#	GENE	OMIM GENE	OMIM DISEASE	OMIM DISEASE	INHERITANCE (AR, AD, XL)
1	ABCA4	601691	248200	Stargardt disease 1, Retinitis pigmentosa 19	AR
2	ABCD1	300371	300100	Adrenoleukodystrophy	XL
3	ACADVL	609575	201475	VLCAD deficiency	AR
4	ADA	608958	102700	Severe combined immunodeficiency due to ADA deficiency	AR
5	AIPL1	604392	604393	Leber congenital amaurosis 4	AR
6	AIRE	607358	240300	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia	AR, AD
7	ALPL	171760	241510	Hypophosphatasia, childhood	AR
8	AMT	238310	605899	Glycine encephalopathy	AR
9	APTX	606350	208920	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	AR
10	ARL6	608845	600151	Bardet-Biedl syndrome 3	AR
11	ARSA	607574	250100	Metachromatic leukodystrophy	AR
12	ASL	608310	207900	Argininosuccinic aciduria	AR
13	ASPA	608034	271900	Canavan disease	AR
14	ATM	607585	208900	Ataxia-telangiectasia	AR
15	ATP7A	300011	309400	Menkes disease	XL
16	ATP7B	606882	277900	Wilson disease	AR
17	BCKDHA	608348	248600	Maple syrup urine disease, type Ia	AR
18	BCKDHB	248611	248600	Maple syrup urine disease, type Ib	AR
19	BEST1	607854	611809	Bestrophinopathy, autosomal recessive	AR
20	BTD	609019	253260	Biotinidase deficiency	AR
21	BTK	300300	307200	Agammaglobulinemia and isolated hormone deficiency	XL
22	CAPN3	114240	253600	Muscular dystrophy, limb-girdle, type 2A	AR
23	CASQ2	114251	611938	Ventricular tachycardia, catecholaminergic polymorphic, 2	AR
24	CAV3	601253	607801	Muscular dystrophy, limb-girdle, type IC	AR
25	CCDC39	613798	613807	Ciliary dyskinesia, primary, 14	AR
26	CCDC40	613799	613808	Ciliary dyskinesia, primary, 15	AR
27	CDH23	605516	601386	Deafness, autosomal recessive 12	AR
28	CEP290	610142	610188	Joubert syndrome 5	AR
29	CERKL	608381	608380	Retinitis pigmentosa 26	AR
30	CFTR	602421	219700	Cystic fibrosis	AR
31	CHAT	118490	254210	Myasthenic syndrome, congenital, 6, presynaptic	AR
32	CHM	300390	303100	Choroideremia	XL
33	CHRNA1	100690	253290	Multiple pterygium syndrome, lethal type	AR
34	CHRND	100720	253290	Multiple pterygium syndrome, lethal type	AR
35	CHRNE	100725	605809	Myasthenic syndrome, congenital, 4A, slow-channel, Myasthenic syndrome, congenital, 4B, fast-channel	AR
36	CLCN1	118425	255700	Myotonia congenita, recessive	AR
37	CNGB1	600724	613767	Retinitis pigmentosa 45	AR
38	COL11A1	120280	228520	Fibrochondrogenesis 1	AR
39	COL11A2	120290	215150	Otospondylomegaepiphyseal dysplasia	AR
40	COL1A2	120160	225320	Ehlers-Danlos syndrome, cardiac valvular form	AR
41	COL2A1	120140	215150	Otospondylomegaepiphyseal dysplasia	AR
42	COL4A5	303630	301050	Alport syndrome	XL
43	COL7A1	120120	226600	Epidermolysis bullosa dystrophica, AR	AR
44	COL9A1	120210	614134	Stickler syndrome, type IV	AR
45	CRB1	604210	600105	Retinitis pigmentosa-12, autosomal recessive	AR
46	CTDP1	604927	604168	Congenital cataracts, facial dysmorphism, and neuropathy	AR
47	CTNS	606272	219800	Cystinosis, nephropathic	AR
48	CYP27A1	606530	213700	Cerebrotendinous xanthomatosis	AR
49	DBT	248610	248600	Maple syrup urine disease, type II	AR
50	DCX	300121	300067	Lissencephaly, X-linked	XL
51	DES	125660	601419	Myopathy, myofibrillar, 1	AR
52	DHCR7	602858	270400	Smith-Lemli-Opitz syndrome	AR
