NBSure™

Newborn Genetic Screening 358-Gene Panel

Detects pathogenic or likely pathogenic variants across 358 genes associated with 411 single gene disorders

Amino Acid Metabolism Disorders (41)

Disorders	Genes	Inheritance
Phenylketonuria	*PAH*	AR
Hyperphenylalaninemia, non-PKU mild	*PAH*	AR
Hyperphenylalaninemia, mild, non-BH4-deficient	*DNAJC12*	AR
Hyperphenylalaninemia, BH4-deficient, A	*PTS*	AR
Hyperphenylalaninemia, BH4-deficient, B	*GCH1*	AR
Hyperphenylalaninemia, BH4-deficient, C	*QDPR*	AR
Hyperphenylalaninemia, BH4-deficient, D	*PCBD1*	AR
Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	*SPR*	AD/AR
Tyrosinemia, type I	*FAH*	AR
Tyrosinemia, type III	*HPD*	AR
Tyrosinemia, type II	*TAT*	AR
Citrullinemia	*ASS1*	AR
Citrullinemia, type II, neonatal-onset	*SLC25A13*	AR
Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	*SLC25A15*	AR
Argininosuccinic aciduria	*ASL*	AR
Argininemia	*ARG1*	AR
Ornithine transcarbamylase deficiency	*OTC*	XL
Gyrate atrophy of choroid and retina with or without ornithinemia	*OAT*	AR
Maple syrup urine disease, type II	*DBT*	AR
Maple syrup urine disease, type Ib	*BCKDHB*	AR
Maple syrup urine disease, type Ia	*BCKDHA*	AR
Branched-chain keto acid dehydrogenase kinase deficiency	*BCKDK*	AR
Dihydrolipoamide dehydrogenase deficiency	*DLD*	AR
Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency	*MAT1A*	AD/AR
Methionine adenosyltransferase deficiency, autosomal recessive	*MAT1A*	AD/AR
Homocystinuria, B6-responsive and nonresponsive types	*CBS*	AR
Homocystinuria due to MTHFR deficiency	*MTHFR*	AR
Homocystinuria-megaloblastic anemia, cbl E type	*MTRR*	AR
Homocystinuria-megaloblastic anemia, cblG complementation type	*MTR*	AR
Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	*AHCY*	AR
Hypermethioninemia due to adenosine kinase deficiency	*ADK*	AR
Glycine N-methyltransferase deficiency	*GNMT*	AR
Hyperprolinemia, type II	*ALDH4A1*	AR
Hyperprolinemia, type I	*PRODH*	AR
Hypervalinemia or hyperleucine-isoleucinemia	*BCAT1*	AR
	BCAT2	AR
Histidinemia	*HAL*	AD/AR
Pyruvate carboxylase deficiency	*PC*	AR
Carbamoylphosphate synthetase I deficiency	*CPS1*	AR
N-acetylglutamate synthase deficiency	*NAGS*	AR
Glycine encephalopathy 1	*GLDC*	*AR*
Glycine encephalopathy 2	*AMT*	*AR*

Fatty Acid Metabolism Disorders (18)

Disorders	Genes	Inheritance
3-hydroxyacyl-CoA dehydrogenase deficiency	*HADH*	AR
Carnitine deficiency, systemic primary	*SLC22A5*	AR
CPT deficiency, hepatic, type IA	*CPT1A*	AR
CPT II deficiency, lethal neonatal	*CPT2*	AR
CPT II deficiency, infantile	*CPT2*	AR
CPT II deficiency, myopathic, stress-induced	*CPT2*	AD/AR
Carnitine-acylcarnitine translocase deficiency	*SLC25A20*	AR
Acyl-CoA dehydrogenase, medium chain, deficiency of	*ACADM*	AR
VLCAD deficiency	*ACADVL*	AR
LCHAD deficiency	*HADHA*	AR
Acyl-CoA dehydrogenase, short-chain, deficiency of	*ACADS*	AR
Glutaric acidemia IIB	*ETFB*	AR
Glutaric acidemia IIC	*ETFDH*	AR
Glutaric acidemia IIA	*ETFA*	AR
Mitochondrial trifunctional protein deficiency 2	*HADHB*	AR
2,4-dienoyl-CoA reductase deficiency	*NADK2*	AR

Paediatric Tumours (2)

