PREQUEL® NIPT
FOR YOUR PEACE OF MIND

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	DISEASE
1.AAAS	Achalasia-addisonianism-alacrimia syndrome
2.ABCA12	Ichthyosis, autosomal recessive 4B (harlequin)
	Ichthyosis, congenital, autosomal recessive 4A
	Surfactant metabolism dysfunction, pulmonary, 3
3.ABCA4	Cone-rod dystrophy 3
	Fundus flavimaculatus
	Macular degeneration, age-related, 2
	Retinal dystrophy, early-onset severe
	Retinitis pigmentosa 19
	Stargardt disease 1
4.ABCB11	Cholestasis, benign recurrent intrahepatic, 2
	Cholestasis, progressive familial intrahepatic 2
5.ABCB4	Cholestasis, intrahepatic, of pregnancy, 3
	Cholestasis, progressive familial intrahepatic 3
	Gallbladder disease 1
6.ABCB7	Anemia, sideroblastic, with ataxia
7.ABCC2	Dubin-Johnson syndrome
8.ABCC6	Arterial calcification, generalized, of infancy, 2
	Pseudoxanthoma elasticum
	Pseudoxanthoma elasticum, forme fruste
9.ABCC8	Diabetes mellitus, noninsulin-dependent
	Diabetes mellitus, permanent neonatal
	Diabetes mellitus, transient neonatal 2
	Hyperinsulinemic hypoglycemia, familial, 1
	Hypoglycemia of infancy, leucine-sensitive
10.ABCD1	Adrenoleukodystrophy
11.ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type
12.ACAD8	Isobutyryl-CoA dehydrogenase deficiency
13.ACAD9	Acyl-CoA dehydrogenase 9 deficiency
14.ACADL	ACYL-CoA DEHYDROGENASE, LONG-CHAIN
15.ACADM	Medium chain acyl-CoA dehydrogenase deficiency
16.ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of
17.ACADSB	2-methylbutyrylglycinuria
18.ACADVL	Very long chain acyl-CoA dehydrogenase deficiency
19.ACAT1	Ketoacidosis due to beta-ketothiolase deficiency
20.ACE	Angiotensin I-converting enzyme, benign serum increase
	Microvascular complications of diabetes 3
	Myocardial infarction, susceptibility to
	Renal tubular dysgenesis
	SARS, progression of
	Stroke, hemorrhagic
21.ACOX1	Peroxisomal acyl-CoA oxidase deficiency
22.ACSF3	Combined malonic and methylmalonic aciduria
23.ACSL4	Mental retardation, X-linked 63
24.ACTN4	Glomerulosclerosis, focal segmental, 1
25.ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency
26.ADAMTS 13	Thrombotic thrombocytopenic purpura, familial

GENE	DISEASE
27.ADAMTS 2	Ehlers-Danlos syndrome, dermatosparaxis type
28.ADAMTS L2	Geleophysic dysplasia 1
29.ADCK3	Autosomal recessive ataxia due to ubiquinone deficiency
30.ADK	Hypermethioninemia due to adenosine kinase deficiency
31.AFF2	Mental retardation, X-linked, FRAXE type
32.AGA	Aspartylglucosaminuria
33.AGL	Glycogen storage disease due to glycogen debranching enzyme deficiency
34.AGPS	Rhizomelic chondrodysplasia punctata type 3
35.AGT	Hypertension, essential, susceptibility to
	Preeclampsia, susceptibility to
	Renal tubular dysgenesis
36.AGTR1	Hypertension, essential
	Renal tubular dysgenesis
37.AGTR2	ANGIOTENSIN II RECEPTOR, TYPE 2
38.AGXT	Hyperoxaluria, primary, type 1
39.AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
40.AHI1	Joubert syndrome with ocular defect
41.AIPL1	Cone-rod dystrophy
	Leber congenital amaurosis 4
	Retinitis pigmentosa, juvenile
42.AIRE	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia
43.ALAS2	Anemia, sideroblastic, 1
	Protoporphyria, erythropoietic, X-linked
44.ALDH3A2	Sjogren-Larsson syndrome
45.ALDH4A1	Hyperprolinemia, type II
46.ALDH5A1	4-hydroxybutyric aciduria
47.ALDH7A1	Epilepsy, pyridoxine-dependent
48.ALDOA	Glycogen storage disease XII
49.ALDOB	Hereditary fructose intolerance
50.ALG1	Congenital disorder of glycosylation type Ik
51.ALG12	Congenital disorder of glycosylation, type Ig
52.ALG2	Congenital disorder of glycosylation, type Ii
53.ALG3	Congenital disorder of glycosylation, type Id
54.ALG6	Congenital disorder of glycosylation type Ic
55.ALG8	Congenital disorder of glycosylation, type Ih
56.ALG9	Congenital disorder of glycosylation, type Il
57.ALMS1	Alström syndrome
58.ALPL	Childhood-onset hypophosphatasia
	Infantile hypophosphatasia
59.ALS2	Amyotrophic lateral sclerosis 2, juvenile
	Primary lateral sclerosis, juvenile
	Spastic paralysis, infantile onset ascending
60.AMACR	Alpha-methylacyl-Coa Racemase deficiency
	Congenital bile acid synthesis defect type 4
61.AMPD1	Myopathy due to myoadenylate deaminase deficiency
62.AMT	Glycine encephalopathy

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63.ANO5	Gnathodiaphyseal dysplasia
	Miyoshi muscular dystrophy 3
	Muscular dystrophy, limb-girdle, autosomal recessive
64.ANTXR2	Hyaline fibromatosis syndrome
65.AP1S1	MEDNIK syndrome
66.AP1S2	Mental retardation, X-linked syndromic 5
67.AP3B1	Hermansky-Pudlak syndrome 2
68.APTX	Ataxia - oculomotor apraxia type 1
69.AQP2	Diabetes insipidus, nephrogenic
70.AR	Complete androgen insensitivity syndrome
	Kennedy disease
	Partial androgen insensitivity syndrome
71.ARG1	Argininemia
72.ARHGEF6	Mental retardation, X-linked 46
73.ARHGEF9	Epileptic encephalopathy, early infantile, 8
74.ARL13B	Joubert syndrome 8
75.ARL6	Bardet-Biedl syndrome 1, modifier of
	Bardet-Biedl syndrome 3
	Retinitis pigmentosa 55
76.ARSA	Metachromatic leukodystrophy
77.ARSB	Mucopolysaccharidosis type 6
78.ARSE	Brachytelephalangic chondrodysplasia punctata
79.ARSF	ARYLSULFATASE F
80.ARX	Early infantile epileptic encephalopathy
81.ASL	Argininosuccinic aciduria
82.ASNS	Asparagine synthetase deficiency
83.ASPA	Canavan disease
84.ASPM	Microcephaly 5, primary, autosomal recessive
85.ASS1	Citrullinemia type I
86.ATIC	AICA-ribosiduria due to ATIC deficiency
87.ATM	Ataxia-telangiectasia
88.ATP6AP2	Mental retardation, X-linked, syndromic, Hedera type
	Parkinsonism with spasticity, X-linked
89.ATP6V0A 2	Cutis laxa, autosomal recessive, type IIA
	Wrinkly skin syndrome
90.ATP6V1B 1	Renal tubular acidosis with deafness
91.ATP7A	Menkes disease
	Occipital horn syndrome
	X-linked distal spinal muscular atrophy
92.ATP7B	Wilson disease
93.ATP8B1	Cholestasis, benign recurrent intrahepatic
	Cholestasis, intrahepatic, of pregnancy, 1
	Cholestasis, progressive familial intrahepatic 1
94.ATR	Seckel syndrome
95.ATRX	Alpha-thalassemia myelodysplasia syndrome, somatic
	Alpha-thalassemia/mental retardation syndrome
	Mental retardation-hypotonic facies syndrome, X-linked

GENE	DISEASE
96.AUH	3-methylglutaconic aciduria type 1
97.B4GALT1	Congenital disorder of glycosylation type 2d
98.B9D2	Joubert syndrome 34
	Meckel syndrome 10
99.BBS1	Bardet-Biedl syndrome 1
100.BBS10	Bardet-Biedl syndrome 10
101.BBS12	Bardet-Biedl syndrome 12
102.BBS2	Bardet-Biedl syndrome 2Z
	Retinitis pigmentosa 74
103.BCHE	Apnea, postanesthetic, susceptibility to, due to BCHE deficiency
	Butyrylcholinesterase deficiency
104.BCKDHA	Maple syrup urine disease (gene BCKDHA)
105.BCKDHB	Maple syrup urine disease (gene BCKDHB)
106.BCOR	Microphthalmia, syndromic 2
107.BCS1L	Björnstad syndrome
	GRACILE syndrome
	Isolated CoQ-cytochrome C reductase deficiency
	Leigh syndrome
108.BEST1	Bestrophinopathy, autosomal recessive
	Macular dystrophy, vitelliform, 2
	Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma
	Retinitis pigmentosa, concentric
	Retinitis pigmentosa-50
	Vitreoretinochoroidopathy
109.BLM	Bloom syndrome
110.BRCA2	Breast cancer, male, susceptibility to
	Breast-ovarian cancer, familial, 2
	Fanconi anemia, complementation group D1
	Glioblastoma 3
	Medulloblastoma
	Pancreatic cancer 2
	Prostate cancer
	Wilms tumor
111.BRIP1	Breast cancer, early-onset, susceptibility to
	Fanconi anemia, complementation group J
	Myasthenic syndrome, fast-channel congenital
	Myasthenic syndrome, slow-channel congenital
112.BRWD3	Mental retardation, X-linked 93
113.BSCL2	Encephalopathy, progressive, with or without lipodystrophy
	Lipodystrophy, congenital generalized, type 2
	Neuropathy, distal hereditary motor, type VA
	Silver spastic paraplegia syndrome
114.BSND	Bartter syndrome, type 4a
	Sensorineural deafness with mild renal dysfunction
115.BTD	Biotinidase deficiency
116.BTK	Isolated growth hormone deficiency type III
	X-linked agammaglobulinemia

