No.	Gene	Condition
1	ACTB	Baraitser-Winter syndrome 1
2	ACTG1	Baraitser-Winter syndrome 2
3	ACTG2	Visceral myopathy 1
4	ACVR1	Fibrodysplasia Ossificans Progressiva
5	ADNP	Autosomal Dominant Mental Retardation 28
6	AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2
7	ANKRD11	KBG Syndrome
8	ARID1A	Autosomal Dominant Mental Retardation 14
9	ARID1B	Coffin-Siris syndrome 1
10	ASXL1	Bohring-Opitz Syndrome
11	ASXL3	Bainbridge-Ropers Syndrome
12	ATP1A2	Alternating Hemiplegia of Childhood 1
13	ATP1A2	Developmental and epileptic encephalopathy 98
14	ATP1A3	Developmental and epileptic encephalopathy 99
15	BCL11A	Dias-Logan syndrome
16	BICD2	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant
17	BRAF	Noonan Syndrome 7
18	BRAF	Cardiofaciocutaneous Syndrome
19	BRAF	LEOPARD syndrome 3
20	CACNA1A	Developmental and epileptic encephalopathy 42
21	CAMTA1	Cerebellar dysfunction with variable cognitive and behavioral abnormalities
22	CASK	Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia
23	CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia
24	CDKL5	Developmental and epileptic encephalopathy 2
25	CHD2	Developmental and epileptic encephalopathy 94
26	CHD7	CHARGE syndrome
27	CHD8	Intellectual developmental disorder with autism and macrocephaly
28	COL11A1	Stickler syndrome, type II
29	COL1A1	Osteogenesis Imperfecta type I
30	COL1A1/COL1A2	Osteogenesis Imperfecta type II
31	COL1A1/COL1A2	Osteogenesis Imperfecta type III
32	COL1A1/COL1A2	Osteogenesis Imperfecta type IV
33	COL2A1	Stickler syndrome, type I
34	COL2A1	Platyspondylic Lethal Skeletal dysplasia, Torrance type

35	COL2A1	Achondrogenesis, type II or hypochondrogenesis
36	COL2A1	SED congenita
37	COL4A1	Porencephaly 1
38	COL9A2	Epiphyseal dysplasia, multiple, 2
39	COL9A3	Epiphyseal dysplasia, multiple, 3, with or without myopathy
40	COMP	Pseudoachondroplasia
41	COMP	Epiphyseal dysplasia, multiple, 1
42	CREBBP	Rubinstein-Taybi Syndrome 1
43	CREBBP	Menke-Hennekam syndrome 1
44	CTCF	Intellectual developmental disorder, autosomal dominant 21
45	CTNNB1	Neurodevelopmental disorder with spastic diplegia and visual defects
46	DNM1	Developmental and epileptic encephalopathy 31
47	DYNC1H1	Intellectual developmental disorder, autosomal dominant 13
48	DYRK1A	Intellectual developmental disorder, autosomal dominant 7
49	EBP	Chondrodysplasia punctata, X-linked dominant
50	EFNB1	Craniofrontonasal dysplasia
51	EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type
52	EHMT1	Kleefstra Syndrome 1
53	EP300	Rubinstein-Taybi Syndrome 2
54	EP300	Menke-Hennekam syndrome 2
55	ERF	Craniosynostosis 4
56	ERF	Chitayat syndrome
57	FBN1	Marfan Syndrome
58	FGFR1/FGFR2	Jackson-Weiss Syndrome
59	FGFR1	Trigonocephaly 1
60	FGFR1/FGFR2	Pfeiffer syndrome
61	FGFR2	Saethre-Chotzen Syndrome
62	FGFR2	Bent Bone Dysplasia Syndrome
63	FGFR2	Beare-Stevenson Cutis Gyrata syndrome
64	FGFR2	Antley-Bixler Syndrome Without Genital Anomalies Or Disordered Steroidogenesis
65	FGFR2	Crouzon syndrome
66	FGFR2	Apert syndrome
67	FGFR3	Thanatophoric Dysplasia, type II
68	FGFR3	SADDAN
69	FGFR3	Muenke Syndrome
70	FGFR3	Crouzon syndrome with acanthosis nigricans
71	FGFR3	Thanatophoric Dysplasia, type I
72	FGFR3	Hypochondroplasia
73	FGFR3	Achondroplasia
74	FLNA	Otopalatodigital syndrome, type II