Disorders	Genes	Inheritance
Wilms tumor, type 1	*WT1*	AD/SMu
Retinoblastoma	*RB1*	AD/SMu

Organic Acid Metabolism Disorders (39)

Disorders	Genes	Inheritance
Methylmalonic aciduria, mut(0) type	*MMUT*	AR
Methylmalonic aciduria and homocystinuria, cblD type	*MMADHC*	AR
Methylmalonic aciduria and homocystinuria, cblC type, digenic	*PRDX1*	AR
Methylmalonic aciduria and homocystinuria, cblC type	*MMACHC*	AR
Methylmalonic aciduria, vitamin B12-responsive, cblA type	*MMAA*	AR
Methylmalonic aciduria, vitamin B12-responsive, cblB type	*MMAB*	AR
Methylmalonyl-CoA epimerase deficiency	*MCEE*	AR
Combined malonic and methylmalonic aciduria	*ACSF3*	AR
Methylmalonic aciduria and homocystinuria, cblF type	*LMBRD1*	AR
Methylmalonate semialdehyde dehydrogenase deficiency	*ALDH6A1*	AR
Methylmalonic aciduria, transient, due to transcobalamin receptor defect	*CD320*	AR
Methylmalonic aciduria and homocystinuria, cblJ type	*ABCD4*	AR
Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	*SUCLA2*	AR
Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)	*SUCLG1*	AR
Transcobalamin II deficiency	*TCN2*	AR
Methylmalonic aciduria and homocysteinemia, cblX type	*HCFC1*	XLR
Propionicacidemia	*PCCA*	AR
	PCCB	
Isovaleric acidemia	*IVD*	AR
Glutaric aciduria I	*GCDH*	AR
Glutaric aciduria III	*SUGCT*	AR
Holocarboxylase synthetase deficiency	*HLCS*	AR
Biotinidase deficiency	*BTD*	AR
3-Methylcrotonyl-CoA carboxylase 1 deficiency	*MCCC1*	AR
3-Methylcrotonyl-CoA carboxylase 2 deficiency	*MCCC2*	AR
HMG-CoA synthase-2 deficiency	*HMGCS2*	AR
Alpha-methylacetoacetic aciduria	*ACAT1*	AR
HMG-CoA lyase deficiency	*HMGCL*	AR
3-methylglutaconic aciduria, type I	*AUH*	AR
Ethylmalonic encephalopathy	*ETHE1*	AR
Malonyl-CoA decarboxylase deficiency	*MLYCD*	AR
Isobutyryl-CoA dehydrogenase deficiency	*ACAD8*	AR
HSD10 mitochondrial disease	*HSD17B10*	XLD
Barth syndrome	*TAFAZZIN*	XLR
3-methylglutaconic aciduria, type VIIA, autosomal dominant	*CLPB*	AD
3-methylglutaconic aciduria, type VIIB, autosomal recessive	*CLPB*	AR
3-methylglutaconic aciduria, type III	*OPA3*	AR
3-methylglutaconic aciduria, type V	*DNAJC19*	AR
3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	*SERAC1*	AR
2-methylbutyrylglycinuria	*ACADSB*	AR

Lysosomal Storage Diseases (29)

Disorders	Genes	Inheritance
Fabry disease	*GLA*	XL
Fabry disease, cardiac variant	*GLA*	XL
Gaucher disease, type I	*GBA1*	AR
Gaucher disease, type II	*GBA1*	AR
Gaucher disease, type III	*GBA1*	AR
Gaucher disease, perinatal lethal	*GBA1*	AR
Gaucher disease, type IIIC	*GBA1*	AR
Gaucher disease, atypical	*PSAP*	.
Glycogen storage disease II	*GAA*	AR
Mucopolysaccharidosis Is	*IDUA*	AR
Mucopolysaccharidosis Ih/s	*IDUA*	AR
Mucopolysaccharidosis Ih	*IDUA*	AR
Mucopolysaccharidosis II	*IDS*	XLR
Mucopolysaccharidosis type IIIA (Sanfilippo A)	*SGSH*	AR
Mucopolysaccharidosis type IIIB (Sanfilippo B)	*NAGLU*	AR
Mucopolysaccharidosis type IIIC (Sanfilippo C)	*HGSNAT*	AR
Mucopolysaccharidosis type IIID	*GNS*	AR
Mucopolysaccharidosis type IVA	*GALNS*	AR
Mucopolysaccharidosis type IVB (Morquio)	*GLB1*	AR
Mucopolysaccharidosis type VI (Maroteaux-Lamy)	*ARSB*	AR
Mucopolysaccharidosis type VII	*GUSB*	AR
Niemann-Pick disease, type A	*SMPD1*	AR
Niemann-Pick disease, type B	*SMPD1*	AR
Niemann-Pick disease, type C1	*NPC1*	AR
Niemann-Pick disease, type D	*NPC1*	AR
Niemann-pick disease, type C2	*NPC2*	AR
Cholesteryl ester storage disease	*LIPA*	AR
Wolman disease	*LIPA*	AR
Krabbe disease	*GALC*	AR