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117.C10orf2	Infantile onset spinocerebellar ataxia
118..3	C3 deficiency
	Hemolytic uremic syndrome, atypical, susceptibility to, 5
	Macular degeneration, age-related, 9
119.CA2	Osteopetrosis with renal tubular acidosis
120.CANT1	Desbuquois dysplasia 1
	Epiphyseal dysplasia, multiple, 7
121.CAPN3	Muscular dystrophy, limb-girdle, autosomal dominant 4
	Muscular dystrophy, limb-girdle, autosomal recessive 1
122.CASK	FG syndrome 4
	Mental retardation and microcephaly with pontine and cerebellar hypoplasia
	Mental retardation, with or without nystagmus
123.CASP10	Autoimmune lymphoproliferative syndrome, type II
124.CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 2
125.CBS	Classical homocystinuria
126.CC2D2A	COACH syndrome
	Joubert syndrome 9
	Meckel syndrome 6
127.CCDC103	Ciliary dyskinesia, primary, 17
128.CCDC39	Ciliary dyskinesia, primary, 14
129.CD19	Immunodeficiency, common variable, 3
130.CD247	Immunodeficiency 25
131.CD2AP	Glomerulosclerosis, focal segmental, 3
132.CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect
133.CD3D	Immunodeficiency 19
134.CD3E	Immunodeficiency 18, SCID variant
135.CD3G	Immunodeficiency 17, CD3 gamma deficient
136.CD40LG	X-linked hyper-IgM syndrome
137.CDH23	Autosomal recessive nonsyndromic sensorineural deafness type DFNB12
138.CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy
	Hypotrichosis, congenital, with juvenile macular dystrophy
139.CDHR1	Cone-rod dystrophy 15
	Retinitis pigmentosa 65
140.CDK5RAP 2	Microcephaly 3, primary, autosomal recessive
141.CDKL5	Epileptic encephalopathy, early infantile, 2
142.CENPJ	Microcephaly 6, primary, autosomal recessive
	Seckel syndrome 4
143.CEP152	Microcephaly 9, primary, autosomal recessive
	Seckel syndrome 5
144.CEP290	Joubert syndrome with oculorenal defect 5
	Senior-Loken syndrome
145.CERKL	Retinitis pigmentosa 26
146.CFH	Basal laminar drusen
	Complement factor H deficiency
	Hemolytic uremic syndrome, atypical, susceptibility to, 1
	Macular degeneration, age-related, 4
147.CFP	Properdin deficiency, X-linked

GENE	DISEASE
148.CFTR	Cystic fibrosis; mucoviscidosis
149.CHM	Choroideremia
150.CHRNA1	Multiple pterygium syndrome, lethal type
	Myasthenic syndrome, fast-channel congenital
	Myasthenic syndrome, slow-channel congenital
151.CHRND	Multiple pterygium syndrome, lethal type
	Myasthenic syndrome, fast-channel congenital
	Myasthenic syndrome, slow-channel congenital
152.CHRNE	Myasthenic syndrome, congenital, 4A, slow-channel
	Myasthenic syndrome, congenital, 4B, fast-channel
	Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency
153.CHRNG	Escobar syndrome
	Multiple pterygium syndrome, lethal type
	Myasthenia gravis, neonatal transient
154.CHST6	Macular corneal dystrophy
155.CIITA	Bare lymphocyte syndrome, type II, complementation group A
	Rheumatoid arthritis, susceptibility to
156.CLCN1	Myotonia congenita, dominant
	Myotonia congenita, recessive
	Myotonia levior, recessive
157.CLCN5	Dent disease
	Hypophosphatemic rickets
	Nephrolithiasis, type I
	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis 308990
158.CLCN7	Autosomal recessive malignant osteopetrosis 4
159.CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis 607626
160.CLDN14	Deafness, autosomal recessive 29
161.CLDN19	Familial hypomagnesemia - hypercalciuria - nephrocalcinosis - severe ocular involvement
162.CLN3	Juvenile neuronal ceroid lipofuscinosis 3
163.CLN5	Late infantile neuronal ceroid lipofuscinosis 5
164.CLN6	Adult neuronal ceroid lipofuscinosis 4A
	Late infantile neuronal ceroid lipofuscinosis 6
165.CLN8	Late infantile neuronal ceroid lipofuscinosis 8
	Progressive epilepsy - intellectual deficit, Finnish type
166.CLRN1	Usher syndrome type 3A
167.CNGA1	Retinitis pigmentosa 49
168.CNGA3	Achromatopsia 2
169.CNGB1	Retinitis pigmentosa 45
170.CNGB3	Achromatopsia 3
	Macular degeneration, juvenile
171.COG1	Congenital disorder of glycosylation, type IIg
172.COG7	Congenital disorder of glycosylation, type IIe
173.COG8	Congenital disorder of glycosylation, type IIh
174. COL11A1	Fibrochondrogenesis 1
	Lumbar disc herniation, susceptibility to
	Marshall syndrome

GENE	DISEASE
175. OL17A1	Generalized junctional epidermolysis bullosa, non-Herlitz type
176.COL18A1	Knobloch syndrome, type 1
177.COL1A1	Caffey disease
	Ehlers-Danlos syndrome, type I
	Ehlers-Danlos syndrome, type VIIA
	Osteogenesis imperfecta, type I
	Osteogenesis imperfecta, type II
	Osteogenesis imperfecta, type III
	Osteogenesis imperfecta, type IV
178.COL1A2	Ehlers-Danlos syndrome, cardiac valvular type
179.COL2A1	Achondrogenesis, type II or hypochondrogenesis
	Avascular necrosis of the femoral head
	Czech dysplasia
	Epiphyseal dysplasia, multiple, with myopia and deafness
	Kniest dysplasia
	Legg-Calve-Perthes disease
	Osteoarthritis with mild chondrodysplasia
	Platyspondylic skeletal dysplasia, Torrance type
	SED congenita
	SMED Strudwick type
	Spondyloepiphyseal dysplasia, Stanescu type
	Spondyloperipheral dysplasia
	Stickler sydrome, type I, nonsyndromic ocular
	Stickler syndrome, type I
	Vitreoretinopathy with phalangeal epiphyseal dysplasia
180.COL4A3	Alport syndrome autosomal recessive (gene COL4A3)
181.COL4A4	Alport syndrome autosomal recessive (gene COL4A4)
182.COL4A5	Alport syndrome
183.COL6A1	Bethlem myopathy
	Ullrich congenital muscular dystrophy
184.COL6A2	Bethlem myopathy
	Ullrich congenital muscular dystrophy
185.COL6A3	Bethlem myopathy
	Ullrich congenital muscular dystrophy
186.COL7A1	Dystrophic epidermolysis bullosa pruriginosa
	Severe generalized recessive dystrophic epidermolysis bullosa
187.COL9A1	Epiphyseal dysplasia, multiple, 6
	Stickler syndrome, type IV
188.COL9A2	Epiphyseal dysplasia, multiple, 2
	Stickler syndrome, type V
189.COQ2	Leigh syndrome with nephrotic syndrome
190.COQ9	Coenzyme Q10 deficiency, primary, 5
191.COX10	Leigh syndrome due to mitochondrial COX4 deficiency
	Mitochondrial complex IV deficiency
192.COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2
	Leigh syndrome due to cytochrome c oxidase deficiency
193.COX6B1	Mitochondrial complex IV deficiency

GENE	DISEASE
194.CPS1	Carbamoylphosphate synthetase deficiency
195.CPT1A	Carnitine palmitoyl transferase 1A deficiency
196.CPT2	Carnitine palmitoyl transferase II deficiency, infantile form
	Carnitine palmitoyl transferase II deficiency, neonatal form
197.CRB1	Leber congenital amaurosis 8
	Pigmented paravenous chorioretinal atrophy
	Retinitis pigmentosa-12
198.CRLF1	Cold-induced sweating syndrome
199.CRTAP	Osteogenesis imperfecta type VII
200.CRX	Cone-rod retinal dystrophy-2
	Leber congenital amaurosis 7
201.CSTB	Unverricht-Lundborg disease
202.CTH	Cystathioninuria
	Homocysteine, total plasma, elevated
203.CTNS	Cystinosis
204.CTSC	Haim-Munk syndrome
	Papillon-Lefevre syndrome
	Periodontitis 1, juvenile
205.CTSD	Adult neuronal ceroid lipofuscinosis 10
206.CTSK	Pycnodysostosis
207.CUL4B	Mental retardation, X-linked, syndromic 15 (Cabezas type)
208.CYBA	Chronic granulomatous disease, autosomal, due to deficiency of CYBA
209.CYBB	Chronic granulomatous disease, X-linked
	Immunodeficiency 34, mycobacteriosis, X-linked
210.CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete
211.CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency
	Aldosteronism, glucocorticoid-remediable
212.CYP11B2	Aldosterone to renin ratio raised
	Hypoaldosteronism, congenital, due to CMO I deficiency
	Hypoaldosteronism, congenital, due to CMO II deficiency
	Low renin hypertension, susceptibility to
213.CYP17A1	17-alpha-hydroxylase/17,20-lyase deficiency
214.CYP19A1	Aromatase deficiency
	Aromatase excess syndrome
215.CYP1B1	Anterior segment dysgenesis 6, multiple subtypes
	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset
216.CYP27A1	Cerebrotendinous xanthomatosis
217.CYP27B1	Vitamin D-dependent rickets, type I
218.CYP4V2	Bietti crystalline corneoretinal dystrophy
219.CYP7B1	Bile acid synthesis defect, congenital, 3
	Spastic paraplegia 5A, autosomal recessive
220.D2HGDH	D-2-hydroxyglutaric aciduria
221.DBT	Classic maple syrup urine disease
222.DCLRE1C	Omenn syndrome
	Severe combined immunodeficiency due to DCLRE1C deficiency
223.DCX	Lissencephaly, X-linked
	Subcortical laminal heteropia, X-linked

