CARRIER SCREENING PANEL

BIOARRAY

1. WHAT IS THE CARRIER SCREENING PANEL?

The carrier screening panel is a genetic test that simultaneously analyses mutations in 298 genes which cause recessive inherited pathologies. Diseases with a recessive inheritance pattern, like cystic fibrosis, are those in which the affected subject inherits two mutations, one from each healthy carrier parent. The theoretical risk for these couples is 25%.

2. WHY TAKING A CARRIER SCREENING?

The purpose of this test is to know if a person is a healthy carrier of gene mutations related to recessive genetic diseases. The knowledge of someone having a carrier status is extremely valuable to determine the specific reproductive risk and to program suitable medical action aimed at avoiding such risk.

3. WHO IS THIS TEST FOR?

a. General population: couples without a personal or familial genetic disease background who, in a preconceptional context, wish to know their risk of conceiving a child affected by a specific recessive inherited disease.

b. Consanguineous couples, who have an increased risk of recessive disease occurrence in their offspring.

c. People of ethnic backgrounds in which some recessive diseases are more prevalent.

d. Couples that will undergo Assisted Reproduction treatment, as a complement to other diagnostic tests.

e. In Assisted Reproduction Units the test can be used for sperm/egg donors, for a better selection and risk reduction of recessive disease transmission.

Gene	Disease	OMIM
CYP17A1	17,20-lyase deficiency	202110
MCCC1	3-methylcrotonyl-CoA carboxylase 1 deficiency	210200
MCCC2	3-methylcrotonyl-CoA carboxylase 2 deficiency	210210
OPA3	3-methylglutaconic aciduria, type III	258501
MTTP	Abetalipoproteinemia	200100
SLC26A2	Achondrogenesis, type Ib	600972
CNGB3	Achromatopsia 3	262300
SLC39A4	Acrodermatitis enteropathica	201100
GDF5	Acromesomelic dysplasia	201250
ACADM	Acyl-coa dehydrogenase, medium-chain, deficiency of	201450
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of	201470
ACADVL	Acyl-CoA dehydrogenase, very long-chain, deficiency of	201475
HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	201810
CYP11B1	Adrenal hyperplasia, congenital, due to steroid 11-beta-hydroxylase deficiency	202010
CYP21A2	Adrenal hyperplasia, congenital / hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency	201910
ABCD1	Adrenoleukodystrophy	300100
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy	218000
TYR	Albinism, oculocutaneous, type IA	203100
TYR	Albinism, oculocutaneous, type IB	606952
SLC45A2	Albinism, oculocutaneous, type IV	606574
TYR	Albinism/waardenburg syndrome, digenic	103470
CYP11B1	Aldosteronism, glucocorticoid-remediable	103900
HGD	Alkaptonuria	203500
SERPINA1	Alpha-1-antitrypsin deficiency	613490
ACAT1	Alpha-methylacetoacetic aciduria	203750
COL4A3	Alport syndrome, recessive	203780
COL4A4	Alport syndrome, recessive	203780
COL4A5	Alport syndrome	301050
MPL	Amegakaryocytic thrombocytopenia, congenital	604498
LAMB3	Amelogenesis imperfecta, type Ia	104530
AR	Androgen insensitivity syndrome	300068, 312300
FGFR2	Antley-Bixler syndrome	207410
POR	Antley-Bixler syndrome	201750
ARG1	Argininemia	207800
ASL	Argininosuccinic aciduria	207900
CYP19A1	Aromatase deficiency	613546
SLC35A3	Arthrogryposis, mental retardation, and seizures	615553
PRPS1	Arts syndrome	301835
AGA	Aspartylglucosaminuria	208400
TTPA	Ataxia with isolated deficiency of vitamin E	277460