75	FLNB	Atelosteogenesis, type I
76	FLNB	Larsen Syndrome
77	FLNB	Atelosteogenesis, type III
78	FLNB	Boomerang dysplasia
79	FOXG1	Rett syndrome, congenital variant
80	FOXP1	Intellectual developmental disorder with language impairment with or without autistic features
81	FREM1	Trigonocephaly 2
82	GABRA1	Developmental and epileptic encephalopathy 19
83	GABRB2	Developmental and epileptic encephalopathy 92
84	GATAD2B	GAND syndrome
85	GFAP	Alexander Disease
86	GNAO1	Developmental and epileptic encephalopathy 17
87	GNAO1	Neurodevelopmental disorder with involuntary movements
88	GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant
89	GRIN2B	Developmental and epileptic encephalopathy 27
90	GRIN2B	Intellectual developmental disorder, autosomal dominant 6, with or without seizures
91	HDAC8	Cornelia de Lange syndrome 5
92	HNRNPK	Au-Kline Syndrome
93	HNRNPU	Developmental and epileptic encephalopathy 54
94	HRAS	Costello Syndrome
95	IFITM5	Osteogenesis imperfecta, type V
96	JAG1	Alagille Syndrome 1
97	KANSL1	Koolen-De Vries Syndrome
98	KAT6B	SBBYSS syndrome
99	KAT6B	Genitopatellar syndrome
100	KCNB1	Developmental and epileptic encephalopathy 26
101	KCNJ2	Andersen syndrome
102	KCNQ2	Developmental and epileptic encephalopathy 7
103	KCNT1	Developmental and epileptic encephalopathy 14
104	KIF1A	NESCAV syndrome
105	KMT2A	Wiedemann-Steiner syndrome
106	KMT2D	Kabuki Syndrome 1
107	KRAS	Cardiofaciocutaneous Syndrome 2
108	KRAS	Noonan Syndrome 3
109	LMNA	Muscular dystrophy, congenital
110	LMNA	Hutchinson-Gilford Progeria Syndrome
111	LZTR1	Noonan Syndrome 10
112	MAP2K1	Cardiofaciocutaneous Syndrome 3
113	MAP2K2	Cardiofaciocutaneous Syndrome 4

114	MECP2	Rett syndrome
115	MED13L	Impaired intellectual development and distinctive facial features with or without cardiac defects
116	MEF2C	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language
117	MSX2	Craniosynostosis 2
118	MSX2	Parietal Foramina With Cleidocranial Dysplasia
119	NALCN	Congenital Contractures Of The Limbs And Face, Hypotonia, And Developmental delay
120	NF1	Neurofibromatosis 1
121	NF2	Neurofibromatosis 2
122	NFIX	Marshall-Smith Syndrome
123	NIPBL	Cornelia de Lange syndrome 1
124	NOTCH2	Hajdu-Cheney Syndrome
125	NOTCH2	Alagille Syndrome 2
126	NR2F1	Bosch-Boonstra-Schaaf Optic Atrophy Syndrome
127	NRAS	Noonan syndrome 6
128	NSD1	Sotos Syndrome 1
129	NSDHL	CHILD syndrome
130	PACS1	Schuurs-Hoeijmakers syndrome
131	PIK3CA	Overgrowth syndrome and/or cerebral malformations
132	PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1
133	PPP2R1A	Intellectual developmental disorder, autosomal dominant 36
134	PPP2R5D	Intellectual developmental disorder, autosomal dominant 35
135	PRKAR1A	Acrodysostosis 1, with or without Hormone Resistance
136	PTPN11	Noonan Syndrome 1
137	PURA	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties
138	RAD21	Cornelia de Lange syndrome 4
139	RAF1	Noonan Syndrome 5
140	RERE	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart
141	RIT1	Noonan Syndrome 8
142	RPS6KA3	Coffin-Lowry Syndrome
143	RUNX2	Metaphyseal Dysplasia with Maxillary Hypoplasia with or without Brachydactyly
144	RUNX2	Cleidocranial dysplasia
145	SATB2	Glass Syndrome
146	SCN1A	Early Infantile Epileptic Encephalopathy 6