GENE	DISEASE
224.DDB2	Xeroderma pigmentosum complementation group E
225.DDC	Aromatic L-amino acid decarboxylase deficiency
226.DFNB59	Deafness, autosomal recessive 59
227.DGUOK	Mitochondrial DNA depletion syndrome, hepatocerebral form due toDGUOK deficiency 3
228.DHCR24	Desmosterolosis
229.DHCR7	Smith-Lemli-Opitz syndrome
230.DHDDS	Congenital disorder of glycosylation, type 1bb
	Developmental delay and seizures with or without movement abnormalities
	Retinitis pigmentosa 59
231.DKC1	Dyskeratosis congenita X-linked
	Hoyeraal-Hreidarsson syndrome
232.DLD	Leigh syndrome
	Maple syrup urine disease
233.DLG3	Mental retardation, X-linked 90
234.DLL3	Autosomal recessive spondylocostal dysostosis 1
235.DMD	Becker muscular dystrophy
	Duchenne muscular dystrophy
236.DMP1	Autosomal recessive hypophosphatemic rickets 1
237.DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus
238.DNAI1	Ciliary dyskinesia, primary, 1, with or without situs inversus
239.DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus
240.DNAJC19	Dilated cardiomyopathy with ataxia
241.DNAL1	Ciliary dyskinesia, primary, 16
242.DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1
243.DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive
244.DOK7	Fetal akinesia deformation sequence
	Myasthenia, limb-girdle, familial
245.DOLK	Congenital disorder of glycosylation, type Im
246.DPAGT1	Congenital disorder of glycosylation type 1j
247.DPM1	Congenital disorder of glycosylation type 1e
248.DPYD	Dihydropyrimidine dehydrogenase deficiency
249.DSP	Lethal acantholytic epidermolysis bullosa
250.DUOX2	Thyroid dyshormonogenesis 6
251.DUOXA2	Thyroid dyshormonogenesis 5
252.DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly
253.DYSF	Miyoshi muscular dystrophy 1
	Muscular dystrophy, limb-girdle, autosomal recessive 2
	Myopathy, distal, with anterior tibial onset
254.EDA	Muscular dystrophy, limb-girdle, autosomal recessive 2
	Ectodermal dysplasia 1, hypohidrotic, X-linked
	Tooth agenesis, selective, X-linked 1
255.EDAR	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant
	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive
	Hair morphology 1, hair thickness
256.EDN3	Waardenburg-Shah syndrome 4B

GENE	DISEASE
257.EDNRB	ABCD syndrome
	Waardenburg-Shah syndrome 4A
258.EFEMP2	Cutis laxa, autosomal recessive, type IB
259.EFNB1	Craniofrontonasal dysplasia
260.EGR2	Charcot-Marie-Tooth disease type 4E
261.EIF2AK3	Wolcott-Rallison syndrome
262.EIF2B5	Leukoencephalopathy with vanishing white matter
	Ovarioleukodystrophy
263.ELK1	MEMBER OF ETS ONCOGENE FAMILY
264.EMD	Emery-Dreifuss muscular dystrophy 1, X-linked
265.ENO3	Glycogen storage disease XIII
266.ENPP1	Autosomal recessive hypophosphatemic rickets 2
267.EPM2A	Epilepsy, progressive myoclonic 2A (Lafora)
268.ERBB3	Lethal congenital contractural syndrome 2
269.ERCC2	Xeroderma pigmentosum/Cockayne syndrome complex complementation group D
270.ERCC3	Xeroderma pigmentosum/Cockayne syndrome complex complementation group B
271.ERCC4	Xeroderma pigmentosum/Cockayne syndrome complex complementation group F
272.ERCC5	Xeroderma pigmentosum/Cockayne syndrome complex complementation group G
273.ERCC6	Cockayne syndrome type B
	COFS syndrome 1
274.ERCC8	Cockayne syndrome type A
275.ESCO2	Roberts syndrome
276.ESPN	Deafness, autosomal recessive 36
	Deafness, neurosensory, without vestibular involvement, autosomal dominant
277.ESRRB	Deafness, autosomal recessive 35
278.ETFA	Glutaric acidemia type 2 (gene ETFA)
279..ETFB	Glutaric acidemia type 2 (gene ETFB)
280.ETFDH	Glutaric acidemia type 2 (gene ETFDH)
281.ETHE1	Ethylmalonic encephalopathy
282.EVC	Ellis-van Creveld syndrome
	Weyers acrodental dysostosis
283.EVC2	Ellis-van Creveld syndrome
284.EXOSC3	Pontocerebellar hypoplasia, type 1B
285.EYS	Retinitis pigmentosa 25
286.F11	Factor XI deficiency, autosomal dominant
	Factor XI deficiency, autosomal recessive
287.F2	Dysprothrombinemia
	Hypoprothrombinemia
	Pregnancy loss, recurrent, susceptibility to, 2
	Stroke, ischemic, susceptibility to
	Thrombophilia due to thrombin defect
288.F5	Budd-Chiari syndrome
	Factor V deficiency
	Pregnancy loss, recurrent, susceptibility to, 1
	Stroke, ischemic, susceptibility to
	Thrombophilia due to activated protein C resistance
	Thrombophilia, susceptibility to, due to factor V Leiden

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289.F8	Hemophilia A
290..F9	Hemophilia B
291.FAH	Tyrosinemia type 1
292.FAM126A	Hypomyelination - congenital cataract
293.FAM161A	Retinitis pigmentosa 28
294.FAM20C	Lethal osteosclerotic bone dysplasia
295.FANCA	Fanconi anemia, complementation group A
296.FANCB	Fanconi anemia, complementation group B
297.FANCC	Fanconi anemia complementation group C
298.FANCD2	Fanconi anemia, complementation group D2
299.FANCE	Fanconi anemia, complementation group E
300.FANCG	Fanconi anemia, complementation group G
301. FANCI	Fanconi anemia, complementation group I
302. FANCL	Fanconi anemia, complementation group L
303.FANCM	Premature ovarian failure 15
304.FAS	Spermatogenic failure 28
	Autoimmune lymphoproliferative syndrome, type IA
305.FASLG	Autoimmune lymphoproliferative syndrome, type IB
306.FASTKD2	Mitochondrial complex IV deficiency
307.FBLN5	Cutis laxa, autosomal dominant 2
	Cutis laxa, autosomal recessive, type IA
	Macular degeneration, age-related, 3
308.FERMT3	Leukocyte adhesion deficiency, type III
309.FGA	Congenital fibrinogen deficiency (gene FGA)
310.FGB	Afibrinogenemia, congenital/Hypofibrinogenemia, congenital
	Dysfibrinogenemia, congenital
311.FGD1	Aarskog-Scott syndrome
	Mental retardation, X-linked syndromic 16
312.FGD4	Charcot-Marie-Tooth disease type 4H
313.FH	Fumaric aciduria
314.FHL1	Uruguay faciocardiomusculoskeletal syndrome
	Emery-Dreifuss muscular dystrophy 6, X-linked
	Myopathy, X-linked, with postural muscle atrophy
	Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset
	Reducing body myopathy, X-linked 1b, with late childhood or adult onset
	Scapuloperoneal myopathy, X-linked dominant
315.FIG4	Amyotrophic lateral sclerosis 11
	Charcot-Marie-Tooth disease, type 4J
	Polymicrogyria, bilateral temporooccipital
	Yunis-Varon syndrome
316. FKRP	Autosomal recessive limb-girdle muscular dystrophy type 2I
	Congenital muscular dystrophy type 5B
	Muscle-eye-brain disease
317. FKTN	Autosomal recessive limb-girdle muscular dystrophy type 2M
	Congenital muscular dystrophy type 4B
	Fukuyama congenital muscular dystrophy

GENE	DISEASE
318.FLNA	Cardiac valvular dysplasia, X-linked
	Congenital short bowel syndrome
	FG syndrome 2
	Frontometaphyseal dysplasia 1
	Heterotopia, periventricular, 1
	Intestinal pseudoobstruction, neuronal
	Melnick-Needles syndrome
	Otopalatodigital syndrome, type I
	Otopalatodigital syndrome, type II
	Terminal osseous dysplasia
319.FLVCR1	Ataxia, posterior column, with retinitis pigmentosa
320.FMR1	Fragile X syndrome
	Fragile X tremor/ataxia syndrome
	Premature ovarian failure 1
321.FOLR1	Neurodegeneration due to cerebral folate transport deficiency
322.FOXG1	Rett syndrome, congenital variant
323.FOXN1	Severe T-cell immunodeficiency - congenital alopecia - nail dystrophy
324.FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked
325.FRAS1	Fraser syndrome (gene FRAS1)
326.FREM2	Fraser syndrome (gene FRAS2)
32.7.FTCD	Glutamate formiminotransferase deficiency
328.FTSJ1	Mental retardation, X-linked 9
329.FUCA1	Fucosidosis
330.FXN	Friedreich ataxia
	Friedreich ataxia with retained reflexes
331. G6PC	Glycogen storage disease due to glucose-6-phosphatase deficiency type 1a
332 6PC3	Síndrome de Dursun
323.G6PD	Favism
	Hemolytic anemia due to G6PD deficiency
334.GAA	Glycogen storage disease due to acid maltase deficiency
335.GALC	Krabbe disease
336.GALE	Galactose epimerase deficiency
337.GALK1	Galactokinase deficiency with cataracts
338.GALNS	Mucopolysaccharidosis IVA
339.GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, 1
340.GALT	Classic galactosemia
341.GAMT	Guanidinoacetate methyltransferase deficiency
342.GAN	Giant axonal neuropathy-1
343.GBA	Fetal Gaucher disease
	Gaucher disease type 2
	Gaucher disease type 3
	Gaucher disease type 3C
344.GBE1	Glycogen storage disease due to glycogen branching enzyme deficiency, childhood combined hepatic and myopathic form
345.GCDH	Glutaryl-CoA dehydrogenase deficiency
346.GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia
	Hyperphenylalaninemia, BH4-deficient, B