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CARRIER SCREENING PANEL 2/6

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Gene	Disease	OMIM
ATM	Ataxia-telangiectasia	208900
SLC26A2	Atelosteogenesis, type II / De la Chapelle dysplasia	256050
AIRE	Autoimmune polyendocrine syndrome, type I, with or without reversible metaphyseal dysplasia	240300
BBS1	Bardet-Biedl syndrome, 1	209900
BBS2	Bardet-Biedl syndrome, 2	615981
TRIM32	Bardet-Biedl syndrome 11	615988
BBS12	Bardet-Biedl syndrome 12	615989
CEP290	Bardet-Biedl syndrome 14	615991
BBS10	Bardet-Biedl syndrome, 10	615987
MKS1	Bardet-Biedl syndrome, 13	615990
CIITA	Bare lymphocyte syndrome, type II, complementation group A	209920
RFX5	Bare lymphocyte syndrome, type II	209920
RFXAP	Bare lymphocyte syndrome, type II	209920
BTD	Biotinidase deficiency	253260
BCS1L	Bjornstad syndrome	262000
BLM	Bloom syndrome	210900
RLBP1	Bothnia retinal dystrophy	607475
GDF5	Brachydactyly	615072, 112600, 113100
F5	Budd-Chiari syndrome	600880
ASPA	Canavan disease	271900
SGCD	Cardiomyopathy, dilated, 1L	606685
FKTN	Cardiomyopathy, dilated, 1X	611615
DMD	Cardiomyopathy, dilated, 3B	302045
SLC22A5	Carnitine deficiency, systemic primary	212140
CPT2	Carnitine palmitoyltransferase II deficiency	608649, 600836, 255110
RAB23	Carpenter syndrome	201000
SLC6A8	Cerebral creatine deficiency syndrome 1	300352
GAMT	Cerebral creatine deficiency syndrome 2	612736
CYP27A1	Cerebrotendinous xanthomatosis	213700
PPT1	Ceroid lipofuscinosis, neuronal 1	256730
TPP1	Ceroid lipofuscinosis, neuronal 2	204500
CLN3	Ceroid lipofuscinosis, neuronal 3	204200
CLN5	Ceroid lipofuscinosis, neuronal 5	256731
CLN6	Ceroid lipofuscinosis, neuronal 6	601780
MFSD8	Ceroid lipofuscinosis, neuronal 7	610951
CLN8	Ceroid lipofuscinosis, neuronal, 8	600143, 610003
CLN6	Ceroid lipofuscinosis type Kufs	204300
PRPS1	Charcot-Marie-Tooth disease 5	311070
GJB1	Charcot-Marie-Tooth disease, X-linked	302800
ABCB11	Cholestasis, progressive familial 2	601847
ABCB11	Cholestasis, benign recurrent intrahepatic, 2	605479

Gene	Disease	OMIM
GDF5	Chondrodysplasia, Grebe type	200700
VPS13A	Choreoacanthocytosis	200150
CHM	Choroideremia	303100
ASS1	Citrullinemia	215700
VPS13B	Cohen syndrome	216550
GUCY2D	Cone-rod dystrophy 6	601777
PROM1	Cone-rod dystrophy 12	612657
PMM2	Congenital disorder of glycosylation, type Ia	212065
MPI	Congenital disorder of glycosylation, type Ib	602579
ALG6	Congenital disorder of glycosylation, type Ic	602522 603147
SLC4A11	Corneal endothelial dystrophy	217400, 217700, 613268
FGFR2	Craniosynostosis, syndromic, dominant	101200, 123500, 101600, 123790, 614592, 123150, 609579
UGT1A1	Crigler-Najjar syndrome, types I, II	218800, 606785
CTH	Cystathioninuria	219500
CFTR	Cystic fibrosis	219700
CTNS	Cystinosis, nephropathic	219800, 219900
CTNS	Cystinosis, non-nephropathic	219750
SLC7A9	Cystinuria	220100
SLC3A1	Cystinuria	220100
HSD17B4	D-bifunctional protein deficiency	261515
CDH23	Deafness 12	601386
USH1C	Deafness 18A	602092
PCDH15	Deafness 23	609533
GJB2	Deafness, recessive 1a	220290
MYO7A	Deafness, recessive 2	600060
MYO15A	Deafness, recessive 3	600316
GJB2	Deafness, dominant 3a	601544
SLC26A4	Deafness, recessive 4	600791
MYO7A	Deafness, dominant 11	601317
PRPS1	Deafness, X-linked 1	304500
GCK	Diabetes permanent neonatal	606176
INS	Diabetes, insulin-dependent, 2	125852
GLIS3	Diabetes, neonatal, with congenital hypothyroidism	610199
GCK	Diabetes, noninsulin-dependent, late onset	125853
ABCC8	Diabetes, noninsulin-dependent	125853
ABCC8	Diabetes, permanent neonatal	606176
KCNJ11	Diabetes, permanent neonatal	606176
SLC26A3	Diarrhea 1, secretory chloride, congenital	214700
SLC26A2	Diastrophic dysplasia	222600

