InterGenetics
DIAGNOSTIC GENETIC CENTER

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#	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	ACTA2	102620	611788	Aortic aneurysm, familial thoracic 6	AD
5	ADA	608958	102700	Severe combined immunodeficiency due to ADA deficiency	AR
6	AIPL1	604392	604393	Leber congenital amaurosis 4	AR
7	AIRE	607358	240300	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia	AR, AD
8	AKAP9	604001	611820	Long QT syndrome-11	AD
9	ALPL	171760	241510	Hypophosphatasia, childhood	AR
10	AMT	238310	605899	Glycine encephalopathy	AR
11	ANK2	106410	600919	Long QT syndrome 4	AD
12	APC	611731	175100	Adenomatous polyposis coli	AD
13	APP	104760	104300	Alzheimer disease 1, familial	AD
14	APTX	606350	208920	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	AR
15	ARL6	608845	600151	Bardet-Biedl syndrome 3	AR
16	ARSA	607574	250100	Metachromatic leukodystrophy	AR
17	ASL	608310	207900	Argininosuccinic aciduria	AR
18	ASPA	608034	271900	Canavan disease	AR
19	ATL1	606439	613708	Neuropathy, hereditary sensory, type ID	AD
20	ATM	607585	208900	Ataxia-telangiectasia	AR
21	ATP2A2	108740	124200	Darier disease, Acrokeratosis verruciformis	AD
22	ATP7A	300011	309400	Menkes disease	XL
23	ATP7B	606882	277900	Wilson disease	AR
24	ATXN1	601556	164400	Spinocerebellar ataxia 1	AD
25	ATXN2	601517	183090	Spinocerebellar ataxia 2	AD
26	ATXN7	607640	164500	Spinocerebellar ataxia 7	AD
27	BCKDHA	608348	248600	Maple syrup urine disease, type Ia	AR
28	BCKDHB	248611	248600	Maple syrup urine disease, type Ib	AR
29	BEST1	607854	611809	Bestrophinopathy, autosomal recessive	AR
30	BMPR1A	601299	174900	Juvenile polyposis syndrome, infantile form	AD
31	BTD	609019	253260	Biotinidase deficiency	AR
32	BTK	300300	307200	Agammaglobulinemia and isolated hormone deficiency	XL
33	CA4	114760	600852	Retinitis pigmentosa 17	AD
34	CACNA1C	114205	611875	Brugada syndrome 3, Timothy syndrome	AD
35	CACNB2	600003	611876	Brugada syndrome 4	AD
36	CAPN3	114240	253600	Muscular dystrophy, limb-girdle, type 2A	AR
37	CASQ2	114251	611938	Ventricular tachycardia, catecholaminergic polymorphic, 2	AR
38	CAV3	601253	607801	Muscular dystrophy, limb-girdle, type IC	AR
39	CCDC39	613798	613807	Ciliary dyskinesia, primary, 14	AR
40	CCDC40	613799	613808	Ciliary dyskinesia, primary, 15	AR
41	CDH23	605516	601386	Deafness, autosomal recessive 12	AR
42	CEP290	610142	610188	Joubert syndrome 5	AR
43	CERKL	608381	608380	Retinitis pigmentosa 26	AR
44	CFTR	602421	219700	Cystic fibrosis	AR
45	CHAT	118490	254210	Myasthenic syndrome, congenital, 6, presynaptic	AR
46	CHD7	608892	214800	CHARGE syndrome	AD
47	CHEK2	604373	609265	Li-Fraumeni syndrome	AD
48	CHM	300390	303100	Choroideremia	XL
49	CHRNA1	100690	253290	Multiple pterygium syndrome, lethal type	AR
50	CHRNB1	100710	616313	Myasthenic syndrome, congenital, 2A, slow-channel	AD
51	CHRND	100720	253290	Multiple pterygium syndrome, lethal type	AR

52	CHRNE	100725	605809	Myasthenic syndrome, congenital, 4A, slow-channel, Myasthenic syndrome, congenital, 4B, fast-channel	AR
53	CLCN1	118425	255700	Myotonia congenita, recessive	AR