53	DKC1	300126	305000	Dyskeratosis congenita, X-linked	XL

54	DLD	238331	246900	Dihydrolipoamide dehydrogenase deficiency	AR
55	DMD	300377	310200	Duchenne muscular dystrophy	XL
56	DNAH11	603339	611884	Ciliary dyskinesia, primary, 7, with or without situs inversus	AR
57	DNAH5	603335	608644	Ciliary dyskinesia, primary, 3, with or without situs inversus	AR
58	DNAI1	604366	244400	Ciliary dyskinesia, primary, 1, with or without situs inversus	AR
59	DNAI2	605483	612444	Ciliary dyskinesia, primary, 9, with or without situs inversus	AR
60	DNM2	602378	615368	Lethal congenital contracture syndrome 5	AR
61	DOK7	610285	254300	Myasthenic syndrome, congenital, 10	AR
62	DSC2	125645	610476	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair	AR
63	DSP	125647	607655	Skin fragility-woolly hair syndrome	AR
64	DYSF	603009	253601	Muscular dystrophy, limb-girdle, type 2B	AR
65	EMD	300384	310300	Emery-Dreifuss muscular dystrophy 1, X-linked	XL
66	EXT1	608177	215300	Chondrosarcoma	AR
67	EYS	612424	602772	Retinitis pigmentosa 25	AR
68	F8	300841	306700	Hemophilia A	XL
69	F9	300746	306900	Hemophilia B	XL
70	FANCA	607139	227650	Fanconi anemia, complementation group A	AR
71	FANCC	613899	227645	Fanconi anemia, complementation group C	AR
72	FANCF	613897	603467	Fanconi anemia, complementation group F	AR
73	FANCG	602956	614082	Fanconi anemia, complementation group G	AR
74	FBXO7	605648	260300	Parkinson disease 15, autosomal recessive	AR
75	FMO3	136132	602079	Trimethylaminuria	AR
76	FRMD7	300628	310700	Nystagmus, infantile periodic alternating, X-linked	XL
77	FXN	606829	229300	Friedreich ataxia	AR
78	GAA	606800	232300	Glycogen storage disease II	AR
79	GALT	606999	230400	Galactosemia	AR
80	GBA	606463	230800	Gaucher disease, type I	AR
81	GBE1	607839	232500	Glycogen storage disease IV	AR
82	GCSH	238330	605899	Glycine encephalopathy	AR
83	GDF5	601146	201250	Acromesomelic dysplasia, Hunter-Thompson type	AR
84	GJB2	121011	220290	Deafness, autosomal recessive 1B	AR
85	GJB3	603324	220290	Deafness, digenic, GJB2/GJB3	AR
86	GJB6	604418	612645	Deafness, autosomal recessive 1B	AR
87	GLA	300644	301500	Fabry disease	XL
88	GLDC	238300	605899	Glycine encephalopathy	AR
89	GNE	603824	269921	Sialuria, Nonaka myopathy	AR, AD
90	GNPTAB	607840	252500	Mucolipidosis II alpha/beta	AR
91	GPC3	300037	312870	Simpson-Golabi-Behmel syndrome, type 1	XL
92	GPR143	300808	300814	Nystagmus 6, congenital, X-linked	XL
93	GUCY2D	600179	204000	Leber congenital amaurosis 1	AR
94	HBB	141900	613985	Thalassemias, beta-	AR
95	HEXA	606869	272800	Tay-Sachs disease	AR
96	HFE	613609	235200	Hemochromatosis	AR
97	HIBCH	610690	250620	3-hydroxyisobutryl-CoA hydrolase deficiency	AR
98	HR	602302	203655	Alopecia universalis	AR
99	IDS	300823	309900	Mucopolysaccharidosis II	XL
100	IDUA	252800	607014	Mucopolysaccharidosis Ih	AR
101	IKBKAP	603722	223900	Dysautonomia, familial	AR
102	IL2RG	308380	312863	Combined immunodeficiency, X-linked, moderate	XL
103	ITGB4	147557	226650	Epidermolysis bullosa, junctional, non-Herlitz type	AR
104	JUP	173325	601214	Naxos disease	AR
105	KCNE1	176261	612347	Jervell and Lange-Nielsen syndrome 2	AR