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Gene	Disease	OMIM
DLD	Dihydrolipoamide dehydrogenase deficiency	246900
DPYD	Dihydropyrimidine dehydrogenase deficiency	274270
POR	Disordered steroidogenesis due to cytochrome P450 oxidoreductase deficiency	613571
GDF5	Du Pan syndrome	228900
ABCC2	Dubin-Johnson syndrome	237500
F2	Dys/hypoprothrombinemia	613679
IKBKAP	Dysautonomia, familial	223900
RTEL1	Dyskeratosis congenita	615190
EDA	Ectodermal dysplasia 1, hypohidrotic	305100
ADAMTS2	Ehlers-Danlos syndrome, type VIIC	225410
EVC2	Ellis-van Creveld syndrome	225500
EVC	Ellis-van Creveld syndrome	225500
EMD	Emery-Dreifuss muscular dystrophy 1	310300
ETHE1	Encephalopathy, ethylmalonic	602473
NR2E3	Enhanced s-cone syndrome	268100
COL7A1	Epidermolysis bullosa dystrophica	226600, 132000, 131750
MMP1	Epidermolysis bullosa dystrophica, modifier of	226600
COL7A1	Epidermolysis bullosa pretibial	131850
COL7A1	Epidermolysis bullosa pruriginosa	604129
COL7A1	Epidermolysis bullosa transient of the newborn	131705
LAMA3	Epidermolysis bullosa, generalized atrophic benign	226650
LAMA3	Epidermolysis bullosa, junctional, Herlitz type	226700
LAMC2	Epidermolysis bullosa, junctional, Herlitz type	226700
LAMB3	Epidermolysis bullosa, junctional, Herlitz type	226700
LAMB3	Epidermolysis bullosa, junctional, non-Herlitz type	226650
LAMC2	Epidermolysis bullosa, junctional, non-Herlitz type	226650
SLC26A2	Epiphyseal dysplasia, multiple, 4	226900
CHRNG	Escobar syndrome	265000
GLA	Fabry disease	301500
F5	Factor V deficiency	227400
F11	Factor XI deficiency	231680 612416
MEFV	Familial mediterranean fever, recessive	249100
MEFV	Familial mediterranean fever, dominant	134610
FANCA	Fanconi anemia, complementation group A	227650
FANCC	Fanconi anemia, complementation group C	227645
FANCG	Fanconi anemia, complementation group G	614082
BRIP1	Fanconi anemia, complementation group J	609054
G6PD	Favism	134700
FMR1	Fragile X mental retardation syndrome	300624
FMR1	Fragile X tremor/ataxia syndrome	300623
ALDOB	Fructose intolerance	229600
FH	Fumarase deficiency	606812