Other Endocrine and Metabolic Disorders (94)

Disorders	Genes	Inheritance
Galactosemia	*GALT*	AR
Galactokinase deficiency with cataracts	*GALK1*	AR
Galactose epimerase deficiency	*GALE*	AR
Galactosemia IV	*GALM*	AR
Fructose intolerance, hereditary	*ALDOB*	AR
Glycogen storage disease type Ia	*G6PC1*	AR
Glycogen storage disease type Ib	*SLC37A4*	AR
Glycogen storage disease type Ic	*SLC37A4*	AR
Glycogen storage disease IIIa	*AGL*	AR
Glycogen storage disease IIIb	*AGL*	AR
Glycogen storage disease IV	*GBE1*	AR
Glycogen storage disease VI	*PYGL*	AR
Glycogen storage disease VII	*PFKM*	AR
Glycogen storage disease, type IXa1	*PHKA2*	XLR
Phosphorylase kinase deficiency of liver and muscle, autosomal recessive	*PHKB*	AR
Glycogen storage disease Ixc	*PHKG2*	AR
Muscle glycogenosis	*PHKA1*	XLR
Fanconi-Bickel syndrome	*SLC2A2*	AR
Glycogen storage disease XII	*ALDOA*	AR
Glycogen storage disease XV	*GYG1*	AR
Glycogen storage disease 0, liver	*GYS2*	AR
Danon disease	*LAMP2*	XLD
Wilson disease	*ATP7B*	AR
Cholestasis, progressive familial intrahepatic 2	*ABCB11*	AR
Cholestasis, progressive familial intrahepatic 1	*ATP8B1*	AR
Crigler-Najjar syndrome, type I	*UGT1A1*	AR
Crigler-Najjar syndrome, type II	*UGT1A1*	AR
Anemia, congenital, nonspherocytic hemolytic, 1, G6PD deficient	*G6PD*	XL
Bile acid synthesis defect, congenital, 1	*HSD3B7*	AR
5-fluorouracil toxicity	*DPYD*	AR
Dihydropyrimidine dehydrogenase deficiency	*DPYD*	AR
Thiopurines, poor metabolism of, 1	*TPMT*	AR
Thiopurines, poor metabolism of, 2	*NUDT15*	AD
Neurofibromatosis-Noonan syndrome	*NF1*	AD
Noonan syndrome 1	*PTPN11*	AD
Noonan syndrome 2	*LZTR1*	AR
Noonan syndrome 3	*KRAS*	AD
Noonan syndrome 4	*SOS1*	AD
Noonan syndrome 5	*RAF1*	AD
Noonan syndrome 6	*NRAS*	AD
Noonan syndrome 7	*BRAF*	AD
Noonan syndrome 8	*RIT1*	AD
Noonan syndrome 9	*SOS2*	AD
Noonan syndrome 10	*LZTR1*	AD
Noonan syndrome 12	*RRAS2*	AD
Noonan syndrome-like disorder with loose anagen hair 2	*PPP1CB*	AD
Noonan syndrome-like disorder with loose anagen hair 1	*SHOC2*	AD
Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia	*CBL*	AD
Congenital myopathy with excess of muscle spindles	*HRAS*	AD
Costello syndrome	*HRAS*	AD
Cardiofaciocutaneous syndrome 3	*MAP2K1*	AD
Cardiofaciocutaneous syndrome 4	*MAP2K2*	AD
Congenital disorder of glycosylation, type Ia	*PMM2*	AR
Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	*CYP21A2*	AR
Lipoid adrenal hyperplasia	*STAR*	AR
Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	*CYP11B1*	AR
Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	*CYP11A1*	.
Thyroid dyshormonogenesis 1	*SLC5A5*	AR
Thyroid dyshormonogenesis 2A	*TPO*	AR
Thyroid dyshormonogenesis 3	*TG*	AR
Thyroid dyshormonogenesis 4	*IYD*	AR
Thyroid dyshormonogenesis 5	*DUOXA2*	AR
Thyroid dyshormonogenesis 6	*DUOX2*	AR
Hypothyroidism, congenital, nongoitrous, 1	*TSHR*	AR
Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia	*PAX8*	AD
Hypothyroidism, congenital, nongoitrous, 4	*TSHB*	AR
Hypothyroidism, congenital, nongoitrous, 5	*NKX2-5*	AD
Hypothyroidism, congenital, nongoitrous, 6	*THRA*	AD
Hypothyroidism, congenital, nongoitrous, 7	*TRHR*	AR
Hypothyroidism, congenital, nongoitrous, 9	*IRS4*	XLR
Thyroid hormone resistance	*THRB*	AD
Hyperparathyroidism, transient neonatal	*TRPV6*	AR
Thyroid hormone metabolism, abnormal, 1	*SECISBP2*	AR
Hypothyroidism, central, and testicular enlargement	*IGSF1*	XLR
Pendred syndrome	*SLC26A4*	AR
Diabetes mellitus, neonatal, with congenital hypothyroidism	*GLIS3*	AR
Choreoathetosis, hypothyroidism, and neonatal respiratory distress	*NKX2-1*	AD
Bamforth-Lazarus syndrome	*FOXE1*	AR
Hyperinsulinemic hypoglycemia, familial, 1	*ABCC8*	AD/AR
Hypoglycemia of infancy, leucine-sensitive	*ABCC8*	AD