106	KCNQ1	607542	220400	Jervell and Lange-Nielsen syndrome	AR
107	KIAA0196	610657	220210	Ritscher-Schinzel syndrome	AR
108	KRT14	148066	601001	Epidermolysis bullosa simplex, recessive 1	AR
109	KRT5	148040	601001	Epidermolysis bullosa simplex, recessive 1	AR
110	L1CAM	308840	304100	Corpus callosum, partial agenesis of	XL
111	LAMB3	150310	226700	Epidermolysis bullosa, junctional, Herlitz type	AR
112	LAMP2	309060	300257	Danon disease	XL
113	LMNA	150330	605588	Charcot-Marie-Tooth disease, type 2B1	AR
114	LRAT	604863	613341	Retinal dystrophy, early-onset severe	AR

115	MECP2	300005	312750	Rett syndrome	XL
116	MED12	300188	305450	Opitz-Kaveggia syndrome	XL
117	MERTK	604705	613862	Retinitis pigmentosa 38	AR
118	MFN2	608507	609260	Charcot-Marie-Tooth disease, type 2A2	AR
119	MLH1	120436	276300	Mismatch repair cancer syndrome	AR
120	MMAA	607481	251100	Methylmalonic aciduria, vitamin B12-responsive	AR
121	MMAB	607568	251110	Methylmalonic aciduria, vitamin B12-responsive,	AR
122	MMACHC	609831	277400	Methylmalonic aciduria and homocystinuria, cblC type	AR
123	MPZ	159440	605253	Neuropathy, congenital hypomyelinating	AR
124	MSH2	609309	276300	Mismatch repair cancer syndrome	AR
125	MTM1	300415	310400	Myotubular myopathy, X-linked	XL
126	MUT	609058	251000	Methylmalonic aciduria, mut(0) type	AR
127	MYH7	160760	255160	Myopathy, myosin storage, autosomal recessive	AR
128	MYO7A	276903	600060	Deafness, autosomal recessive 2	AR
129	NKX2-5	600584	217095	Conotruncal heart malformations, variable	AR
130	NPC1	607623	257220	Niemann-Pick disease, type D	AR
131	NPC2	601015	607625	Niemann-pick disease, type C2	AR
132	NR2E3	604485	268100	Enhanced S-cone syndrome	AR
133	OCA2	611409	203200	Albinism, oculocutaneous, type II	AR
134	OCRL	300535	309000	Lowe syndrome	XL
135	OTC	300461	311250	Ornithine transcarbamylase deficiency	XL
136	PAH	612349	261600	Phenylketonuria	AR
137	PAX3	606597	268220	Rhabdomyosarcoma 2, alveolar	AR
138	PAX6	607108	206700	Gillespie syndrome	AR
139	PCDH15	605514	609533	Deafness, autosomal recessive 23	AR
140	PEX1	602136	214100	Peroxisome biogenesis disorder 1A (Zellweger)	AR
141	PEX10	602859	614870	Peroxisome biogenesis disorder 6A (Zellweger)	AR
142	PEX13	601789	614883	Peroxisome biogenesis disorder 11A (Zellweger)	AR
143	PEX14	601791	614887	Peroxisome biogenesis disorder 13A (Zellweger)	AR
144	PEX19	600279	614886	Peroxisome biogenesis disorder 12A (Zellweger)	AR
145	PEX26	608666	614872	Peroxisome biogenesis disorder 7A (Zellweger)	AR
146	PEX3	603164	614882	Peroxisome biogenesis disorder 10A (Zellweger)	AR
147	PEX5	600414	214110	Peroxisome biogenesis disorder 2A (Zellweger)	AR
148	PINK1	608309	605909	Parkinson disease 6, early onset	AR
149	PKHD1	606702	263200	Polycystic kidney and hepatic disease	AR
150	PLEC	601282	613723	Muscular dystrophy, limb-girdle, type 2Q	AR
151	PLOD1	153454	225400	Ehlers-Danlos syndrome, type VI	AR
152	PMM2	601785	212065	Congenital disorder of glycosylation, type Ia	AR
153	PMP22	601097	118220	Charcot-Marie-Tooth disease type 1A, Dejerine-Sottas disease	AR
154	POLG	174763	203700	Mitochondrial DNA depletion syndrome 4A (Alpers type)	AR