Gene	Disease	OMIM
GALK1	Galactokinase deficiency with cataracts	230200
GALE	Galactose epimerase deficiency	230350
GALT	Galactosemia	230400
GBA	Gaucher disease, perinatal lethal types, I, II, III, IIIC	608013, 230800, 230900, 231000, 231005
SLC12A3	Gitelman syndrome	263800
GCDH	Glutaric acidemia I	231670
ETFA	Glutaric acidemia IIA	231680
ETFB	Glutaric acidemia IIB	231680
ETFDH	Glutaric acidemia IIC	231680
GNMT	Glycine N-methyltransferase deficiency	606664
G6PC	Glycogen storage disease Ia	232200
SLC37A4	Glycogen storage disease Ib	232220
SLC37A4	Glycogen storage disease Ic	232240
GAA	Glycogen storage disease II	232300
AGL	Glycogen storage disease IIIa, IIIb	232400
GBE1	Glycogen storage disease IV	232500
PFKM	Glycogen storage disease VII	232800
GLB1	GM1-gangliosidosis, types I, II, III	230500, 230600, 230650
GM2A	GM2-gangliosidosis, AB variant	272750
PRPS1	Gout/ phosphoribosylpyrophosphate synthetase superactivity	300661
BCS1L	GRACILE syndrome	603358
CYBB	Granulomatous disease, chronic	306400
PEX1	Heimler syndrome 1	234580
PEX6	Heimler syndrome 2	616617
HFE	Hemochromatosis	235200
HFE2	Hemochromatosis, type 2A	602390
TFR2	Hemochromatosis, type 3	604250
HBA2	Hemoglobin H disease	613978
HBA1	Hemoglobin H disease	613978
G6PD	Hemolytic anemia due to g6pd deficiency	300908
F8	Hemophilia A	306700
F9	Hemophilia B	306900
CPT1A	Hepatic carnitine palmitoyltransferase deficiency, type IA	255120
HPS1	Hermansky-Pudlak syndrome 1	203300
HPS3	Hermansky-Pudlak syndrome 3	614072
HAL	Histidinemia	235800
HLCS	Holocarboxylase synthetase deficiency	253270
CBS	Homocystinuria / hyperhomocysteinemic thrombosis	236200
MTHFR	Homocystinuria due to deficiency of methylenetetrahydrofolate reductase	236250
HMGCL	Hydroxymethylglutaryl-coa lyase deficiency	246450

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Gene	Disease	OMIM
MVK	Hyper IgD syndrome	260900
UGT1A1	Hyperbilirubinemia, transient familial neonatal	237900
AMT	Hyperglycinemia encephalopathy	605899
GLDC	Hyperglycinemia encephalopathy	605899
LIAS	Hyperglycinemia, lactic acidosis, and seizures	614462
ABCC8	Hyperinsulinemic hypoglycemia, familial, 1	256450
KCNJ11	Hyperinsulinemic hypoglycemia, familial, 2	601820
GCK	Hyperinsulinemic hypoglycemia, familial, 3	602485
LPL	Hyperlipidemia, familial combined	144250
AHCY	Hypermethioninemia with S-adenosylhomocysteine hydrolase deficiency	613752
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	238970
AGXT	Hyperoxaluria, primary, type I	259900
GRHPR	Hyperoxaluria, primary, type II	260000
HOGA1	Hyperoxaluria, primary, type III	613616
PTS	Hyperphenylalaninemia	261640
INS	Hyperproinsulinemia	616214
ALDH4A1	Hyperprolinemia type II	239510
CYP11B2	Hypoaldosteronism, congenital, due to cmo I deficiency	203400
CYP11B2	Hypoaldosteronism, congenital, due to cmo II deficiency	610600
ABCC8	Hypoglycemia, infantile	240800
ALPL	Hypophosphatasia	146300, 241510, 241500
AR	Hypospadias 1	300633
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia	218700
TSHR	Hypothyroidism, congenital, nongoitrous, 1	275200
TSHB	Hypothyroidism, congenital, resistant to TSH	275200
TGM1	Ichthyosis, congenital, 1	242300
CYBB	Immunodeficiency 34, mycobacteriosis	300645
SLC17A5	Infantile sialic acid storage disease	269920
IVD	Isovaleric acidemia	243500
TMEM216	Joubert syndrome 2	608091
MKS1	Joubert syndrome 28	617121
CEP290	Joubert syndrome 5	610188
ARL13B	Joubert syndrome 8	612291
GJB2	Keratitis-ichthyosis-deafness, dominant	148210, 602540
GJB2	Keratoderma, palmoplantar, with deafness	148350
GALC	Krabe disease	245200
FGFR2	LADD syndrome	149730
LAMA3	Laryngoonychocutaneous syndrome	245660
GUCY2D	Leber congenital amaurosis 1	204000
LCA5	Leber congenital amaurosis 5	604537
RDH12	Leber congenital amaurosis 13	612712

