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

trisonim® NeoSeq evalúa con una sensibilidad y especificidad del 99%, la presencia de 6.246 variantes patogénicas o probablemente patogénicas, *de novo* en 155 genes asociados a 202 patologías de origen monogénico con un patrón de herencia autosómico dominante. El listado completo de las variantes está a su disposición bajo petición. info@nimgenetics.com