54	CNGB1	600724	613767	Retinitis pigmentosa 45	AR
55	COL11A1	120280	228520	Fibrochondrogenesis 1	AR
56	COL11A2	120290	215150	Otospondylomegaepiphyseal dysplasia	AR
57	COL1A1	120150	166200	Osteogenesis imperfecta type I, II, III, IV, Ehlers-Danlos syndrome, classic	AD
58	COL1A2	120160	225320	Ehlers-Danlos syndrome, cardiac valvular form	AR
59	COL2A1	120140	215150	Otospondylomegaepiphyseal dysplasia	AR
60	COL3A1	120180	130050	Ehlers-Danlos syndrome, type IV	AD
61	COL4A1	120130	175780	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, Porencephaly 1	AD
62	COL4A5	303630	301050	Alport syndrome	XL
63	COL5A1	120215	130000	Ehlers-Danlos syndrome, classic type	AD
64	COL5A2	120190	130000	Ehlers-Danlos syndrome, classic type	AD
65	COL7A1	120120	226600	Epidermolysis bullosa dystrophica, AR	AR
66	COL9A1	120210	614134	Stickler syndrome, type IV	AR
67	CRB1	604210	600105	Retinitis pigmentosa-12, autosomal recessive	AR
68	CRX	602225	120970	Cone-rod retinal dystrophy-2, Leber congenital amaurosis 7	AD
69	CTDP1	604927	604168	Congenital cataracts, facial dysmorphism, and neuropathy	AR
70	CTNS	606272	219800	Cystinosis, nephropathic	AR
71	CYP27A1	606530	213700	Cerebrotendinous xanthomatosis	AR
72	DBT	248610	248600	Maple syrup urine disease, type II	AR
73	DCX	300121	300067	Lissencephaly, X-linked	XL
74	DES	125660	601419	Myopathy, myofibrillar, 1	AR
75	DHCR7	602858	270400	Smith-Lemli-Opitz syndrome	AR
76	DKC1	300126	305000	Dyskeratosis congenita, X-linked	XL
77	DLD	238331	246900	Dihydrolipoamide dehydrogenase deficiency	AR
78	DMD	300377	310200	Duchenne/Becker muscular dystrophy	XL
79	DNAH11	603339	611884	Ciliary dyskinesia, primary, 7, with or without situs inversus	AR
80	DNAH5	603335	608644	Ciliary dyskinesia, primary, 3, with or without situs inversus	AR
81	DNAI1	604366	244400	Ciliary dyskinesia, primary, 1, with or without situs inversus	AR
82	DNAI2	605483	612444	Ciliary dyskinesia, primary, 9, with or without situs inversus	AR
83	DNM2	602378	615368	Lethal congenital contracture syndrome 5	AR
84	DOK7	610285	254300	Myasthenic syndrome, congenital, 10	AR
85	DSC2	125645	610476	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair	AR
86	DSG2	125671	610193	Arrhythmogenic right ventricular dysplasia 10	AD
87	DSP	125647	607655	Skin fragility-woolly hair syndrome	AR
88	DYSF	603009	253601	Muscular dystrophy, limb-girdle, type 2B	AR
89	ELN	130160	185500	Supravalvar aortic stenosis	AD
90	EMD	300384	310300	Emery-Dreifuss muscular dystrophy 1, X-linked	XL
91	ENG	131195	187300	Telangiectasia, hereditary hemorrhagic, type 1	AD
92	EXT1	608177	215300	Chondrosarcoma	AR
93	EYA1	601653	602588	Branchiootic syndrome 1 , Anterior segment anomalies with or without cataract, Branchiootorenal syndrome 1, with or without cataracts	AD
94	EYS	612424	602772	Retinitis pigmentosa 25	AR
95	F8	300841	306700	Hemophilia A	XL
96	F9	300746	306900	Hemophilia B	XL
97	FANCA	607139	227650	Fanconi anemia, complementation group A	AR
98	FANCC	613899	227645	Fanconi anemia, complementation group C	AR
99	FANCF	613897	603467	Fanconi anemia, complementation group F	AR
