter syndrome, antenatal type 1	SLC12A1	15	15q21.1 deletion	146200
266	BP1-BP2 breakpoint region in 15q11.2	TUBGCP5, NIPA1, NIPA2, CYFIP1	15	15q11.2 microdeletion	
267	Cardiomyopathy, familial hypertrophic/Congenital heart defect	ACTC1	15	15q14 deletion	612098
268	Cleft plate, congenital heart defect	GREM1, GJD2, MEIS2	15	15q13.3 deletion	
269	Diaphragmatic hernia, congenital	NR2F2, CHD2	15	15q26.1-q26.2 deletion	142340
270	Epilepsy, juvenile myoclonic	CHRNA7	15	15q13.3 deletion	
271	Epilepsy, nocturnal frontal lobe, type2/Autosomal-dominant nocturnal frontal lobe epilepsy (ADNFLE)	CHRNA5, CHRNA3, CHRNB4	15	15q25.1 deletion	
272	Johanson-Blizzard syndrome (JBS)	UBR1	15	15q15.2 deletion	243800
273	Marfan syndrome, type I (MFS1)	FBN1	15	15q21.1 deletion	154700
274	Mental retardation	ARHGAP11A, CHRM5	15	15q13.3 deletion	
275	Oculocutaneous albinism 2	OCA2	15	15q13.1 microdeletion	203200
276	Prader –Willi syndrome (PWS)	PWS-SRO, SNRPN, HBII-85 C/D box, NDN, all snoRNAs, snoRNA cluster	15	15q11.2-q12 deletion	176270
277	Prader –Willi syndrome (PWS) imprinting center	PWS imprinting center	15	15q11.2 deletion	
278	16p11.2 microdeletion/duplication (AUTS14)		16	16p11.2	611913
279	16p11.2-p12.2 microdeletion syndrome		16	16p12.1 deletion	
280	16p13.11 recurrent microdeletion predisposing to autism and/or mental retardation		16	16p13.11 microdeletion/duplication	
281	16p13.3 deletion; Rubinstein-Taybi syndrome, severe	DNASE1, TRAP1, CREBBP	16	16p13.3 deletion	610543
282	Alpha thalassemia-MR syndrome (ATR-16)	HBA1, HBA2	16	16p13.3 deletion	141750

283	Autism susceptibility locus	A2BP1	16	16p13.2 deletion	
284	Bilateral frontoparietal polymicrogyria/focal or multifocal epilepsy	GPR56	16	16q13 deletion	606854
285	Epilepsy, idiopathic generalized (EIG6)	CACNA1H	16	16p13.3 deletion	
286	Gitelman syndrome	SLC12A3	16	16q13	263800
287	Lymphedema hereditary, II	FOXC2	16	16q24.1 deletion	
288	Multicentric osteolysis, nodulosis, and arthropathy (MONA)	MMP2	16	16q12.2 deletion	
289	Polycystic kidney disease	PKD1	16	16p13.3	601313
290	Polycystic kidney disease, infantile severe, with tuberous sclerosis	TSC2, PKD1	16	16p13.3 deletion	600273
291	Pseudoxanthoma elasticum, forme fruste	ABCC6	16	16p13.11 deletion	
292	Rubinstein-Taybi syndrome (RSTS)	CREBBP	16	16p13.3	180849
293	Townes-Brocks syndrome	SALL1	16	16q12.1 deletion	107480
294	Tuberous sclerosis 2	TSC2	16	16p13.3	191100
295	17q21.31 microdeletion/microduplication syndrome	MAPT, CRHR1	17	17q21.31 deletion	610443
296	Andersen syndrome	KCNJ2	17	17q24.3 deletion	170390
297	Autism susceptibility locus (AUTS7)	ITGB3	17	17q21.32	610676
298	Autism susceptibility locus	SLC6A4	17	17q11.2	182138
299	Autism susceptibility locus	DLG4	17	17p13.1 deletion	