Disorders	Genes	Inheritance
Hyperinsulinemic hypoglycemia, familial, 2	*KCNJ11*	AD/AR
Hyperinsulinemic hypoglycemia, familial, 3	*GCK*	AD
Hyperinsulinemic hypoglycemia, familial, 4	*HADH*	AR
Hyperinsulinemic hypoglycemia, familial, 5	*INSR*	AD
Hyperinsulinism-hyperammonemia syndrome	*GLUD1*	AD
Hyperinsulinemic hypoglycemia, familial, 7	*SLC16A1*	AD
Hyperproinsulinemia	*INS*	AD
Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young	*HNF4A*	AD
MODY, type I	*HNF4A*	AD
Laron dwarfism	*GHR*	AR
Growth hormone deficiency, isolated, type IV	*GHRHR*	AR
Growth hormone deficiency, isolated, type IA	*GH1*	AR
Growth hormone deficiency, isolated, type IB	*GH1*	-
Growth hormone deficiency, isolated, type II	*GH1*	AD

Skeletal Disorders (18)

Disorders	Genes	Inheritance
Hypophosphatemic rickets, X-linked dominant	*PHEX*	XLD
Hypophosphatemic rickets with hypercalciuria	*SLC34A3*	AR
Hypophosphatemic rickets, AR	*DMP1*	AR
Hypophosphatemic rickets	*CLCN5*	XLR
Hypophosphatemic rickets, autosomal recessive, 2	*ENPP1*	AR
Vitamin D-dependent rickets, type I	*CYP27B1*	AR
Rickets, vitamin D-resistant, type IIA	*VDR*	AR
Osteogenesis imperfecta, type I	*COL1A1*	AD
Osteogenesis imperfecta, type II	*COL1A1*	AD
	COL1A2	
Osteogenesis imperfecta, type III	*COL1A1*	AD
	COL1A2	
Osteogenesis imperfecta, type IV	*COL1A1*	AD
	COL1A2	
Osteogenesis imperfecta, type VII	*CRTAP*	AR
Osteogenesis imperfecta, type VIII	*P3H1*	AR
Thanatophoric dysplasia, type I	*FGFR3*	AD
Thanatophoric dysplasia, type II	*FGFR3*	AD
Hypophosphatasia, infantile	*ALPL*	AR
Hypophosphatasia, childhood	*ALPL*	AR
Cartilage-hair hypoplasia	*RMRP*	AR

Hematologic Disorders (26)