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	DISEASE
347.GCSH	Glycine encephalopathy
348.GDAP1	Autosomal dominant Charcot-Marie-Tooth disease type 2K
	Autosomal recessive Charcot-Marie-Tooth disease with hoarseness
	Autosomal recessive intermediate Charcot-Marie-Tooth disease type A
	Charcot-Marie-Tooth disease type 4A
349.GDF5	Acromesomelic dysplasia, Hunter-Thompson type
	Acromesomelic dysplasia, Hunter-Thompson type
	Brachydactyly, type A2
	Brachydactyly, type C
	Chondrodysplasia, Grebe type
	Du Pan syndrome
	Multiple synostoses syndrome 2
	Osteoarthritis-5
	Symphalangism, proximal, 1B
350.GDI1	Mental retardation, X-linked 41
351.GFM1	Hepatoencephalopathy due to combined oxidative phosphorylation deficiency type 1
352.GHRHR	Growth hormone deficiency, isolated, type IV
353.GJA1	Atrioventricular septal defect 3
	Craniometaphyseal dysplasia, autosomal recessive
	Erythrokeratodermia variabilis et progressiva 3
	Hypoplastic left heart syndrome 1
	Oculodentodigital dysplasia
	Oculodentodigital dysplasia, autosomal recessive
	Palmoplantar keratoderma with congenital alopecia
	Syndactyly, type III
354.GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1
355.GJB2	Autosomal recessive nonsyndromic sensorineural deafness type DFNB1A (gene GJB2)
355.GJB3	Deafness, autosomal dominant 2B
	Deafness, autosomal dominant, with peripheral neuropathy
	Deafness, autosomal recessive
	Deafness, digenic, GJB2/GJB3
	Erythrokeratodermia variabilis et progressiva 1
357.GJB6	Deafness, autosomal dominant 3B
	Deafness, autosomal recessive 1B
	Deafness, digenic GJB2/GJB6
	Ectodermal dysplasia 2, Clouston type
358.GJC2	Pelizaeus-Merzbacher-like due to GJC2 mutation
359.GK	Glycerol kinase deficiency
360.GLA	Fabry disease
361.GLB1	GM1 gangliosidosis type 1
	GM1 gangliosidosis type 2
	GM1 gangliosidosis type 3
	Mucopolysaccharidosis type 4B
362.GLDC	Glycine encephalopathy
363.GLE1	Lethal congenital contracture syndrome type 1
364.GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism
365.GM2A	GM2-gangliosidosis, AB variant

GENE	DISEASE
366.GNAS	ACTH-independent macronodular adrenal hyperplasia
	McCune-Albright syndrome, somatic, mosaic
	Osseous heteroplasia, progressive
	Pituitary adenoma 3, multiple types, somatic
	Pseudohypoparathyroidism Ia
	Pseudohypoparathyroidism Ib
	Pseudohypoparathyroidism Ic
	Pseudopseudohypoparathyroidism
367.GNE	Nonaka myopathy
	Sialuria
368.GNMT	Glycine N-methyltransferase deficiency
369.GNPTAB	Mucolipidosis type 2
	Mucolipidosis type 3
370.GNPTG	Mucolipidosis III gamma
371.GNRHR	Fertile eunuch syndrome
	Hypogonadotropic hypogonadism 7 without anosmia
372.GNS	Mucopolysaccharidosis type IIID
373. GORAB	Bernard-Soulier syndrome, type A1 (recessive)
	Bernard-Soulier syndrome, type A2 (dominant)
	Nonarteritic anterior ischemic optic neuropathy, susceptibility to
	von Willebrand disease, platelet-type
374.GP1BA	Bernard-Soulier syndrome, type A1 (recessive)
	Bernard-Soulier syndrome, type A2 (dominant)
	Nonarteritic anterior ischemic optic neuropathy, susceptibility to
	von Willebrand disease, platelet-type
375.GP1BB	Bernard-Soulier syndrome, type B
	Giant platelet disorder, isolated
376.GP9	Bernard-Soulier syndrome, type C
377.GPC3	Simpson-Golabi-Behmel syndrome, type 1
378.GPR143	Nystagmus 6, congenital, X-linked
	Ocular albinism, type I, Nettleship-Falls type
379.GPR179	Night blindness, congenital stationary (complete), 1E, autosomal recessive
380.GPR98	Usher syndrome type 2C
381.GRHPR	Hyperoxaluria, primary, type II
382.GRIA3	Mental retardation, X-linked 94
383.GRIK2	Mental retardation, autosomal recessive, 6
384.GRM6	Night blindness, congenital stationary (complete), 1B, autosomal recessive
385.GRXCR1	Deafness, autosomal recessive 25
386.GSS	Glutathione synthetase deficiency with 5-oxoprolinuria
387.GTF2H5	Trichothiodystrophy, complementation group A
388.GUCY2D	Choroidal dystrophy, central areolar 1
	Cone-rod dystrophy 6
	Leber congenital amaurosis 1
389..GUSB	Mucopolysaccharidosis type 7
390.HADH	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency
391.HADHA	Mitochondrial trifunctional protein deficiency
392.HADHB	Mitochondrial trifunctional protein deficiency

GENE	DISEASE
393.HAL	Histidinemia
394.HAMP	Histidinemia
395.HAX1	Neutropenia, severe congenital 3, autosomal recessive
396.HBA1	Alpha-thalassemia
397.HBA2	Erythrocytosis 7
	Heinz body anemia
	Hemoglobin H disease, deletional and nondeletional
	Thalassemia, alpha-
398.HBB	Beta-thalassemia
	Sickle cell anemia
399.HCCS	Linear skin defects with multiple congenital anomalies 1
400.HESX1	Combined pituitary hormone deficiencies, genetic forms
401.HEXA	Tay-Sachs disease
402.HEXB	Sandhoff disease
403. HFE	Alzheimer disease, susceptibility to
	Hemochromatosis
	Microvascular complications of diabetes 7
	Porphyria cutanea tarda, susceptibility to
	Porphyria variegata, susceptibility to
	Transferrin serum level QTL2
	Hemochromatosis, type 2A
404.HGD	Alkaptonuria
405.HGF	Deafness, autosomal recessive 39
406.HGSNAT	Sanfilippo syndrome type C
407.HIBCH	Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency
408.HLCS	Holocarboxylase synthetase deficiency
409.HMGCL	3-hydroxy-3-methylglutaric aciduria
410.HMOX1	Heme oxygenase-1 deficiency
	Pulmonary disease, chronic obstructive, susceptibility to
411.HOGA1	Hyperoxaluria, primary, type III
412.HP	Anhaptoglobinemia
	Hypohaptoglobinemia
413.HPD	Tyrosinemia type 3
414.HPRT1	Kelley-Seegmiller syndrome
	Lesch-Nyhan syndrome
415.HPS1	Hermansky-Pudlak syndrome 1
416.HPS3	Hermansky-Pudlak syndrome 3
417.HSD11B2	Apparent mineralocorticoid excess
418.HSD17B10	17-beta-hydroxysteroid dehydrogenase X deficiency
419. HSD17B3	Pseudohermaphroditism, male, with gynecomastia
420.HSD17B4	Bifunctional enzyme deficiency
	Perrault syndrome
421.HSD3B2	3-beta-hydroxysteroid dehydrogenase, type II, deficiency
422.HSPD1	Leukodystrophy, hypomyelinating, 4
	Spastic paraplegia 13, autosomal dominant
423.HSPG2	Schwartz-Jampel syndrome

GENE	DISEASE
424. TRA1	CARASIL syndrome
	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2
	Macular degeneration, age-related, 7
	Macular degeneration, age-related, neovascular type
425.HUWE1	Mental retardation, X-linked syndromic, Turner type
426.HYAL1	Mucopolysaccharidosis type IX
427.HYLS1	Hydrolethalus syndrome
428.ICOS	Immunodeficiency, common variable, 1
429.DH3B	Retinitis pigmentosa 46
430.IDS	Mucopolysaccharidosis type 2
431.IDUA	Mucopolysaccharidosis Ih
	Mucopolysaccharidosis Ih/s
	Mucopolysaccharidosis Is
432.IFNGR1	Immunodeficiency 27A, mycobacteriosis, AR
433.IFNGR2	Immunodeficiency 28, mycobacteriosis
434.IFT80	Jeune syndrome
435.IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia
436. GF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency
437.IGHMBP2	Spinal muscular atrophy with respiratory distress
438.IKBKAP	Familial dysautonomia
439.IKBKG	Ectodermal dysplasia, hypohidrotic, with immune deficiency
	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency
	Immunodeficiency 33
	Incontinentia pigmenti, type II
440.IL12B	Immunodeficiency 29, mycobacteriosis
441.IL12RB1	Immunodeficiency 30
442.IL1RAPL1	Mental retardation, X-linked 21/34
443.IL1RN	Interleukin 1 receptor antagonist deficiency
444.IL2RA	Diabetes, mellitus, insulin-dependent, susceptibility to, 10
	Immunodeficiency 41 with lymphoproliferation and autoimmunity
445.IL2RG	T-B+ severe combined immunodeficiency due to gamma chain deficiency
446.IMPDH1	Leber congenital amaurosis 11
	Retinitis pigmentosa 10
447.IMPG2	Macular dystrophy, vitelliform, 5
	Retinitis pigmentosa 56
448.INSR	Leprechaunism
449.INVS	Nephronophthisis 2, infantile
450.IQCB1	Senior-Loken syndrome 5
451.IQSEC2	Mental retardation, X-linked 1/78
452.ISCU	Myopathy with lactic acidosis, hereditary
453.ITGA6	Junctional epidermolysis bullosa - pyloric atresia
454.ITGB4	Junctional epidermolysis bullosa with piloric atresia
	Junctional epidermolysis bullosa, non-Herlitz type
455.IVD	Isovaleric acidemia