300	Autism susceptibility locus	NUFIP2	17	17q11.2 deletion	
301	Autism susceptibility locus	CNTNAP1	17	17q21.31 deletion	
302	Campomelic dysplasia	SOX9	17	17q24.3 deletion	114290
303	Canavan disease	ASPA	17	17p13.3 deletion	
304	Charcot-Marie-Tooth disease type 1A(CMT1A)	PMP22	17	17p12 duplication	118220
305	Cystinosis, Nephropathic (CTNS)	CTNS	17	17p13.2-p13.3 deletion	219800
306	Hereditary neuropathy with pressure palsies (HNPP)	PMP22	17	17p12 deletion	162500
307	Li-Fraumeni 1 (LFS)	TP53	17	17p13.1	151623
308	Lissencephaly 1	PAFAH1B1 (LIS1)	17	17p13.3 deletion	607432
309	Miller-Dieker lissencephaly syndrome (MDLS)	PAFAH1B1 (LIS1), YWHAE	17	17p13.3 deletion	247200
310	Neurofibromatosis I (NF1)	NF1	17	17q11.2 deletion	162200
311	Osteogenesis imperfecta type IV	COL1A1	17	17q21.33 deletion	166220

312	Osteogenesis imperfecta congenita; type IIA	COL1A2	17	17q21.33 deletion	166210
313	Potocki-Lupski syndrome (PTLS)	RAI1	17	17p11.2 duplication	610883
314	Renal cysts and diabetes (RCAD)/epilepsy	HNF1B (TCF2)	17	17q12 deletion/duplication	137920
315	Smith-Magenis syndrome (SMS)	RAI1	17	17p11.2 deletion	182290
316	Symphalangisms proximal, Multiple synostoses syndrome 1, Stapes ankylosis with broad thumb and toes	NOG	17	17q22 deletion	186500
317	Van Buchem disease(Hyperostosis corticalis generalisata)	SOST and downstream non coding region	17	17q21.31	239100
318	18p deletion syndrome		18	18p11.1-p11.3 deletion	146390
319	18q deletion syndrome		18	18q22.3-q23 deletion	601808
320	Dyggve-Melchior-Clausen syndrome (DMC) (autosomal recessive)	DYM	18	18q21.1 deletion/duplication	
321	Holoprosencephaly 4 (HPE4)	TGIF1	18	18p11.31 deletion	142946
322	Juvenile polyposis syndrome (JPS), MADH4-related	SMAD4 (MADH4)	18	18q21.2 microdeletion	174900
323	Pitt-Hopkins syndrome	TCF4	18	18q21.2 deletion	610954
324	Susceptibility to adolescent-onset idiopathic generalized epilepsy	ME2	18	18q21.2 deletion	154270
325	CADASIL (Cerebral-Arteriopathy, Autosomal Dominant, with Subcortical Infarcts and Leukoencephalopathy)	NOTCH3	19	19p13.12 deletion	125310
326	Epilepsy idiopathic generalized/ Migraine/Spinocerebellar ataxia 6	CACNA1A	19	19q21.2 deletion	
327	Familial hypercholesterolemia, autosomal dominant	LDLR	19	19p13.2	143890
328	Generalized epilepsy with febrile seizures- plus type 1	SCN1B	19	19q13.11 deletion	604233
329	Maple syrup urine disease, classic, type IA	BCKDHA	19	19q13.2	608348
330	Mitochondrial complex I deficiency; NADH-coenzyme Q reductase deficiency (nuclear-encoded)	NDUFA11	19	19p13.3	252010

331	Persistent Mullerian duct syndrome (PMDS), type I /Sex ambiguity	AMH (MIS)	19	19p13.3 deletion	261550
332	Pseudoachondroplasia (PSACH)	COMP	19	19p13.11 deletion	177170