Disorders	Genes	Inheritance
Thalassemia, beta	*HBB*	.
Thrombocytopenia with beta-thalassemia, X-linked	*GATA1*	XLR
Diamond-Blackfan anemia 1	*RPS19*	AD
Diamond-Blackfan anemia 3	*RPS24*	AD
Diamond-Blackfan anemia 5	*RPL35A*	AD
Diamond-Blackfan anemia 6	*RPL5*	AD
Diamond-Blackfan anemia 7	*RPL11*	AD
Diamond-Blackfan anemia 8	*RPS7*	AD
Diamond-Blackfan anemia 9	*RPS10*	AD
Shwachman-Diamond syndrome 1	*SBDS*	AR
Fanconi anemia, complementation group A	*FANCA*	AR
Fanconi anemia, complementation group B	*FANCB*	XLR
Fanconi anemia, complementation group C	*FANCC*	AR
Fanconi anemia, complementation group D1	*BRCA2*	AR
Fanconi anemia, complementation group D2	*FANCD2*	AR
Fanconi anemia, complementation group E	*FANCE*	AR
Fanconi anemia, complementation group F	*FANCF*	AR
Fanconi anemia, complementation group I	*FANCI*	AR
Fanconi anemia, complementation group Q	*ERCC4*	AR
Hemophagocytic lymphohistiocytosis, familial, 3	*UNC13D*	AR
Hemophagocytic lymphohistiocytosis, familial, 4	*STX11*	AR
Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease	*STXBP2*	AR
Hemophilia A	*F8*	XLR
Hemophilia B	*F9*	XLR
Paroxysmal nocturnal hemoglobinuria, somatic	*PIGA*	.
Paroxysmal nocturnal hemoglobinuria 2	*PIGT*	AD/SMu

Digestive Diseases (4)

Disorders	Gene	Inheritance
Inflammatory bowel disease 28, early onset, autosomal recessive	*IL10RA*	AR
Inflammatory bowel disease 25, early onset, autosomal recessive	*IL10RB*	AR
Gastrointestinal defects and immunodeficiency syndrome	*TTC7A*	AR
Inflammatory skin and bowel disease, neonatal, 1	*ADAM17*	AR

Immune Diseases (32)

Disorders	Genes	Inheritance
Neutropenia, severe congenital 1, autosomal dominant	*ELANE*	AD
Neutropenia, severe congenital 3, autosomal recessive	*HAX1*	AR
Neutropenia, severe congenital 4, autosomal recessive	*G6PC3*	AR
Neutropenia, severe congenital, X-linked	*WAS*	XLR
Chediak-Higashi syndrome	*LYST*	AR
Severe combined immunodeficiency, Athabascan type	*DCLRE1C*	AR
Severe combined immunodeficiency, X-linked	*IL2RG*	XLR
Immunodeficiency 124, severe combined	*NHEJ1*	AR
Severe combined immunodeficiency due to ADA deficiency	*ADA*	AR/SMo
Severe combined immunodeficiency, B cell-negative	*RAG1*	AR
Combined cellular and humoral immune defects with granulomas	*RAG1*	AR
	RAG2	
Severe combined immunodeficiency, autosomal recessive, T-negative/B-positive type	*JAK3*	AR
Autoimmune disease, multisystem, infantile-onset, 2	*ZAP70*	AR
Immunodeficiency 48	*ZAP70*	AR
Hyper-IgE syndrome 1, autosomal dominant, with recurrent infections	*STAT3*	AD
T-cell immunodeficiency, congenital alopecia, and nail dystrophy	*FOXN1*	AR
T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant	*FOXN1*	AD
Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	*FOXP3*	XLR
Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity	*RAG1*	.
Immunodeficiency 104, severe combined	*IL7R*	AR
Immunodeficiency 49, severe combined	*BCL11B*	AD
Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities	*BCL11B*	AD
Wiskott-Aldrich syndrome	*WAS*	XLR
Lymphoproliferative syndrome, X-linked, 1	*SH2D1A*	XLR
Lymphoproliferative syndrome, X-linked, 2	*XIAP*	XLR
Chronic granulomatous disease, X-linked	*CYBB*	XLR
Chronic granulomatous disease 1, autosomal recessive	*NCF1*	AR
Chronic granulomatous disease 2, autosomal recessive	*NCF2*	AR
Chronic granulomatous disease 3, autosomal recessive	*NCF4*	AR
Chronic granulomatous disease 4, autosomal recessive	*CYBA*	AR
Agammaglobulinemia, X-linked 1	*BTK*	XLR
Immunodeficiency, X-linked, with hyper-IgM	*CD40LG*	XLR