Gene	Disease	OMIM
LRPPRC	Leigh syndrome, french canadian type	220111
BCS1L	Leigh syndrome	256000
LHCGR	Leydig cell hypoplasia with hypergonadotropic hypogonadism / pseudohermaphroditism / LH resistance (female)	238320
DYSF	Limb-girdle muscular dystrophy, type 2B	253601
LMF1	Lipase deficiency	246650
STAR	Lipoid congenital adrenal hyperplasia	201710
LPL	Lipoprotein lipase deficiency	238600
SLC7A7	Lysinuric protein intolerance	222700
CNGB3	Macular degeneration, juvenile	248200
PROM1	Macular dystrophy 2	608051
MFSD8	Macular dystrophy with central cone involvement	616170
MAN2B1	Mannosidosis, Alpha B, types I, II	248500
BCKDHA	Maple syrup urine disease, type Ia	248600
BCKDHB	Maple syrup urine disease, type Ib	248600
DBT	Maple syrup urine disease, type II	248600
PYGM	McArdle disease	232600
TMEM216	Meckel syndrome	603194
MKS1	Meckel syndrome 1	249000
CEP290	Meckel syndrome 4	611134
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A	613925
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2B with/without mental retardation	613926
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts 1	604004
ATP7A	Menkes disease	309400
ARSA	Metachromatic leukodystrophy	250100
MAT1A	Methionine adenosyltransferase I/III deficiency	250850
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	277400
MMAB	Methylmalonic aciduria, cblB type	251110
MUT	Methylmalonic aciduria, mut(0) type	251000
MMAA	Methylmalonic aciduria, vitamin B12-responsive	251100
MMADHC	Methylmalonic aciduria/homocystinuria, type cblD, variants 1, 2	277410
MCEE	Methylmalonyl-CoA epimerase deficiency	251120
MVK	Mevalonic aciduria	610377
RFXANK	MHC class II deficiency, complementation group B	209920
POLG	Mitochondrial ataxia rec.	607459
BCS1L	Mitochondrial complex III deficiency, nuclear type 1	124000
POLG	Mitochondrial DNA depletion syndrome 4A	203700
POLG	Mitochondrial DNA depletion syndrome 4B	613662
MPV17	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	256810
DYSF	Miyoshi muscular dystrophy 1	254130
IL2RG	Moderate combined immunodeficiency	312863

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Gene	Disease	OMIM
INS	MODY type 10	613370
KCNJ11	MODY type 13	616329
GCK	MODY-II	125851
GNPTAB	Mucolipidosis alpha/beta II	252500
GNPTAB	Mucolipidosis alpha/beta III	252600
MCOLN1	Mucolipidosis IV	252650
IDUA	Mucopolysaccharidosis Ih/s	607015
IDUA	Mucopolysaccharidosis Ih	607014
IDUA	Mucopolysaccharidosis Is	607016
IDS	Mucopolysaccharidosis, type II	309900
SGSH	Mucopolysaccharidosis, type IIIA (Sanfilippo A)	252900
NAGLU	Mucopolysaccharidosis, type IIIB (sanfilippo B)	252920
HGSNAT	Mucopolysaccharidosis, type IIIC (Sanfilippo C)	252930
GNS	Mucopolysaccharidosis, type IIID	252940
GLB1	Mucopolysaccharidosis type IVB (Morquio)	253010
CHRND	Multiple pterygium syndrome, lethal type	253290
CHRNA1	Multiple pterygium syndrome, lethal type	253290
CHRNG	Multiple pterygium syndrome, lethal type	253290
SUMF1	Multiple sulfatase deficiency	272200
GDF5	Multiple synostoses syndrome 2	610017
DMD	Muscular dystrophy, Becker type	300376
DMD	Muscular dystrophy, Duchenne type	310200
CAPN3	Muscular dystrophy, limb-girdle, type 2A	253600
SGCG	Muscular dystrophy, limb-girdle, type 2C	253700
SGCA	Muscular dystrophy, limb-girdle, type 2D	608099
SGCB	Muscular dystrophy, limb-girdle, type 2E	604286
SGCD	Muscular dystrophy, limb-girdle, type 2F	601287
TRIM32	Muscular dystrophy, limb-girdle, type 2H	254110
POMT1	Muscular dystrophy-dystroglycanopathy, type A1	236670
POMGNT1	Muscular dystrophy-dystroglycanopathy type A3, B3, C3	253280, 613151, 613157
FKTN	Muscular dystrophy-dystroglycanopathy, type A4	253800
FKRP	Muscular dystrophy-dystroglycanopathy, type A5	613153
POMT1	Muscular dystrophy-dystroglycanopathy, type B1	613155
FKTN	Muscular dystrophy-dystroglycanopathy, type B4	613152
FKRP	Muscular dystrophy-dystroglycanopathy, type B5	606612
POMT1	Muscular dystrophy-dystroglycanopathy, type C1	609308
FKTN	Muscular dystrophy-dystroglycanopathy, type C4	611588
FKRP	Muscular dystrophy-dystroglycanopathy, type C5	607155
CHRNA1	Myasthenic syndrome, congenital, 1A, slow-channel	601462
CHRNA1	Myasthenic syndrome, congenital, 1B, fast-channel	608930
CHRND	Myasthenic syndrome, congenital, 3A, slow-channel	616321
CHRND	Myasthenic syndrome, congenital, 3B, fast-channel	616322
CHRND	Myasthenic syndrome, congenital, 3C, acetylcholine receptor deficiency	616323
AMPD1	Myopathy due to myoadenylate deaminase deficiency	615511