333	Spondylocostal dysostosis autosomal recessive; (SCDO1)	DLL3	19	19q13.2 deletion	277300
334	20q13.33 deletion/Autosomal-dominant nocturnal frontal lobe epilepsy, Benign familial neonatal convulsions (type 1), hypotrichosis–lymphedema–telangiectasia	ARFGAP1, CHRNA4, KCNQ2, SOX18	20	20q13.33 deletion	
335	Alagille syndrome (AGS)	JAG1	20	20p12.2 deletion	118450
336	Brachydactyly, type C (BDC)	GDF5	20	20q11.22 deletion	113100
337	Chondrodysplasia, grebe tybe	GDF5 (CDMP1)	20	20q11.22 deletion	200700
338	Duane radial ray syndrome	SALL4	20	20q13.2	607323
339	Immunodeficiency-centromeric Instability-Facial anomalies syndrome	DNMT3B	20	20q11.21 deletion	
340	McCune-Albright syndrome	GNAS	20	20q13.32 deletion	174800
341	McKusick-Kaufman syndrome (MKKS)	MKKS	20	20p12.2 deletion	236700
342	Mitochondrial complex I deficiency; NADH-coenzyme Q reductase deficiency (nuclear-encoded)	C20orf7	20	20p12.1	252010
343	Shah-Waardenburg syndrome	EDN3	20	20q13.32 deletion	277580
344	Susceptibility to attention-deficit hyperactivity disorder (ADHD)	SNAP25	20	20p12.2 deletion	600322
345	Wolff-Parkinson-White	BMP2	20	20p12.3 deletion	
346	Alzheimer disease, early onset with cerebral amyloid angiopathy	APP	21	21q21.3 microdeletion/duplication	104300
347	Blepharophimosis/vormian hypoplasia/exotropia/DD	POFUT2	21	21q22.3 deletion	
348	Down syndrome critical regions	RCAN1 (DSCR1)	21	21q22.12	190685
349	Down syndrome critical regions	PSMG1 (DSCR2), DSCR3, DSCR4, PIGP, DSCR8, DSCR9, DSCR10	21	21q22.2-q22.3	190685
350	Epilepsy, juvenile absence (JAE)	GRIK1	21	21q21.3 deletion	138245
351	Holoprosencephaly 1 (HPE1)	TRAPPC10 (TMEM1), LSS	21	21q22.3 deletion	236100

352	22q11.2 duplication syndrome reciprocal to DGS deletion	TBX1	22	22q11.2 duplication	608363
353	22q11.2 distal microdeletion syndrome	BCR	22	22q11.2 deletion	
354	22q13.3 deletion/Autism (Phelan-Mcdermid syndrome)	SHANK3, ARSA	22	22q13.33 deletion	606232
355	Cat eye syndrome (CES)	CECR1, CECR5, CECR6, CECR7, CRKL	22	inv dup(22)(q11.2)	115470
356	DiGeorge/velocardiofacial syndrome DGS1/VCFS / Autism	GNB1L, TBX1	22	22q11.2 deletion	188400
357	Metachromatic leukodystrophy	ARSA	22	22q13.33 deletion	250100
358	Neurofibromatosis II (NF2)	NF2	22	22q12.2 deletion	101000
359	Rubinstein-Taybi syndrome (RSTS)	EP300	22	22q13.2	180849
360	Waardenburg-Shah syndrome	SOX10	22	22q13.1 deletion	277580
361	Xp11.3 deletion syndrome/Retinitis pigmentosa X-linked RP2/ XLMR	ZNF674, RP2	X	Xp11.3 deletion	300578
362	Xp11.3 deletion with mental retardation (MRX92)	ZNF674	X	Xp11.3 deletion	300573
363	Adrenal hypoplasia, congenital (AHC)	NROB1 (DAX1)	X	Xp21.2 deletion	300200
364	Adrenoleukodystrophy; (ALD)	ABCD1	X	Xq28 deletion	300100