GENE	DISEASE
456. IYD	Thyroid dyshormonogenesis 4
457.AK3	T-B+ severe combined immunodeficiency due to JAK3 deficiency
458.KCNJ1	Antenatal Bartter syndrome
459.KCNJ11	Diabetes mellitus, transient neonatal, 3
	Diabetes mellitus, type 2, susceptibility to
	Diabetes, permanent neonatal, with or without neurologic features
	Hyperinsulinemic hypoglycemia, familial, 2
	Maturity-onset diabetes of the young, type 13
460.KCNJ13	Leber congenital amaurosis 16
	Snowflake vitreoretinal degeneration
461.KCNV2	Retinal cone dystrophy 3B
462.KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type
463..KIAA2022	Mental retardation, X-linked 98
464.KIF7	Acrocallosal syndrome
	Al-Gazali-Bakalinova syndrome
	Hydrolethalus syndrome 2
	Joubert syndrome 12
465.L1CAM	Corpus callosum hypoplasia-retardation-adducted thumbs-spasticity- hydrocephalus syndrome
	Masa syndrome
466.LAMA2	Congenital muscular dystrophy type 1A
467.LAMA3	Junctional epidermolysis bullosa, Herlitz type (gene LAMA3)
	Junctional epidermolysis bullosa, Herlitz type (gene LAMB3)
	Junctional epidermolysis bullosa, non-Herlitz type (gene LAMA3)
468.LAMB2	Nephrotic syndrome, type 5, with or without ocular abnormalities
	Pierson syndrome
469.LAMB3	Junctional epidermolysis bullosa, non-Herlitz type (gene LAMB3)
470.LAMC2	Junctional epidermolysis bullosa, Herlitz type (gene LAMC2
	unctional epidermolysis bullosa, non-Herlitz type (gene LAMC2)
471.LAMP2	Danon disease
472..LARGE	Congenital muscular dystrophy type 1D
	Muscle-eye-brain disease
473.LBR	Greenberg dysplasia
474.LCA5	Leber congenital amaurosis 5
475.LDHA	Glycogen storage disease XI
476.LDLR	Hypercholesterolemia, familial, 1
	LDL cholesterol level QTL2
477.LDLRAP1	Hypercholesterolemia, familial, 4
478.LEPRE1	Osteogenesis imperfecta type 8

GENE	DISEASE
479. LHCGR	Leydig cell adenoma, somatic, with precocious puberty
	Leydig cell hypoplasia with hypergonadotropic hypogonadism
	Leydig cell hypoplasia with pseudohermaphroditism
	Luteinizing hormone resistance, female
	Precocious puberty, male
480.LHFPL5	Deafness, autosomal recessive 67
481.LHX3	Combined pituitary hormone deficiency with spine abnormalities
482.LIFR	Stüve-Wiedemann syndrome
483.LIG4	Severe combined immunodeficiency with sensitivity to ionizing radiation
484.LIPA	Cholesteryl ester storage disease
	Wolman disease
485.LIPH	Hypotrichosis 7
	Woolly hair, autosomal recessive 2 with or without hypotrichosis
486.LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type
487.LMNA	Charcot-Marie-Tooth disease axonal type 2B1
	Lethal restrictive dermopathy
	Mandibuloacral dysplasia with type A lipodystrophy
488.LOXHD1	Deafness, autosomal recessive 77
489.LPL	Combined hyperlipidemia, familial
	High density lipoprotein cholesterol level QTL 11
	Lipoprotein lipase deficiency
490.LRAT	Leber congenital amaurosis 14
491.LRP2	Retinal dystrophy, early-onset severe
	Retinitis pigmentosa, juvenile
	Donnai-Barrow syndrome
492.LRP5	Bone mineral density variability 1
	Exudative vitreoretinopathy 4
	Hyperostosis, endosteal
	Osteopetrosis, autosomal dominant 1
	Osteoporosis
	Osteoporosis-pseudoglioma syndrome
	Osteosclerosis
	Polycystic liver disease 4 with or without kidney cysts
	van Buchem disease, type 2
493.LRPPRC	French-Canadian type Leigh syndrome
494.LRTOMT	Deafness, autosomal recessive 63
495.LYST	Chediak-Higashi syndrome

GENE	DISEASE
496. AGT1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia
497.MAK	Retinitis pigmentosa 62
498.MAN2B1	Mannosidosis, alpha-, types I and II
499.MARVEL D2	Deafness, autosomal recessive 49
500.MAT1A	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency
	Methionine adenosyltransferase deficiency, autosomal recessive
501.MATN3	Epiphyseal dysplasia, multiple, 5
	Osteoarthritis susceptibility 2
	Spondyloepimetaphyseal dysplasia
502.MBTPS2	Ichthyosis follicularis - alopecia - photophobia
503.MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency
504.MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency
505.MCEE	Methylmalonyl-CoA epimerase deficiency
506.MCOLN1	Mucolipidosis type 4
507.MCPH1	Microcephaly 1, primary, autosomal recessive
508.MECP2	Severe neonatal-onset encephalopathy with microcephaly
509.MED12	X-linked intellectual deficit with marfanoid habitus
510.MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy
511.MED25	Basel-Vanagait-Smirin-Yosef syndrome
	Charcot-Marie-Tooth disease, type 2B2
512.MEFV	Familial Mediterranean fever
513.MERTK	Retinitis pigmentosa 38
514.MESP2	Spondylocostal dysostosis 2, autosomal recessive
515.MFRP	Microphthalmia, isolated 5
516.MFSD8	Nanophthalmos 2
	Nanophthalmos 2
	Late infantile neuronal ceroid lipofuscinosis
517.MGAT2	Congenital disorder of glycosylation type 2a
518.MID1	Opitz GBBB syndrome, type I
519.MKKS	Bardet-Biedl syndrome 6
520.MKS1	McKusick-Kaufman syndrome
	McKusick-Kaufman syndrome
	Meckel syndrome type 1
521.MLC1	Megalencephalic leukoencephalopathy with subcortical cysts
522 MLYCD	Malonyl-CoA decarboxylase deficiency
523.MMAA	Vitamin B12-responsive methylmalonic acidemia type cblA
524.MMAB	Vitamin B12-responsive methylmalonic acidemia type cblB
525.MMACHC	Methylmalonic acidemia with homocystinuria, type cblC
	Methylmalonic acidemia with homocystinuria, type cblD
526.MMADHC	Homocystinuria, cblD type, variant 1
527.MOCS1	Methylmalonic aciduria and homocystinuria, cblD type
	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A (gene MOCS1)

GENE	DISEASE
528.MOCS2	Methylmalonic aciduria and homocystinuria, cblD type
	Methylmalonic aciduria, cblD type, variant 2
	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A (gene MOCS2)
529.MOGS	Congenital disorder of glycosylation, type IIb
530.MPDU1	Congenital disorder of glycosylation, type If
531.MPI	Congenital disorder of glycosylation type 1b
532.MPL	Thrombocythemia 2
	Thrombocytopenia, congenital amegakaryocytic
533.MPV17	Methylmalonic aciduria, cblD type, variant 2
	Navajo neurohepatopathy
534.MPZ	Charcot-Marie-Tooth disease, type 1B
	Charcot-Marie-Tooth disease, type 2I
	Charcot-Marie-Tooth disease, type 2J
	Dejerine-Sottas disease
	Neuropathy, congenital hypomyelinating
	Roussy-Levy syndrome
535.MRE11	Ataxia-telangiectasia-like disorder 1
536.MRPS16	Combined oxidative phosphorylation defect type 2
537.MRPS22	Combined oxidative phosphorylation defect type 5
538.MTHFR	Homocystinuria due to MTHFR deficiency
539.MTM1	X-linked centronuclear myopathy
540. MTMR2	Charcot-Marie-Tooth disease, type 4B1
541.MTR	Homocystinuria-megaloblastic anemia, cblG complementation type
	Neural tube defects, folate-sensitive, susceptibility to
542.MTRR	Homocystinuria-megaloblastic anemia, cbl E type
	Neural tube defects, folate-sensitive, susceptibility to
543.MTTP	Abetalipoproteinemia
544.MUT	Metabolic syndrome, protection against
	Vitamin B12-unresponsive methylmalonic acidemia type mut-
545.MVK	Mevalonic aciduria
546.MYD88	Macroglobulinemia, Waldenstrom
	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency
547.MYO15A	Deafness, autosomal recessive 3
548.MYO3A	Deafness, autosomal recessive 30
549.MYO5A	Griscelli disease type 1
550.MYO6	Deafness, autosomal dominant 22
551.MYO7A	Autosomal recessive nonsyndromic sensorineural deafness type DFNB2
	Usher syndrome type 1
552.NAGA	Kanzaki disease
553.NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B)

GENE	DISEASE
554.NAGS	Hyperammonemia due to N-acetylglutamate synthetase deficiency
555.NBN	Aplastic anemia
	Nijmegen breakage syndrome
556.NDP	Exudative vitreoretinopathy 2, X-linked
	Norrie disease
557.NDRG1	Charcot-Marie-Tooth disease, type 4D
558.NDUFA1	Mitochondrial complex I deficiency, nuclear type 12
	Mitochondrial complex I deficiency
559. NDUFA7	Mitochondrial complex I deficiency, nuclear type 12
560. NDUFAF2	Mitochondrial complex I deficiency, nuclear type 1
	Leigh syndrome
	Mitochondrial complex I deficiency
561.NDUFAF4	Mitochondrial complex I deficiency
562.NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16
563.NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency
	Mitochondrial complex I deficiency
564.NDUFS4	Leigh syndrome
	Mitochondrial complex I deficiency
565.NDUFS5	NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 5
566.NDUFS6	Complex I, mitochondrial respiratory chain, deficiency of
567.NDUFS7	Leigh syndrome
568.NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency
569.NDUFV1	Mitochondrial complex I deficiency
570.NEB	Nemaline myopathy 2
571.NEFL	Charcot-Marie-Tooth disease, dominant intermediate G
572.NEU. 1	Sialidosis, type I
	Sialidosis, type II
573. NEUROG 3	Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells
574.NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation
575.NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora)
576.NHP2	Dyskeratosis congenita, autosomal recessive 2
577.NHS	Cataract 40, X-linked
	Nance-Horan syndrome
578.NKX2-1	Chorea, hereditary benign
	Choreoathetosis, hypothyroidism, and neonatal respiratory distress
	Thyroid cancer, nonmedullary, 1
579.NKX2-5	Atrial septal defect 7, with or without AV conduction defects
	Conotruncal heart malformations, variable
	Hypoplastic left heart syndrome 2
	Hypothyroidism, congenital nongoitrous, 5
	Tetralogy of Fallot
580.NKX2-5	Ventricular septal defect 3