147	SCN1A	Developmental and epileptic encephalopathy 6B, non-Dravet
148	SCN2A	Developmental and epileptic encephalopathy 11
149	SCN2A	Episodic ataxia, type 9
150	SCN8A	Developmental and epileptic encephalopathy 13
151	SCN8A	Cognitive Impairment With Or Without Cerebellar Ataxia
152	SETBP1	Schinzel-Giedion syndrome
153	SETBP1	Intellectual developmental disorder, autosomal dominant 29
154	SETD2	Luscan-Lumish Syndrome
155	SETD5	Intellectual developmental disorder, autosomal dominant 23
156	SHANK3	Phelan-McDermid Syndrome
157	SHOC2	Noonan-Like Syndrome with Loose Anagen Hair
158	SKI	Shprintzen-Goldberg Syndrome
159	SLC25A24	Fontaine Progeroid Syndrome
160	SMAD3	Loeys-Dietz syndrome 3
161	SMAD4	Myhre Syndrome
162	SMARCA2	Nicolaides-Baraitser Syndrome
163	SMARCA2	Blepharophimosis-impaired intellectual development syndrome
164	SMARCA4	Autosomal Dominant Mental Retardation 16
165	SMARCB1	Autosomal Dominant Mental Retardation 15
166	SMARCE1	Coffin-Siris Syndrome 5
167	SMC1A	Cornelia de Lange syndrome 2
168	SMC3	Cornelia de Lange syndrome 3
169	SOS1	Noonan Syndrome 4
170	SOS2	Noonan Syndrome 9
171	SOX9	Campomelic Dysplasia
172	SPECC1L	Opitz GBBB Syndrome, Type II
173	SPTAN1	Developmental and epileptic encephalopathy 5
174	SRCAP	Floating-Harbor Syndrome
175	SRCAP	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities
176	STAT3	Hyper-IgE recurrent infection syndrome
177	STXBP1	Developmental and epileptic encephalopathy 4
178	SYNGAP1	Intellectual developmental disorder, autosomal dominant 5
179	TBL1XR1	Pierpont Syndrome
180	TBL1XR1	Intellectual developmental disorder, autosomal dominant 41
181	TBX5	Holt-Oram Syndrome
182	TCF4	Pitt-Hopkins syndrome
183	TGFB2	Loeys-Dietz syndrome 4
184	TGFBR1	Loeys-Dietz syndrome 1
185	TGFBR2	Loeys-Dietz syndrome 2

186	TRAF7	Cardiac, Facial, and Digital Anomalies with Developmental Delay
187	TRPS1	Trichorhinophalangeal syndrome, type I
188	TSC1	Tuberous Sclerosis-1
189	TSC2	Tuberous sclerosis-2
190	TUBA1A	Lissencephaly 3
191	TUBB	Symmetric circumferential skin creases, congenital, 1
192	TUBB	Cortical Dysplasia, Complex, with Other Brain Malformations 6
193	TUBB2A	Cortical Dysplasia, Complex, with Other Brain Malformations 5
194	TUBB4A	Leukodystrophy, hypomyelinating, 6
195	TWIST1	Saethre-Chotzen syndrome with or without eyelid anomalies
196	TWIST1	Craniosynostosis 1
197	TWIST1	Sweeney-Cox syndrome
198	TWIST1	Robinow-Sorauf Syndrome
199	WDR45	Neurodegeneration with brain iron accumulation 5
200	ZBTB20	Primrose syndrome
201	ZC4H2	Wieacker-Wolff syndrome, female-restricted
202	ZEB2	Mowat-Wilson syndrome