365	Albinism, ocular type 1	GPR143 (OA1)	X	Xp22.2 deletion	300500
366	Allan-Herndon-Dudley syndrome, X-linked	SLC16A2	X	Xq13.2 deletion	300523
367	Alport syndrome, X-linked (ATS)	COL4A5	X	Xq22.3 deletion	301050
368	Androgen insensitivity syndrome (AIS)	AR	X	Xq12 deletion	300068
369	ATRX, XLMR-Hypotonic facies syndrome, ATRX, and others	ATRX	X	Xq21.1 deletion/duplication	
370	Autism susceptibility locus	ASMT	X	Xp22.33 deletion	300015
371	Autism susceptibility locus	ASMTL	X	Xp22.33 deletion	300162
372	Autism / Asperger syndrome-1, X-linked; AUTSX1	NLGN3	X	Xq13.1 deletion	300425
373	Autism, X-linked, susceptibility to, AUTSX2 (XLMR)	NLGN4X	X	Xp22.32 deletion	300495
374	Autism, X-linked, susceptibility to, AUTSX3 (XLMR)	MECP2	X	Xq28 deletion	300496

375	Bipolar disorder/Bipolar affective disorder susceptibility locus	VAMP7 (SYBL1)	X	Xq28 deletion	300053
376	Börjeson-Forssman-Lehmann syndrome	PHF6	X	Xq26.2 deletion	301900
377	Brunner syndrome/Monoamine oxidase-A deficiency/Antisocial behavior following childhood maltreatment	MAOA	X	Xp11.3 deletion	
378	Bruton agammaglobulinemia tyrosine kinase (X-linked)	BTK	X	Xq22.1 deletion	300300
379	Cerebellar hypoplasia	OPHN1	X	Xq12 deletion	
380	Charcot-Marie-Tooth disease, X-linked, 1 (CMTX1)	GJB1	X	Xq13.1	302800
381	Chondrodysplasia punctata, X-linked recessive	ARSE (CDPX1)	X	Xp22.33 deletion	302950
382	Choroideremia	CHM	X	Xq21.2 deletion	
383	Coffin-Lowry syndrome	RPS6KA3 (RSK2)	X	Xp22.12 deletion	303600
384	Cornelia de Lange syndrome (CDLS), X-linked	SMC1A (SMC1L1)	X	Xp11.22 deletion	300590
385	Craniofrontal dysplasia; CFND	EFNB1	X	Xq11.2q13.1	304110
386	Creatine deficiency syndrome / X-linked mental retardation (XLMR)	SLC6A8	X	Xq28 deletion	300352
387	Danon disease	LAMP2	X	Xq24 deletion	300257
388	Deafness, X-linked 2 (DNFX2)	POU3F4	X	Xq21.1 deletion	
389	Dent disease 1	CLCN5	X	Xp11.22 deletion	300009
390	Diabetes insipidus, nephrogenic, X-linked	AVPR2	X	Xq28 deletion	304800
391	Dosage sensitive sex reversal (DSS)	NR0B1 (DAX1)	X	Xp21.2 duplication	300018
392	Down syndrome, transient myeloproliferative disorder and megakaryoblastic leukemia	GATA1	X	Xp11.23	190685
393	Duchenne/Becker muscular dystrophy (large del/dup)	DMD	X	Xp21.2 deletion/duplication	310200
394	Dyskeratosis congenita, X-linked	DKC1	X	Xq28 deletion	
395	Ectodermal dysplasia, X-linked hypohidrotic	EDA	X	Xq13.1 deletion	
396	Epilepsy	EFHC2	X	Xp11.3 deletion	
397	Epilepsy, X-linked	SYN1	X	Xp11.23/11.3 deletion	
398	Fabry disease	GLA	X	Xq22.1 deletion	301500

399	Faciogenital dysplasia/Aarskog-Scott syndrome	FGD1	X	Xp11.22 deletion	
400	FG syndrome 1 (FGS1; Opitz-Kaveggia syndrome)	MED12	X	Xq13	305450
401	FG syndrome 2 (FGS2)	FLNA	X	Xq28 deletion	300321
402	FG syndrome 4 (FGS4)	CASK	X	Xp11.4	300422