GENE	DISEASE
581.NLGN3	Asperger syndrome susceptibility, X-linked 1
	Autism susceptibility, X-linked 1
582.NLGN4X	Mental retardation, X-linked
583.NLRP7	Hydatidiform mole, recurrent, 1
584.NMNAT1	Leber congenital amaurosis 9
585.NOP10	Dyskeratosis congenita, autosomal recessive 1
586.NPC1	Niemann-Pick disease type C1
587.NPC2	Niemann-Pick disease type C2
588.NPHP1	Joubert syndrome 4
589.NPHP3	Renal-hepatic-pancreatic dysplasia
	Senior-Loken syndrome 1
590.NPHP4	Senior-Loken syndrome
591.NPHS1	Nephrotic syndrome, type 1
592.NPHS2	Nephrotic syndrome, type 2
593.NR0B1	46XY sex reversal 2, dosage-sensitive
594.NR2E3	Enhanced S-cone syndrome
	Retinitis pigmentosa 37
595.NR5A1	46XY sex reversal 3
	Adrenocortical insufficiency
596.NSD1	Beckwith-Wiedemann syndrome
	Sotos syndrome 1
597.NSDHL	CHILD syndrome
	CK syndrome
598.NSUN2	Mental retardation, autosomal recessive 5
599.NTRK1	Hereditary sensory and autonomic neuropathy type 4
600.NUP62	Infantile bilateral striatal necrosis
601.NXF5	NUCLEAR RNA EXPORT FACTOR 5
602.NYX	Night blindness, congenital stationary (complete), 1A, X-linked
603.OAT	Gyrate atrophy of choroid and retina with or without ornithinemia
604.OCA2	Albinism, brown oculocutaneous
	Albinism, oculocutaneous, type II
	Skin/hair/eye pigmentation 1, blond/brown hair
	Skin/hair/eye pigmentation 1, blue/nonblue eyes
605.OCRL	Dent disease 2Oc
	Oculocerebrorenal syndrome
606.OFD1	Simpson-Golabi-Behmel syndrome type 2
607.OPA3	3-methylglutaconic aciduria type 3
608.OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance
609.ORAI1	Immunodeficiency 9
	Myopathy, tubular aggregate, 2
610.OSTM1	Osteopetrosis, autosomal recessive 5
611.OTC	Ornithine transcarbamylase deficiency

COMPLETE GENETIC SCAN (CGS)

NON-INVASIVE PRENATAL SCREENING

GENE	DISEASE
612.OTOA	Deafness, autosomal recessive 22
613.OTO F	Auditory neuropathy, autosomal recessive, 1
614.OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency
615.PAH	Phenylketonuria
616.PAK3	Mental retardation, X-linked 30/47
617.PALB2	Fanconi anemia, complementation group N
618.PANK2	Pantothenate kinase-associated neurodegeneration
619.PAX3	Craniofacial-deafness-hand syndrome
	Rhabdomyosarcoma 2, alveolar
	Waardenburg syndrome, type 1
	Waardenburg syndrome, type 3
620.PAX6	Aniridia
	Anterior segment dysgenesis 5, multiple subtypes
	Cataract with late-onset corneal dystrophy
	Coloboma of optic nerve
	Coloboma, ocular
	Foveal hypoplasia 1
	Keratitis
	Morning glory disc anomaly
	Optic nerve hypoplasia
621.PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia
622.PC	Pyruvate carboxylase deficiency
623.PCBD1	Hyperphenylalaninemia, BH4-deficient, D
624.PCCA	Propionic acidemia (gene PCCA)
625.PCCB	Propionic acidemia (gene PCCB)
626.PCDH15	Deafness, autosomal recessive 23
	Usher syndrome, type 1D/F digenic
	Usher syndrome, type 1F
627.PCDH19	Epileptic encephalopathy, early infantile, 9
628.PDE6A	Retinitis pigmentosa 43
629.PDE6B	Night blindness, congenital stationary, autosomal dominant 2
	Retinitis pigmentosa-40
630.PDE6C	Cone dystrophy 4
631.PDE6G	Retinitis pigmentosa 57
632.PDHA1	Leigh syndrome, X-linked
633.PDHB	Pyruvate dehydrogenase E1-beta deficiency
634.PDHX	Lacticacidemia due to PDX1 deficiency
635.PDP1	Pyruvate dehydrogenase phosphatase deficiency
636.PDSS1	Deafness - encephaloneuropathy - obesity - valvulopathy
637.PDSS2	Leigh syndrome with nephrotic syndrome
638.PDX1	Diabetes mellitus, type II, susceptibility to
	MODY, type IV
	Pancreatic agenesis 1
639.PDZD7	Deafness, autosomal recessive 57
	Retinal disease in Usher syndrome type IIA, modifier of
	Usher syndrome, type IIC, GPR98/PDZD7 digenic

GENE	DISEASE
640.PEPD	Prolidase deficiency
641.PEX1	Zellweger syndrome 1A
642.PEX10	Peroxisome biogenesis disorder 6A (Zellweger)
	Peroxisome biogenesis disorder 6B
643.PEX12	Neonatal adrenoleukodystrophy (gene PEX12)
644.PEX13	Peroxisome biogenesis disorder 11A (Zellweger)
	Peroxisome biogenesis disorder 11B
645.PEX2	Peroxisome biogenesis disorder 5A (Zellweger)
646.PEX26	Peroxisome biogenesis disorder 5B
	Peroxisome biogenesis disorder 5B
	Neonatal adrenoleukodystrophy (gene PEX26)
	Zellweger syndrome 7A
647.PEX5	Neonatal adrenoleukodystrophy (gene PEX5)
648.PEX6	Heimler syndrome 2
649.PEX7	Peroxisome biogenesis disorder 4A (Zellweger)
	Peroxisome biogenesis disorder 4B
	Rhizomelic chondrodysplasia punctata type 1
650.PFKM	Glycogen storage disease VII
651.PGK1	Phosphoglycerate kinase 1 deficiency
652 PGM1	Congenital disorder of glycosylation, type It
653.PHF8	Mental retardation syndrome, X-linked, Siderius type
654.PHGDH	Neu-Laxova syndrome 1
	Phosphoglycerate dehydrogenase deficiency
655.PHKG2	Cirrhosis due to liver phosphorylase kinase deficiency
	Glycogen storage disease IXc
656.PHYH	Refsum disease
657.PKHD1	Autosomal recessive polycystic kidney disease
658.PKLR	Hemolytic anemia due to red cell pyruvate kinase deficiency
659.PLA2G6	Infantile neuroaxonal dystrophy 2A
	Infantile neuroaxonal dystrophy 2B
660.PLCE1	Nephrotic syndrome, tupe 3
661.PLEC	Epidermolysis bullosa simplex with muscular dystrophy
	Epidermolysis bullosa simplex with pyloric atresia
	Limb girdle dystrophy with epidermolysis bullosa simplex
662.PLEKHG5	Autosomal recessive distal spinal muscular atrophy type 4
663.PLG	Plasminogen deficiency type 1
664.PLOD1	Ehlers-Danlos syndrome type 6
665.PLP1	Spastic paraplegia type 2, X-linked
666.PMM2	Congenital disorder of glycosylation type 1a
667.PMP22	Charcot-Marie-Tooth disease, type 1A
	Charcot-Marie-Tooth disease, type 1E
	Dejerine-Sottas disease
	Roussy-Levy syndrome
668.PNPO	Pyridoxal phosphate-responsive seizures

GENE	DISEASE
669.POLG	Alpers syndrome
	Autosomal recessive progressive external ophthalmoplegia
	Mitochondrial neurogastrointestinal encephalomyopathy
	Sensory ataxic neuropathy - dysarthria - ophthalmoparesis
670.POLR1C	Leukodystrophy, hypomyelinating, 11
671.POMGNT 1	Treacher Collins syndrome 3
	Treacher Collins syndrome 3
	Autosomal recessive limb-girdle muscular dystrophy type C
	Congenital muscular dystrophy with cerebellar involvement
	Walker-Warburg syndrome (gene POMGNT1)
672.POMT1	Autosomal recessive limb-girdle muscular dystrophy type C
	Congenital muscular dystrophy with cerebellar involvement
	Walker-Warburg syndrome (gene POMT1)
673.POMT2	Autosomal recessive limb-girdle muscular dystrophy type C
	Congenital muscular dystrophy with cerebellar involvement
	Walker-Warburg syndrome (gene POMT2)
674.POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis
675.POU1F1	Combined pituitary hormone deficiencies, genetic forms
676.POU3F4	Deafness, X-linked 2
677.PPT1	Adult neuronal ceroid lipofuscinosis
678.PQBP1	Renpenning syndrome
679.PRCD	Retinitis pigmentosa 36
680.PRF1	Hemophagocytic lymphohistiocytosis, familial, 2
681.PRKRA	Dystonia 16
682.PRODH	Hyperprolinemia, type I
	Schizophrenia, susceptibility to, 4
683.PROM1	Cone-rod dystrophy 12
684.PROP1	Macular dystrophy, retinal, 2
	Combined pituitary hormone deficiencies, genetic forms
685.PRPS1	Retinitis pigmentosa 41
	Lethal ataxia with deafness and optic atrophy
	X-linked Charcot-Marie-Tooth disease type 5
686.PRSS12	Macular dystrophy, retinal, 2
	Retinitis pigmentosa 41
	Stargardt disease 4
	Stargardt disease 4
	Mental retardation, autosomal recessive 1
687.PRX	Charcot-Marie-Tooth disease type 4F
688.PSAP	Encephalopathy due to prosaposin deficiency
	Krabbe disease
	Metachromatic leukodystrophy