100	FANCG	602956	614082	Fanconi anemia, complementation group G	AR
101	FBN1	134797	154700	Marfan syndrome	AD
102	FBXO7	605648	260300	Parkinson disease 15, autosomal recessive	AR
103	FGFR1	136350	123150	Jackson-Weiss syndrome, Pfeiffer syndrome, Trigonocephaly 1, Osteoglophonic dysplasia	AD
104	FGFR3	134934	100800	Achondroplasia, Hypochondroplasia, Thanatophoric dysplasia, type I	AD
105	FMO3	136132	602079	Trimethylaminuria	AR

106	FOXL2	605597	110100	Blepharophimosis, epicanthus inversus, and ptosis, type 1 & 2	AD
107	FRMD7	300628	310700	Nystagmus, infantile periodic alternating, X-linked	XL
108	FSCN2	607643	607921	Retinitis pigmentosa 30	AD
109	FXN	606829	229300	Friedreich ataxia	AR
110	GAA	606800	232300	Glycogen storage disease II	AR
111	GALT	606999	230400	Galactosemia	AR
112	GATA4	600576	607941	Atrial septal defect 2, Atrioventricular septal defect 4, Tetralogy of Fallot	AD
113	GBA	606463	230800	Gaucher disease, type I	AR
114	GBE1	607839	232500	Glycogen storage disease IV	AR
115	GCSH	238330	605899	Glycine encephalopathy	AR
116	GDF5	601146	201250	Acromesomelic dysplasia, Hunter-Thompson type	AR
117	GJB2	121011	220290	Deafness, autosomal recessive 1B	AR
118	GJB3	603324	220290	Deafness, digenic, GJB2/GJB3	AR
119	GJB6	604418	612645	Deafness, autosomal recessive 1B	AR
120	GLA	300644	301500	Fabry disease	XL
121	GLDC	238300	605899	Glycine encephalopathy	AR
122	GNE	603824	269921	Sialuria, Nonaka myopathy	AR, AD
123	GNPTAB	607840	252500	Mucolipidosis II alpha/beta	AR
124	GPC3	300037	312870	Simpson-Golabi-Behmel syndrome, type 1	XL
125	GPD1L	611778	611777	Brugada syndrome 2	AD
126	GPR143	300808	300814	Nystagmus 6, congenital, X-linked	XL
127	GUCY2D	600179	204000	Leber congenital amaurosis 1	AR
128	HCN4	605206	613123	Brugada syndrome 8, Sick sinus syndrome 2	AD
129	HEXA	606869	272800	Tay-Sachs disease	AR
130	HFE	613609	235200	Hemochromatosis	AR
131	HIBCH	610690	250620	3-hydroxyisobutryl-CoA hydrolase deficiency	AR
132	HMBS	609806	176000	Porphyria, acute intermittent	AD
133	HR	602302	203655	Alopecia universalis	AR
134	IDS	300823	309900	Mucopolysaccharidosis II	XL
135	IDUA	252800	607014	Mucopolysaccharidosis Ih	AR
136	IKBKAP	603722	223900	Dysautonomia, familial	AR
137	IL2RG	308380	312863	Combined immunodeficiency, X-linked, moderate	XL
138	IMPDH1	146690	180105	Retinitis pigmentosa 10, Leber congenital amaurosis 11	AD
139	ITGB4	147557	226650	Epidermolysis bullosa, junctional, non-Herlitz type	AR
140	JAG1	601920	118450	Alagille syndrome	AD
141	JUP	173325	601214	Naxos disease	AR
142	KCNE1	176261	612347	Jervell and Lange-Nielsen syndrome 2, Long QT syndrome 5	AR, AD
143	KCNE2	603796	613693	Long QT syndrome 6	AD
144	KCNE3	604433	613119	Brugada syndrome 6	AD
145	KCNH2	152427	613688	Long QT syndrome 2, Short QT syndrome 1	AD
146	KCNJ2	600681	170390	Andersen syndrome, Short QT syndrome 3	AD
147	KCNQ1	607542	220400	Jervell and Lange-Nielsen syndrome	AR
148	KCNQ4	603537	600101	Deafness, autosomal dominant 2A	AD
149	KIAA0196	610657	220210	Ritscher-Schinzel syndrome	AR
150	KLHL7	611119	612943	Retinitis pigmentosa 42	AD
151	KRAS	190070	609942	Noonan syndrome 3, Cardiofaciocutaneous syndrome 2	AD