403	FG syndrome 5 (FGS5)	MID2	X	Xq22.3	300581
404	Focal dermal hypoplasia (Goltz syndrome), XLMR	PORCN	X	Xp11.23 deletion	
405	Fragile-X mental retardation syndrome (FMR1)	FMR1	X	Xq27.3 microdeletion	300624
406	Granulomatous disease, chronic X-linked	CYBB	X	Xp11.4 deletion	306400
407	Hemophilia A, (Factor 8, hemophilia)	F8	X	Xq28 deletion	306700
408	Hemophilia B, (Factor 9, hemophilia)	F9	X	Xq27.1 deletion	306900
409	Heterotopia, periventricular, X-linked dominant/Otopalatodigital syndrome type 1 (OPD)/multifocal epilepsy in females	FLNA	X	Xq28 deletion	
410	Hunter/Mucopolysaccharidosis, type II (MPS2)	IDS	X	Xq28 deletion	309900
411	Hyperekplexia and epilepsy/X-linked mental retardation (XLMR)	ARHGEF9	X	Xq11.1 deletion	
412	Hyperglycerolemia (Glycerol kinase deficiency)	GK	X	Xp21.3-p21.2 deletion	307030
413	Hypophosphatemic rickets, X-linked dominant	PHEX	X	Xp22.11 deletion	
414	Ichthyosis, X-linked (steroid sulfatase deficiency)	STS	X	Xp22.31 deletion	308100
415	Incontinentia pigmenti (IP2)	IKBKG (NEMO)	X	Xq28 deletion	308300
416	Infantile spasm syndrome, X-linked (ISSX), West, Proud, XLAG, Partington, multifocal epilepsy	ARX	X	Xp21.3 deletion	308350
417	Kallmann syndrome 1 (KAL1)	KAL1	X	Xp22.31 deletion	308700
418	Leri-Weill dyschondrosteosis (LWD) / short stature	SHOX (SHOXY)	X	Xpter-p22.33 deletion	127300

419	Lesch-Nyhan syndrome (LNS)	HPRT1 (HPRT)	X	Xq26.2 deletion	300322
420	Lowe oculocerebrorenal syndrome	OCRL	X	Xq25 deletion	309000
421	LUBS (MRXSL) / MECP2 duplication	MECP2	X	Xq28 duplication	300260
422	MASA syndrome, X-linked mental retardation (XLMR)	L1CAM	X	Xq28 deletion	
423	Menkes disease (MNK), Cutis laxa / Occipital horn syndrome (OHS)	ATP7A	X	Xq21.1 deletion	
424	Mental retardation X-linked with isolate growth hormone deficiency (MRGH)	SOX3	X	Xq27.1 deletion or duplication	300123
425	Mental retardation, X-linked 21 (MRX21)	IL1RAPL1	X	Xp21.3 deletion	300143
426	Mental retardation, X-linked, syndromic, Turner type/X-linked mental retardation (XLMR)	HUWE1	X	Xp11.22 deletion	
427	Mental retardation, X-linked, with Epilepsy (XMRE)	ATP6AP2	X	Xp11.4 deletion	
428	Microphthalmia syndromic (MCOPS2)	BCOR	X	Xp11.4 deletion	300166
429	Microphthalmia, syndromic 7 (MCOPS7)/Microphthalmia with linear skin defects (MLS)	HCCS	X	Xp22.2 deletion	309801
430	Mitochondrial complex I deficiency; NADH-coenzyme Q reductase deficiency (nuclear-encoded)	NDUFA1	X	Xq24	252010
431	Mohr-Tranebjaerg syndrome	TIMM8A	X	Xq22.1 deletion	304700
432	Nance-Horan	NHS	X	Xp22.13 deletion	
433	Norrie disease	NDP	X	Xp11.3 deletion	
434	Opitz BBB syndrome	MID1	X	Xp22.2 deletion	300000
435	Ornithine transcarbamylase deficiency	OTC	X	Xp11.4 deletion	311250