GENE	DISEASE
689.PSAT1	Phosphoserine aminotransferase deficiency
690.PTEN	Neu-Laxova syndrome 2
	Bannayan-Riley-Ruvalcaba syndrome
	Cowden syndrome 1
	Lhermitte-Duclos syndrome
	Macrocephaly/autism syndrome
691.PTH1R	Chondrodysplasia, Blomstrand type
	Eiken syndrome
	Failure of tooth eruption, primary
	Metaphyseal chondrodysplasia, Murk Jansen type
692.PTS	Hyperphenylalaninemia, BH4-deficient, A
693.PUS1	Myopathy, lactic acidosis, and sideroblastic anemia 1
694.PYGM	Glycogen storage disease due to muscle glycogen phosphorylase deficiency
695.QDPR	Hyperphenylalaninemia, BH4-deficient, C
696.RAB23	Carpenter syndrome
697.RAB27A	Griscelli disease type 2
698.RAB39B	Mental retardation, X-linked 72
699.RAB3GAP 1	Micro syndrome
700.RAB3GAP 2	Cataract - intellectual deficit - hypogonadism
701.RAD51C	Fanconi anemia, complementation group O
702.RAG1	Breast-ovarian cancer, familial, susceptibility to, 3
	Breast-ovarian cancer, familial, susceptibility to, 3
	Combined immunodeficiency with skin granulomas
	Omenn syndrome (gene RAG1)
	Severe combined immunodeficiency due to complete RAG1/2 deficiency
703.RAG2	Combined immunodeficiency with skin granulomas
	Omenn syndrome (gene RAG2)
	Severe combined immunodeficiency due to complete RAG1/2 deficiency
704.RAPSN	Fetal akinesia deformation sequence
705.RARS2	Pontocerebellar hypoplasia, type 6
706.RAX	Microphthalmia, isolated 3
707.RDH12	Leber congenital amaurosis 13
708.RDX	Deafness, autosomal recessive 24
709.RELN	Lissencephaly syndrome, Norman-Roberts type
710.REN	Hyperproreninemia
	Hyperuricemic nephropathy, familial juvenile 2
	Renal tubular dysgenesis
711.RFT1	Congenital disorder of glycosylation, type In
712.RGR	Retinitis pigmentosa
713.RHO	Night blindness, congenital stationary, autosomal dominant 1
	Retinitis pigmentosa 4, autosomal dominant or recessive
	Retinitis punctata albescens

GENE	DISEASE
714.RLBP1	Bothnia retinal dystrophy
	Fundus albipunctatus
	Newfoundland rod-cone dystrophy
	Retinitis punctata albescens
715.RMRP	Anauxetic dysplasia
	Cartilage-hair hypoplasia
	Metaphyseal dysplasia without hypotrichosis
716.RNASEH2 A	Aicardi-Goutieres syndrome 4
717.RNASEH2 B	Aicardi-Goutieres syndrome 2
718.RNASEH2 C	Aicardi-Goutieres syndrome 3
719.RP2	Retinitis pigmentosa 2
720.RPE65	Leber congenital amaurosis 2
	Retinitis pigmentosa 20
721.RPGR	Cone-rod dystrophy, X-linked, 1
	Macular degeneration, X-linked atrophic
	Retinitis pigmentosa 3
	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness
722.RPGRIP1L	Joubert syndrome with hepatic defect
	Meckel syndrome, type 5
723.RPL10	Autism, susceptibility to, X-linked 5
724.RPS6KA3	Coffin-Lowry syndrome
	Mental retardation, X-linked 19
725.RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)
	Mitochondrial DNA depletion syndrome 8B (MNGIE type)
726.RS1	Retinoschisis
727.RYR1	Central core disease
	King-Denborough syndrome
	Malignant hyperthermia susceptibility 1
	Minicore myopathy with external ophthalmoplegia
	Neuromuscular disease, congenital, with uniform type 1 fiber
728.SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay
729.SAG	Oguchi disease-1
	Retinitis pigmentosa 47
730.SAMD9	MIRAGE syndrome
	Tumoral calcinosis, familial, normophosphatemic
731.SAMHD1	Aicardi-Goutieres syndrome 5
	Chilblain lupus 2
732.SBDS	Shwachman-Diamond syndrome
733.SBF2	Charcot-Marie-Tooth disease, type 4B2

GENE	DISEASE
734.SC5DL	Lathosterolosis
735.SCN2A	Epileptic encephalopathy, early infantile, 11
	Seizures, benign familial infantile, 3
736.SCNN1A	Pseudohypoaldosteronism type 1, autosomal recessive (gene SCNN1A)
737.SCNN1B	Pseudohypoaldosteronism type 1, autosomal recessive (gene SCNN1B)
738.SCNN1G	Pseudohypoaldosteronism type 1, autosomal recessive (gene SCNN1G)
739.SCO1	Mitochondrial complex IV deficiency
740.SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1
741.SEMA4A	Cone-rod dystrophy 10
	Retinitis pigmentosa 35
742.SEPN1	Rigid spine syndrome
743.SEPSECS	Pontocerebellar hypoplasia type 2D
744.SERPINA1	Emphysema due to AAT deficiency
	Emphysema-cirrhosis, due to AAT deficiency
	Hemorrhagic diathesis due to antithrombin Pittsburgh
	Pulmonary disease, chronic obstructive, susceptibility to
745.SETX	Amyotrophic lateral sclerosis 4, juvenile
	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2
746.SFTPB	Surfactant metabolism dysfunction, pulmonary, 1
747.SFTPC	Surfactant metabolism dysfunction, pulmonary, 2
748.SGCA	Muscular dystrophy, limb-girdle, autosomal recessive 3
749.SGCB	Muscular dystrophy, limb-girdle, autosomal recessive 4
750.SGCD	Cardiomyopathy, dilated, 1L
	Muscular dystrophy, limb-girdle, autosomal recessive 6
751.SGCG	Muscular dystrophy, limb-girdle, autosomal recessive 5
752.SGSH	Mucopolysaccharidosis type 3A (Sanfilippo syndrome type A)
753.SH2D1A	X-linked lymphoproliferative disease
754.SH3TC2	Charcot-Marie-Tooth disease, type 4C
	Mononeuropathy of the median nerve, mild
755.SHROOM 4	Stocco dos Santos X-linked mental retardation syndrome
756.SIL1	Marinesco-Sjögren syndrome
757.SIX6	Optic disc anomalies with retinal and/or macular dystrophy
758.SLC12A1	Antenatal Bartter syndrome type 1
759.SLC12A3	Gitelman syndrome
760.SLC12A6	Corpus callosum agenesis - neuronopathy
761.SLC16A2	Allan-Herndon-Dudley syndrome
762.SLC17A5	Free sialic acid storage disease, infantile form
763.SLC19A2	Thiamine-responsive megaloblastic anemia syndrome
764.SLC22A5	Carnitine deficiency, systemic primary

GENE	DISEASE
765.SLC24A1	Night blindness, congenital stationary (complete), 1D, autosomal recessive
766.SLC25A13	Citrullinemia, adult-onset type II
	Citrullinemia, type II, neonatal-onset
767.SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria
768.SLC25A20	Carnitine-acylcarnitine translocase deficiency
769.SLC25A22	Early infantile epileptic encephalopathy
770.SLC26A2	Achondrogenesis type 1B
	Atelosteogenesis type II
	Diastrophic dwarfism
	Multiple epiphyseal dysplasia type 4
771.SLC26A3	Diarrhea 1, secretory chloride, congenital
772.SLC26A4	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct
	Pendred syndrome
773.SLC26A5	Deafness, autosomal recessive 61
774.SLC35A1	Congenital disorder of glycosylation type 2f
775.SLC35C1	Congenital disorder of glycosylation type 2c
776.SLC35D1	Schneckenbecken dysplasia
777.SLC37A4	Glycogen storage disease due to glucose-6-phosphatase deficiency type b
778.SLC37A4	Glycogen storage disease due to glucose-6-phosphatase deficiency type c
779.SLC39A4	Acrodermatitis enteropathica
780.SLC3A1	Cystinuria
781.SLC45A2	Albinism, oculocutaneous, type IV
	Skin/hair/eye pigmentation 5, black/nonblack hair
	Skin/hair/eye pigmentation 5, dark/fair skin
	Skin/hair/eye pigmentation 5, dark/light eyes
782.SLC46A1	Folate malabsorption, hereditary
783.SLC4A11	Congenital hereditary endothelial dystrophy type II
	Corneal dystrophy - perceptive deafness
784.SLC5A5	Folate malabsorption, hereditary
785.SLC6A19	Hartnup disorder
	Hyperglycinuria
	Iminoglycinuria, digenic
786.SLC6A8	X-linked creatine transporter deficiency
787.SLC7A7	Lysinuric protein intolerance
788.SLC7A9	Cystinuria
789.SLC9A6	Mental retardation, X-linked syndromic, Christianson type
790.SLX4	Fanconi anemia, complementation group P
791.SMARCAL1	Fanconi anemia, complementation group P
792.SMN1	Proximal spinal muscular atrophy type 1
	Proximal spinal muscular atrophy type 2
	Proximal spinal muscular atrophy type 3
	Proximal spinal muscular atrophy type 4