152	KRT14	148066	601001	Epidermolysis bullosa simplex, recessive 1	AR
153	KRT5	148040	601001	Epidermolysis bullosa simplex, recessive 1	AR
154	L1CAM	308840	304100	Corpus callosum, partial agenesis of	XL
155	LAMB3	150310	226700	Epidermolysis bullosa, junctional, Herlitz type	AR
156	LAMP2	309060	300257	Danon disease	XL
157	LMNA	150330	605588	Charcot-Marie-Tooth disease, type 2B1	AR
158	LRAT	604863	613341	Retinal dystrophy, early-onset severe	AR
159	MAPT	157140	260540	Supranuclear palsy, progressive atypical	AR
160	MC1R	155555	203200	{Albinism, oculocutaneous, type II, modifier of}	AR
161	MECP2	300005	312750	Rett syndrome	XL
162	MED12	300188	305450	Opitz-Kaveggia syndrome	XL
163	MEN1	613733	131100	Multiple endocrine neoplasia 1	AD
164	MERTK	604705	613862	Retinitis pigmentosa 38	AR

165	MFN2	608507	609260	Charcot-Marie-Tooth disease, type 2A2	AR
166	MLH1	120436	276300	Mismatch repair cancer syndrome	AR
167	MMAA	607481	251100	Methylmalonic aciduria, vitamin B12-responsive	AR
168	MMAB	607568	251110	Methylmalonic aciduria, vitamin B12-responsive,	AR
169	MMACHC	609831	277400	Methylmalonic aciduria and homocystinuria, cblC type	AR
170	MPZ	159440	605253	Neuropathy, congenital hypomyelinating	AR
171	MSH2	609309	276300	Mismatch repair cancer syndrome	AR
172	MTM1	300415	310400	Myotubular myopathy, X-linked	XL
173	MUT	609058	251000	Methylmalonic aciduria, mut(0) type	AR
174	MYH11	160745	132900	Aortic aneurysm, familial thoracic 4	AD
175	MYLK	600922	613780	Aortic aneurysm, familial thoracic 7	AD
176	MYO7A	276903	600060	Deafness, autosomal recessive 2	AR
177	NF1	613113	162200	Neurofibromatosis, type 1	AD
178	NF2	607379	101100	Neurofibromatosis, type 2	AD
179	NIPBL	608667	122470	Cornelia de Lange syndrome 1	AD
180	NKX2-5	600584	217095	Conotruncal heart malformations, variable	AR
181	NPC1	607623	257220	Niemann-Pick disease, type D	AR
182	NPC2	601015	607625	Niemann-pick disease, type C2	AR
183	NR2E3	604485	268100	Enhanced S-cone syndrome	AR
184	NRAS	164790	613224	Noonan syndrome 6	AD
185	NSD1	606681	117550	Sotos syndrome 1, Beckwith-Wiedemann syndrome	AD
186	OCA2	611409	203200	Albinism, oculocutaneous, type II	AR
187	OCRL	300535	309000	Lowe syndrome	XL
188	OTC	300461	311250	Ornithine transcarbamylase deficiency	XL
189	PABPN1	602279	164300	Oculopharyngeal muscular dystrophy	AD
190	PAFAH1B1	601545	607432	Lissencephaly 1, Subcortical laminar heterotopia	AD
191	PAH	612349	261600	Phenylketonuria	AR
192	PAX3	606597	268220	Rhabdomyosarcoma 2, alveolar	AR
193	PAX6	607108	206700	Gillespie syndrome	AR
194	PCDH15	605514	609533	Deafness, autosomal recessive 23	AR
195	PEX1	602136	214100	Peroxisome biogenesis disorder 1A (Zellweger)	AR
196	PEX10	602859	614870	Peroxisome biogenesis disorder 6A (Zellweger)	AR
197	PEX13	601789	614883	Peroxisome biogenesis disorder 11A (Zellweger)	AR
198	PEX14	601791	614887	Peroxisome biogenesis disorder 13A (Zellweger)	AR
199	PEX19	600279	614886	Peroxisome biogenesis disorder 12A (Zellweger)	AR
200	PEX26	608666	614872	Peroxisome biogenesis disorder 7A (Zellweger)	AR
201	PEX3	603164	614882	Peroxisome biogenesis disorder 10A (Zellweger)	AR