Gene	Disease	OMIM
ACTA1	Myopathy, congenital, with fiber-type disproportion	255310
MTM1	Myotubular myopathy	310400
NEB	Nemaline myopathy 2	256030
ACTA1	Nemaline myopathy 3, congenital	161800
INS	Neonatal diabetes	606176
NPHS1	Nephrotic syndrome, type 1	256300
NPHS2	Nephrotic syndrome, type 2	600995
PHGDH	Neu-laxova syndrome 1	256520
NME1	Neuroblastoma	256700
HAX1	Neutropenia, severe congenital, 3	610738
VPS45	Neutropenia, severe congenital, type 5	615285
SMPD1	Niemann-pick disease, type A	257200
SMPD1	Niemann-pick disease, type B	607616
NPC1	Niemann-Pick disease, types C1, D	257220
NPC2	Niemann-pick disease, type C2	607625
NBN	Nijmegen syndrome	251260
GNE	Nonaka myopathy	605820
ATP7A	Occipital horn syndrome	304150
DCLRE1C	Omenn syndrome	603554
OPA3	Optic atrophy 3 with cataract	165300
OTC	Ornithine transcarbamylase deficiency	311250
PROP1	Panhypopituitarism	262600
SLC26A4	Pendred syndrome	274600
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	264470
PEX1	Peroxisome biogenesis disorder 1A (Zellweger)	214100
PEX1	Peroxisome biogenesis disorder 1B (NALD/IRD)	601539
PEX6	Peroxisome biogenesis disorder 4A (Zellweger)	614862
PEX6	Peroxisome biogenesis disorder 4B	614863
PEX2	Peroxisome biogenesis disorder 5A (Zellweger)	614866
PEX2	Peroxisome biogenesis disorder 5B	614867
PEX10	Peroxisome biogenesis disorder 6A (Zellweger)	614870
PEX10	Peroxisome biogenesis disorder 6B	614871
PEX7	Peroxisome biogenesis disorder 9B	614879
HSD17B4	Perrault syndrome 1	233400
AMH	Persistent Mullerian duct syndrome, type I	261550
AMHR2	Persistent Mullerian duct syndrome, type II	261550
PAH	Phenylketonuria	261600
PHGDH	Phosphoglycerate dehydrogenase deficiency	601815
PKHD1	Polycystic kidney and hepatic disease	263200
GBE1	Polyglucosan body neuropathy, adult form	263570
SEPSECS	Pontocerebellar hypoplasia, type 2D	613811
MVK	Porokeratosis 3, dominant	175900
BCHE	Postanesthesia apnea	-
FMR1	Premature ovarian failure	311360
POLG	Progressive external ophthalmoplegia dom. 1	157640