GENE	DISEASE
793.SMN2	Spinal muscular atrophy, type III, modifier of
794.SMPD1	Niemann-Pick disease type A
	Niemann-Pick disease type B
795.SMS	Mental retardation, X-linked, Snyder-Robinson type
796.SNAI2	Piebaldism
	Waardenburg syndrome, type 2D
797.SNAP29	Cerebral dysgenesis-neuropathy-ichthyosis- palmoplantar keratoderma syndrome
798.SOX3	Mental retardation, X-linked, with isolated growth hormone deficiency
	Panhypopituitarism, X-linked
799.SP110	Hepatic venoocclusive disease with immunodeficiency
800.SPG11	Amyotrophic lateral sclerosis 5, juvenile
	Charcot-Marie-Tooth disease, axonal, type 2X
	Spastic paraplegia 11, autosomal recessive
801.SPG20	Troyer syndrome
802.SPG7	Spastic paraplegia 7, autosomal recessive
803.SRD5A2	Pseudovaginal perineoscrotal hypospadias
804.SRD5A3	Congenital disorder of glycosylation, type Iq
	Kahrizi syndrome
805.SRPX2	Rolandic epilepsy, mental retardation, and speech dyspraxia
806.ST3GAL3	Epileptic encephalopathy, early infantile, 15
	Mental retardation, autosomal recessive 12
807.ST3GAL5	Amish infantile epilepsy syndrome
808.STAR	Congenital lipoid adrenal hyperplasia
809.STAT1	Immunodeficiency 31A, mycobacteriosis, autosomal dominant
	Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive
	Immunodeficiency 31C, autosomal dominant
810.STIL	Microcephaly 7, primary, autosomal recessive
811.STIM1	Immunodeficiency 10
	Myopathy, tubular aggregate, 1
	Stormorken syndrome
812.STRA6	Syndromic microphthalmia type 9
813.STRC	Deafness, autosomal recessive 16
814.STX11	Hemophagocytic lymphohistiocytosis, familial, 4
815.STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5
816.SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)
817.SUCLG1	Fatal infantile lactic acidosis with methylmalonic aciduria
818.SUMF1	Multiple sulfatase deficiency
819.SUOX	Sulfocysteinuria
820.SURF1	Leigh syndrome, due to COX deficiency
821.SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders
822.SYP	Mental retardation, X-linked 96

GENE	DISEASE
823.TAF1	Dystonia-Parkinsonism, X-linked
	Mental retardation, X-linked, syndromic 33
824.TAT	Tyrosinemia type 2
825.TAZ	Barth syndrome
826.TBCE	Hypoparathyroidism - intellectual deficit - dysmorphism syndrome
827.TCAP	Cardiomyopathy, hypertrophic, 25
	Muscular dystrophy, limb-girdle, autosomal recessive 7
828.TCF4	Pitt-Hopkins syndrome
829.TCIRG1	Autosomal recessive malignant osteopetrosis 1
830.TCN2	Transcobalamin II deficiency
831.TECTA	Deafness, autosomal dominant 8/12
	Deafness, autosomal recessive 21
832TERT	Dyskeratosis congenita, autosomal dominant 2
	Dyskeratosis congenita, autosomal recessive 4
	Leukemia, acute myeloid
	Melanoma, cutaneous malignant, 9
	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1
833TFR2	Hemochromatosis, type 3
834.TG	Autoimmune thyroid disease, susceptibility to, 3
	Thyroid dyshormonogenesis 3
835.TGM1	Ichthyosis, congenital, autosomal recessive 1
836.TH	Autosomal recessive dopa-responsive dystonia
837.THRA	Hypothyroidism, congenital, nongoitrous, 6
838.THRB	Thyroid hormone resistance
	Thyroid hormone resistance, autosomal recessive
	Thyroid hormone resistance, selective pituitary
839.TIMM8A	Mohr-Tranebjaerg syndrome
840.TK2	Mitochondrial DNA depletion syndrome, myopathic form
841.TLR3	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2
	HIV1 infection, resistance to
842.TMC1	Deafness, autosomal dominant 36
	Deafness, autosomal recessive 7
843.TMEM216	Joubert syndrome 2
	Meckel syndrome 2
844.TMEM67	COACH syndrome
	Joubert syndrome 6
845.TMIE	Deafness, autosomal recessive 6
846.TMPRSS3	Deafness, autosomal recessive 8/10
847.TNFRSF1 1B	Paget disease, juvenile
848.TNNT1	Nemaline myopathy 5, Amish type
849.TPO	Thyroid dyshormonogenesis 2A
850.TPP1	Neuronal ceroid lipofuscinosis 2

GENE	DISEASE
851.TPRN	Deafness, autosomal recessive 79
852.TRAPPC9	Mental retardation, autosomal recessive 13
853.TRDN	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness
854.TREX1	Aicardi-Goutières syndrome
855.TRIM32	Bardet-Biedl syndrome 11
	Muscular dystrophy, limb-girdle, autosomal recessive 8
856.TRIM37	MULIBREY nanism
857.TRIOBP	Deafness, autosomal recessive 28
858.TRMU	Deafness, mitochondrial, modifier of
	Liver failure, transient infantile
859.TSEN54	Pontocerebellar hypoplasia type 2A
	Pontocerebellar hypoplasia type 4
860.TSFM	Fatal mitochondrial disease due to combined oxidative phosphorylationdeficiency 3
861.TSHB	Isolated thyroid-stimulating hormone deficiency
862 TSHR	Hyperthyroidism, familial gestational
	Hyperthyroidism, nonautoimmune
	Hypothyroidism, congenital, nongoitrous, 1
	Thyroid adenoma, hyperfunctioning, somatic
	Thyroid carcinoma with thyrotoxicosis
863.TSPAN7	Mental retardation, X-linked 58
864.TSPYL1	Sudden infant death with dysgenesis of the testes syndrome
865.TTC37	Trichohepatoenteric syndrome 1
866.TTN	Cardiomyopathy, dilated, 1G
	Cardiomyopathy, familial hypertrophic, 9
	Muscular dystrophy, limb-girdle, autosomal recessive 10
	Myopathy, myofibrillar, 9, with early respiratory failure
	Salih myopathy
	Tibial muscular dystrophy, tardive
867.TTPA	Ataxia with vitamin E deficiency
868.TUBA1A	Lissencephaly 3
869. TUFM	Combined oxidative phosphorylation deficiency 4
870.TULP1	Leber congenital amaurosis 15
	Retinitis pigmentosa 14
871.TUSC3	ntal retardation, autosomal recessive 7
872.TYK2	Immunodeficiency 35
873.TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
874.TYR	Albinism, oculocutaneous, type IA
	Albinism, oculocutaneous, type IB
	Melanoma, cutaneous malignant, susceptibility to, 8
	Skin/hair/eye pigmentation 3, blue/green eyes
	Skin/hair/eye pigmentation 3, light/dark/freckling skin
	Waardenburg syndrome/albinism, digenic

GENE	DISEASE
875.TYRP1	Albinism, oculocutaneous, type III
	Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)
876.UBA1	X-linked spinal muscular atrophy type 2
877.UBE2A	Mental retardation, X-linked syndromic, Nascimento-type
878.UBE3A	Angelman syndrome
879.UBR1	Johanson-Blizzard syndrome
880.UGT1A1	Bilirubin, serum level of, QTL1
	Crigler-Najjar syndrome, type I
	Crigler-Najjar syndrome, type II
	Gilbert syndrome
	Hyperbilirubinemia, familial transient neonatal
881.UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3
882.UNC93B1	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1
883.UPF3B	Mental retardation, X-linked, syndromic 14
884.UQCRB	Mitochondrial respiratory chain complex III deficiency
885.UQCRQ	Mitochondrial respiratory chain complex III deficiency
886.UROS	Porphyria, congenital erythropoietic
887.USH1C	Autosomal recessive nonsyndromic sensorineural deafness type DFNB18
	Usher syndrome type 1C
888.USH1G	Usher syndrome type 1G
889.USH2A	Usher syndrome type 2A
890.USP9X	Mental retardation, X-linked 99
	Mental retardation, X-linked 99, syndromic, female-restricted
891. VDR	Vitamin D-dependent rickets type 2A
892.VLDLR	Cerebellar ataxia - intellectual deficit - dysequilibrium syndrome
893.VPS13A	Choreoacanthocytosis
894.VPS13B	Cohen Syndrome type 1
895.VPS33B	Arthrogryposis - renal dysfunction - cholestasis
896.VRK1	Pontocerebellar hypoplasia type 1A
897.VSX2	Microphthalmia with coloboma 3
	Microphthalmia, isolated 2
898.VWF	von Willebrand disease, type 1
	von Willebrand disease, types 2A, 2B, 2M, and 2N
	von Willibrand disease, type 3
899.WAS	Neutropenia, severe congenital, X-linked
	Thrombocytopenia, X-linked
	Thrombocytopenia, X-linked, intermittent
	Wiskott-Aldrich syndrome
900.WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations
901.WFS1	Wolfram syndrome 1
902.WISP3	Arthropathy, progressive pseudorheumatoid, of childhood
	Spondyloepiphyseal dysplasia tarda with progressive arthropathy

GENE	DISEASE
903.WNT10A	Odontoonychodermal dysplasia
	Schopf-Schulz-Passarge syndrome
	Tooth agenesis, selective, 4
904.WNT3	Tetra-amelia, autosomal recessive
905.WNT7A	Aplasia/hypoplasia of limbs and pelvis
	Fibular hypoplasia or aplasia - femoral bowing - oligodactyly
906.WRN	Werner syndrome
907.XIAP	Lymphoproliferative syndrome, X-linked, 2
908.XPA	Xeroderma pigmentosum complementation group A
909.XPC	Xeroderma pigmentosum, group C
910.ZDHHC9	Mental retardation, X-linked syndromic, Raymond type
911ZEB2	Mowat-Wilson syndrome
912.ZFYVE6	Spastic paraplegia 15, autosomal recessive
913.ZIC3	Congenital heart defects, nonsyndromic, 1, X-linked
	Heterotaxy, visceral, 1, X-linked
914.ZMPSTE24	Lethal restrictive dermopathy
	Mandibuloacral dysplasia with type B lipodystrophy
915. ZNF469	Brittle cornea syndrome
916. ZNF711	Mental retardation, X-linked 97