202	PEX5	600414	214110	Peroxisome biogenesis disorder 2A (Zellweger)	AR
203	PINK1	608309	605909	Parkinson disease 6, early onset	AR
204	PKD1	601313	173900	Polycystic kidney disease, adult type I	AD
205	PKD2	173910	613095	Polycystic kidney disease 2	AD
206	PKHD1	606702	263200	Polycystic kidney and hepatic disease, infantile	AR
207	PKP2	602861	609040	Arrhythmogenic right ventricular dysplasia 9	AD
208	PLEC	601282	613723	Muscular dystrophy, limb-girdle, type 2Q	AR
209	PLOD1	153454	225400	Ehlers-Danlos syndrome, type VI	AR
210	PMM2	601785	212065	Congenital disorder of glycosylation, type Ia	AR
211	PMP22	601097	145900	Dejerine-Sottas disease	AR
212	POLG	174763	203700	Mitochondrial DNA depletion syndrome 4A (Alpers type)	AR
213	PPT1	600722	256730	Ceroid lipofuscinosis, neuronal, 1	AR
214	PRCD	610598	610599	Retinitis pigmentosa 36	AR
215	PROM1	604365	612095	Retinitis pigmentosa 41	AR
216	PRPF31	606419	600138	Retinitis pigmentosa 11	AD
217	PRPF8	607300	600059	Retinitis pigmentosa 13	AD
218	PRPH2	179605	608133	Retinitis pigmentosa 7 and digenic	AR
219	PSEN1	104311	607822	Alzheimer disease, type 3 with spastic paraparesis and unusual plaques and apraxia	AD
220	PSEN2	600579	606889	Alzheimer disease-4	AD
221	PTCH1	601309	610828	Holoprosencephaly-7	AR
222	PTPN11	176876	163950	Noonan syndrome 1, LEOPARD syndrome 1	AD
223	RAG1	179615	601457	Severe combined immunodeficiency, B cell-negative	AR
224	RAG2	179616	601457	Severe combined immunodeficiency, B cell-negative	AR

225	RAI1	607642	182290	Smith-Magenis syndrome	AD
226	RAPSN	601592	208150	Fetal akinesia deformation sequence	AR
227	RB1	614041	180200	Retinoblastoma	AD
228	RDH12	608830	612712	Leber congenital amaurosis 13	AR
229	RET	164761	191830	Renal agenesis	AR
230	RHO	180380	136880	Retinitis punctata albescens	AR
231	ROR2	602337	269310	Robinow syndrome, autosomal recessive	AR
232	RP9	607331	180104	Retinitis pigmentosa 9	AD
233	RPE65	180069	204100	Leber congenital amaurosis 2	AR
234	RPGR	312610	304020	Cone-rod dystrophy, X-linked, 1	XL
235	RPGRIP1	605446	608194	Cone-rod dystrophy 13	AR
236	RPL11	604175	612562	Diamond-Blackfan anemia 7	AD
237	RPL35A	180468	612528	Diamond-Blackfan anemia 5	AD
238	RPS10	603632	613308	Diamond-Blackfan anemia 9	AD
239	RPS19	603474	105650	Diamond-Blackfan anemia 1	AD
240	RPS24	602412	610629	Diamond-blackfan anemia 3	AD
241	RPS26	603701	613309	Diamond-Blackfan anemia 10	AD
242	RPS6KA3	300075	303600	Coffin-Lowry syndrome	XL
243	RPS7	603658	612563	Diamond-Blackfan anemia 8	AD
244	RS1	300839	312700	Retinoschisis	XL
245	RSPH4A	612647	612649	Ciliary dyskinesia, primary, 11	AR
246	RSPH9	612648	612650	Ciliary dyskinesia, primary, 12	AR
247	RYR1	180901	117000	Central core disease, Minicore myopathy with external ophthalmoplegia	AR, AD
248	RYR2	180902	600996	Arrhythmogenic right ventricular dysplasia 2, Ventricular tachycardia, catecholaminergic polymorphic, 1	AD
249	SALL4	607343	607323	Duane-radial ray syndrome, IVIC syndrome	AD
250	SCN1B	600235	612838	Brugada syndrome 5, Epilepsy, generalized, with febrile seizures plus, type 1	AD