436	Orofaciodigital syndrome (OFD1)	OFD1 (CXorf5)	X	Xp22.2 deletion	311200
437	Pelizaeus-Merzbacher disease (PMD)	PLP1	X	Xq22.2 duplication or deletion	312080
438	Perisylvian polymicrogyria (Polymicrogyria, bilateral perisylvian; BPP)	SRPX2	X	Xq22.1	
439	Phosphoglycerate kinase deficiency	PGK1	X	Xq21.1 deletion	

440	Phosphoribosylpyrophosphate synthetase superactivity	PRPS1	X	Xq22.3 deletion	
441	Pyruvate dehydrogenase deficiency	PDHA1	X	Xp22.12 deletion	
442	Renpenning syndrome 1 (RENS1)/Sutherland-Haax XLMR syndrome/Golabi-Ito-Hall syndrome	PQBP1	X	Xp11.23 deletion	
443	Retinoschisis, X-linked, juvenile (RS1)	RS1	X	Xp22.13 deletion	312700
444	Rett syndrome (RTT)	MECP2	X	Xq28 deletion	312750
445	Rett-like, Infantile spasm syndrome, X-linked (ISSX)	CDKL5	X	Xp21.3 deletion	300203
446	Siderius type X-linked mental retardation /Cleft lip/palate	PHF8	X	Xp11.22 deletion	
447	Simpson-Golabi-Behmel syndrome type 1;(SGBS1)	GPC3	X	Xq26.2 deletion	
448	Stocco dos Santos XLMR	SHROOM4 (KIAA1202)	X	Xp11.22 deletion	300434
449	Von Hippel-Lindau syndrome	VBP1	X	Xq28 deletion	
450	Wilms tumor,somatic	GPC3	X	Xq26	194070
451	X-inactivation, familial skewed, XIST deficiency	XIST	X	Xq13.2 usually ring X chromosome	314670
452	X-linked heterotaxy, visceral	ZIC3	X	Xq26.3 deletion	306955
453	X-linked idiopathic short statue (ISS)	SHOX (SHOXY)	X	Xp22.33 deletion	300582
454	X-linked lymphoproliferative syndrome (XLP1)	SH2D1A	X	Xq25 deletion	308240
455	X-linked mental retardation (XLMR)	CASK	X	Xp11.4 deletion	
456	X-linked mental retardation MRXS10	HSD17B10 (HADH2)	X	Xp11.22 deletion	300220
457	X-linked mental retardation (XLMR)	ELK1	X	Xp11.23 deletion	
458	X-linked mental retardation (XLMR)	SLC38A5	X	Xp11.23 deletion	
459	X-linked mental retardation (XLMR)	ZNF41	X	Xp11.3 deletion	
460	X-linked mental retardation (XLMR)	VCX3A	X	Xp22.31 deletion	
461	X-linked mental retardation (XLMR)	KLF8	X	Xp11.21 deletion	
462	X-linked mental retardation (XLMR)	GDI1	X	Xq28 deletion	
463	X-linked mental retardation (XLMR)	NXF5	X	Xq22.1 deletion	
464	X-linked mental retardation (XLMR)	ZMYM3 (ZNF261)	X	Xq13.3 deletion	300061
465	X-linked mental retardation (XLMR)	KIAA2022	X	Xq13.3 deletion	

466	X-linked mental retardation (XLMR)	ACSL4	X	Xq22.3 deletion	
467	X-linked mental retardation (XLMR) (MRX30)	PAK3	X	Xq22.3 deletion	300558
468	X-linked mental retardation (XLMR)	AGTR2	X	Xq23 deletion	
469	X-linked mental retardation (XLMR)	ZDHHC9	X	Xq25 deletion	
470	X-linked mental retardation (XLMR)	AFF2 (FMR2)	X	Xq28 deletion	309548
471	X-linked mental retardation (XLMR) (MRX45)	ZNF81	X	Xp11.23 deletion	
472	X-linked mental retardation (XLMR) (MRX46)	ARHGEF6	X	Xq26.3 deletion	
473	X-linked mental retardation (XLMR) (MRX54)	ARX	X	Xp21.3	300419