155	PPT1	600722	256730	Ceroid lipofuscinosis, neuronal, 1	AR
156	PRCD	610598	610599	Retinitis pigmentosa 36	AR
157	PROM1	604365	612095	Retinitis pigmentosa 41	AR
158	PRPH2	179605	608133	Retinitis pigmentosa 7 and digenic	AR
159	RAG1	179615	601457	Severe combined immunodeficiency, B cell-negative	AR
160	RAG2	179616	601457	Severe combined immunodeficiency, B cell-negative	AR
161	RAPSN	601592	208150	Fetal akinesia deformation sequence	AR
162	RDH12	608830	612712	Leber congenital amaurosis 13	AR
163	RET	164761	191830	Renal agenesis	AR
164	RHO	180380	136880	Retinitis punctata albescens	AR
165	ROR2	602337	269310	Robinow syndrome, autosomal recessive	AR
166	RPE65	180069	204100	Leber congenital amaurosis 2	AR
167	RPGR	312610	304020	Cone-rod dystrophy, X-linked, 1	XL
168	RPGRIP1	605446	608194	Cone-rod dystrophy 13	AR
169	RPS6KA3	300075	303600	Coffin-Lowry syndrome	XL
170	RS1	300839	312700	Retinoschisis	XL
171	RSPH4A	612647	612649	Ciliary dyskinesia, primary, 11	AR
172	RSPH9	612648	612650	Ciliary dyskinesia, primary, 12	AR
173	RYR1	180901	117000	Central core disease	AR
174	SCN5A	600163	608567	Sick sinus syndrome 1	AR
175	SCN9A	603415	243000	Insensitivity to pain, congenital	AR
176	SEMA4A	607292	610283	Cone-rod dystrophy 10	AR

177	SERPINA1	107400	613490	Emphysema due to AAT deficiency	AR
178	SGCD	601411	601287	Muscular dystrophy, limb-girdle, type 2F	AR
179	SLC25A13	603859	605814	Citrullinemia, type II, neonatal-onset	AR
180	SLC25A4	103220	615418	Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type)	AR
181	SLC26A4	605646	600791	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	AR
182	SOD1	147450	105400	Amyotrophic lateral sclerosis 1	AR
183	SPATA7	609868	604232	Leber congenital amaurosis 3	AR
184	SPG7	602783	607259	Spastic paraplegia 7, autosomal recessive	AR
185	TAF1	313650	314250	Dystonia-Parkinsonism, X-linked	XL
186	TAZ	300394	302060	Barth syndrome	XL
187	TNNT1	191041	605355	Nemaline myopathy 5, Amish type	AR
188	TNXB	600985	606408	Ehlers-Danlos syndrome due to tenascin X deficiency	AR
189	TTPA	600415	277460	Ataxia with isolated vitamin E deficiency	AR
190	TULP1	602280	613843	Leber congenital amaurosis 15	AR
191	TYR	606933	203100	Albinism, oculocutaneous, type IA	AR
192	USH1C	605242	276904	Usher syndrome, type 1C	AR
193	USH2A	608400	276901	Usher syndrome, type 2A	AR
194	WAS	300392	313900	Thrombocytopenia, X-linked	AR
195	WRN	604611	277700	Werner syndrome	XL
196	WT1	607102	256370	Nephrotic syndrome, type 4	AR

NOTE: The applied methodology may not necessarily detect all the possible pathogenic mutations of each gene/disease. Generally, the analysis covers a large percentage (~95%) of mutations in exons-coding regions of the genes, while it does not generally detect mutations in non-coding regions/introns and exonic deletions or duplications >15-20 base-pairs.

The test also includes MLPA analysis for the detection of deletions/duplications of the SMN1 and DMD genes, associated with the genetic disorders Spinal Muscular Atrophy - SMA (OMIM 253300) and Duchenne/Becker muscular dystrophy (DMD/BMD - OMIM 310200).
In females only, we also include carrier testing for Fragile X syndrome.