251	SCN3B	608214	613120	Brugada syndrome 7	AD
252	SCN4B	608256	611819	Long QT syndrome-10	AD
253	SCN5A	600163	608567	Sick sinus syndrome 1	AR
254	SCN9A	603415	243000	Insensitivity to pain, congenital	AR
255	SEMA4A	607292	610283	Cone-rod dystrophy 10	AR
256	SERPINA1	107400	613490	Emphysema due to AAT deficiency	AR
257	SERPING1	606860	106100	Angioedema, hereditary, types I and II	AD
258	SGCD	601411	601287	Muscular dystrophy, limb-girdle, type 2F	AR
259	SH3BP2	602104	118400	Cherubism	AD
260	SIX1	601205	605192	Deafness, autosomal dominant 23, Brachiootic syndrome 3	AD
261	SIX5	600963	610896	Branchiootorenal syndrome 2	AD
262	SLC25A13	603859	605814	Citrullinemia, type II, neonatal-onset	AR
263	SLC25A4	103220	615418	Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type)	AR
264	SLC26A4	605646	600791	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	AR
265	SMAD3	603109	613795	Loeys-Dietz syndrome, type 3	AD
266	SMAD4	600993	175050	Polyposis, juvenile intestinal, Myhre syndrome	AD
267	SNCA	163890	168601	Parkinson disease 1 and 4	AD
268	SNRNP200	601664	610359	Retinitis pigmentosa 33	AD
269	SNTA1	601017	612955	Long QT syndrome 12	AD
270	SOD1	147450	105400	Amyotrophic lateral sclerosis 1	AR
271	SOS1	182530	610733	Noonan syndrome 4, Fibromatosis, gingival	AD
272	SOX9	608160	114290	Campomelic dysplasia, Campomelic dysplasia with autosomal sex reversal, Acampomelic campomelic dysplasia	AD
273	SPATA7	609868	604232	Leber congenital amaurosis 3	AR
274	SPG7	602783	607259	Spastic paraplegia 7, autosomal recessive	AR
275	TAF1	313650	314250	Dystonia-Parkinsonism, X-linked	XL
276	TAZ	300394	302060	Barth syndrome	XL
277	TBX5	601620	142900	Holt-Oram syndrome	AD
278	TCOF1	606847	154500	Treacher Collins syndrome 1	AD
279	TGFBR1	190181	609192	Loeys-Dietz syndrome, type 1	AD

280	TGFBR2	190182	610168	Loeys-Dietz syndrome, type 2	AD
281	TMEM43	612048	614302	Emery-Dreifuss muscular dystrophy 7, AD	AD
282	TNNI3	191044	611880	Cardiomyopathy, dilated, 2A	AR
283	TNNT1	191041	605355	Nemaline myopathy 5, Amish type	AR
284	TNXB	600985	606408	Ehlers-Danlos syndrome due to tenascin X deficiency	AR
285	TOPORS	609507	609923	Retinitis pigmentosa 31	AD
286	TP53	191170	202300	Adrenal cortical carcinoma	AR
287	TSC1	605284	191100	Tuberous sclerosis-1	AD
288	TSC2	191092	613254	Tuberous sclerosis-2	AD
289	TTPA	600415	277460	Ataxia with isolated vitamin E deficiency	AR
290	TTR	176300	105210	Amyloidosis, hereditary, transthyretin-related	AD
291	TULP1	602280	613843	Leber congenital amaurosis 15	AR
292	TWIST1	601622	101400	Saethre-Chotzen syndrome, Craniosynostosis type 1, Robinow-Sorauf syndrome	AD
293	TXNDC3	607421	610852	Ciliary dyskinesia, primary, 6	AR
294	TYR	606933	203100	Albinism, oculocutaneous, type IA	AR
295	USH1C	605242	276904	Usher syndrome, type 1C	AR
296	USH2A	608400	276901	Usher syndrome, type 2A	AR
297	VHL	608537	193300	von Hippel-Lindau syndrome	AD
298	WAS	300392	313900	Thrombocytopenia, X-linked	XL
299	WRN	604611	277700	Werner syndrome	AR
300	WT1	607102	256370	Nephrotic syndrome type 4, Denys-Drash syndrome	AR, AD

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.