474	X-linked mental retardation (XLMR) (MRX58)	TSPAN7 (TM4SF2)	X	Xp11.4 deletion	300210
475	X-linked mental retardation (XLMR) (MRX59)	AP1S2	X	Xp22.2 deletion	
476	X-linked mental retardation (XLMR) (MRX9)	FTSJ1	X	Xp11.23 deletion	
477	X-linked mental retardation (XLMR) (MRX90)/Autism	DLG3	X	Xq13.1 deletion	
478	X-linked mental retardation (XLMR) (MRX91)	ZDHHC15	X	Xq13.3 deletion	
479	X-linked mental retardation (XLMR) (MRX94)	GRIA3	X	Xq25 deletion	
480	X-linked mental retardation (XLMR) with short stature, small testes muscle wasting, and tremor	CUL4B	X	Xq24 deletion	
481	X-linked mental retardation (XLMR), syndromic, Chrystianson type	SLC9A6	X	Xq26.3 deletion	
482	X-linked lissencephaly/multifocal spasms (LISX)	DCX	X	Xq22.3 deletion	300067
483	X-linked lissencephaly with ambiguous genitalia (LISX2)	ARX	X	Xp21.3	300215
484	XLMR / Snyder-Robinson syndrome	SMS	X	Xp22.11 deletion	
485	XLMR/Autism	KDM5C (JARID1C)	X	Xp11.22 deletion	314690
486	Xp pseudoautosomal region (PAR1)		X	Xpter-p22.33	
487	Xq pseudoautosomal region (PAR2)	SPRY3, VAMP7, IL9R, WASH6P	X	Xq28-qter	
488	Autism susceptibility locus	ASMT	Y	Yp11.3 deletion	300015
489	Autism susceptibility locus	ASMTL	Y	Yp11.3 deletion	300162
490	Azoospermia factor A (AZFa), Y-linked	USP9Y	Y	Yq11.21 interstitial deletion	415000
491	Azoospermia factor B (AZFb), Y-linked		Y	Yq11.21 interstitial deletion	415000

492	Azoospermia factor C (AZFc), Y-linked			Y	Yq11.21 interstitial deletion	415000
493	Azoospermia factor regions (AZFb+AZFc), Y-linked			Y	Yq11.21 interstitial deletion	415000
494	Bipolar disorder/Bipolar affective disorder susceptibility locus	VAMP7 (SYBL1)		Y	Yq12	300053
495	Gonadal dysgenesis, XY female type/Sex reversal X/Y translocations, (Sex determination male; XX male syndrome)	SRY		Y	Yp11.31 translocation/deletion	480000
496	Leri-Weill dyschondrosteosis (LWD) / short stature	SHOX (SHOXY)		Y	Ypter-Yp11.32 deletion	127300
497	X-linked idiopathic short stature (ISS)	SHOX (SHOXY)		Y	Ypter-p11.32 deletion	300582
498	XLMR/Autism	KDM5D (JARID1D)		Y	Yq11.222 deletion	426000
499	Yp pseudoautosomal region (PAR1)			Y	Ypter-p11.32	
500	Yq pseudoautosomal region (PAR2)	SPRY3, VAMP7, IL9R, WASH6P		Y	Yq12-qter	
501	Mitochondrial NADH dehydrogenase component of complex I, deficiency of (mitochondrial-encoded)	MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND5, MT-ND6, MT-TS2	Mitochondria			252010
502	Transient neonatal diabetes mellitus (hyperglycemia); Paternal UPD			6	paternal UPD of chr 6	
I	All 41 unique subtelomeric regions	Multiple	41 sites		41 sites	
II	All 43 unique pericentromeric regions	Multiple	43 sites		43 sites	
III	Aneuploidy for 24 chromosomes	Multiple	24 chromosomes		24 chromosomes	