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Gene	Disease	OMIM
POLG	Progressive external ophthalmoplegia rec. 1,	258450
PCCA	Propionic acidemia	606054
PCCB	Propionic acidemia	606054
HSD17B3	Pseudohermaphroditism, male, with gynecomastia	264300
SRD5A2	Pseudovaginal perineoscrotal hypospadias	264600
CTSK	Pycnodysostosis	265800
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	312170
PDHB	Pyruvate dehydrogenase E1-beta deficiency	614111
NR2E3	Retinitis pigmentosa 37	611131
PROM1	Retinitis pigmentosa 41	612095
DHDDS	Retinitis Pigmentosa 59	613861
CLRN1	Retinitis pigmentosa 61	614180
HGSNAT	Retinitis pigmentosa 73	616544
BBS2	Retinitis pigmentosa 74	616562
POMGNT1	Retinitis Pigmentosa 76	617123
RLBP1	Retinitis punctata albescens	136880
RS1	Retinoschisis	312700
PEX7	Rhizomelic chondrodysplasia punctata, type 1	215100
SLC17A5	Salla disease	604369
HEXB	Sandhoff disease, infantile, juvenile, and adult forms	268800
TH	Segawa syndrome	605407
CEP290	Senior-Loken syndrome 6	610189
BSND	Sensorineural deafness with renal dysfunction (Bartter syndrome type 4a)	602522
ADA	Severe combined immunodeficiency, due to adenosine deaminase deficiency / partial deficiency	102700
IL2RG	Severe combined immunodeficiency	300400
DCLRE1C	Severe combined immunodeficiency Athabaskan-type	602450
GNE	Sialuria	269921
HBB	Sickle cell anemia	603903
ALDH3A2	Sjogren-Larsson syndrome	270200
SLC45A2	Skin/hair/eye pigmentation, variation in, 5	227240
DHCR7	Smith-Lemli-Opitz syndrome	270400
SACS	Spastic ataxia, Charlevoix-Saguenay type	270550
AR	Spinal and bulbar muscular atrophy of Kennedy	313200
SMN1	Spinal muscular atrophy 1, 2, 3, 4	253300, 253550, 253400, 271150
ATP7A	Spinal muscular atrophy, distal	300489
TPP1	Spinocerebellar ataxia 7	609270
PROM1	Stargardt disease 4	603786
LIFR	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome	601559
HEXA	Tay-Sachs disease	272800
F5	Thrombophilia due to protein C resistance	188055

Gene	Disease	OMIM
F9	Thrombophilia due to factor IX defect	300807
F2	Thrombophilia	188050
SLC5A5	Thyroid dyshormonogenesis 1	274400
TPO	Thyroid dyshormonogenesis 2A	274500
TG	Thyroid dyshormonogenesis 3	274700
IYD	Thyroid dyshormonogenesis 4	274800
DUOXA2	Thyroid dyshormonogenesis 5	274900
DUOX2	Thyroid dyshormonogenesis 6	607200
EDA	Tooth agenesis, selective	313500
HADHA	Trifunctional protein deficiency	609015
FAH	Tyrosinemia, type I	276700
MYO7A	Usher syndrome, type IB	276900
USH1C	Usher syndrome, type IC	276904
PCDH15	Usher syndrome, type ID/F, digenic	601067
CDH23	Usher syndrome, type ID/F, digenic	601067
PCDH15	Usher syndrome, type IF	602083
USH2A	Usher syndrome, type IIa	276901
CLRN1	Usher syndrome, type IIIa	276902
CFTR	Vas deferens, congenital bilateral aplasia of	277180
VWF	Von Willebrand disease, type 1	193400
VWF	Von Willebrand disease, types 2A, 2B, 2M, 2N	613554
VWF	Von Willebrand disease, type 3	277480
WRN	Werner syndrome	277700
EVC2	Weyers acrofacial dysostosis	193530
EVC	Weyers acrofacial dysostosis	193530
ATP7B	Wilson disease	277900

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