Cardiovascular Disease (1)

Disorders	Genes	Inheritance
Pulmonary hypertension, familial primary, 1, with or without HHT	*BMPR2*	AD

Skin Disorders (26)

Disorders	Genes	Inheritance
Epidermolysis bullosa dystrophica inversa	*COL7A1*	AR
Epidermolysis bullosa dystrophica, autosomal recessive	*COL7A1*	AR
Epidermolysis bullosa, pretibial	*COL7A1*	AD/AR
Epidermolysis bullosa pruriginosa	*COL7A1*	AD/AR
Epidermolysis bullosa simplex 2F, with mottled pigmentation	*KRT5*	AD
Epidermolysis bullosa, junctional 1A, intermediate	*LAMB3*	AR
Epidermolysis bullosa, junctional 1B, severe	*LAMB3*	AR
Epidermolysis bullosa, junctional 5B, with pyloric atresia	*ITGB4*	AR
Epidermolysis bullosa simplex 1A, generalized severe	*KRT14*	AD
Epidermolysis bullosa, junctional 2B, severe	*LAMA3*	AR
Epidermolysis bullosa, junctional 2A, intermediate	*LAMA3*	AR
Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous	*LAMA3*	AR
Epidermolysis bullosa, junctional 3B, severe	*LAMC2*	AR
Epidermolysis bullosa, junctional 3A, intermediate	*LAMC2*	AR
Epidermolysis bullosa, lethal acantholytic	*DSP*	AR
Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome	*ITGA3*	AR
Epidermolysis bullosa simplex 5A, Ogna type	*PLEC*	AD
Epidermolysis bullosa simplex 5B, with muscular dystrophy	*PLEC*	AR
Epidermolysis bullosa simplex 5C, with pyloric atresia	*PLEC*	AR
Dyskeratosis congenita, autosomal dominant 1	*TERC*	AD
Dyskeratosis congenita, autosomal dominant 2	*TERT*	AD/AR
Dyskeratosis congenita, autosomal dominant 3	*TINF2*	AD
Dyskeratosis congenita, autosomal recessive 2	*NHP2*	AR
Dyskeratosis congenita, autosomal recessive 3	*WRAP53*	AR
Albinism, oculocutaneous, type IA	*TYR*	AR
Albinism, oculocutaneous, type IB	*TYR*	AR

Neuromuscular Disorders (32)

Disorders	Genes	Inheritance
Adrenoleukodystrophy	*ABCD1*	XLR
Muscular dystrophy, limb-girdle, autosomal recessive 1	*CAPN3*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 2	*DYSF*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 3	*SGCA*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 4	*SGCB*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 5	*SGCG*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 6	*SGCD*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 7	*TCAP*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 8	*TRIM32*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 10	*TTN*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 12	*ANO5*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 17	*PLEC*	AR
Muscular dystrophy, limb-girdle, autosomal recessive 23	*LAMA2*	AR
Duchenne muscular dystrophy	*DMD*	XLR
Becker muscular dystrophy	*DMD*	XLR
Emery-Dreifuss muscular dystrophy 2, autosomal dominant	*LMNA*	AD
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1	*POMT1*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2	*POMT2*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3	*POMGNT1*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	*FKTN*	AR
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5	*FKRP*	AR
Ullrich congenital muscular dystrophy 1A	*COL6A1*	AD/AR
Ullrich congenital muscular dystrophy 1B	*COL6A2*	AD/AR
Ullrich congenital muscular dystrophy 1C	*COL6A3*	AD/AR
Spinal muscular atrophy-1	*SMN1*	AR
Spinal muscular atrophy-2	*SMN1*	AR
Spinal muscular atrophy-3	*SMN1*	AR
Spinal muscular atrophy-4	*SMN1*	AR
Spinal muscular atrophy, X-linked 2, infantile	*UBA1*	XLR
Neuronopathy, distal hereditary motor, X-linked	*ATP7A*	XLR
Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant	*BICD2*	AD
Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant	*BICD2*	AD

Ophthalmologic and Otorhinolaryngologic Diseases (43)

Disorders	Genes	Inheritance
Deafness, autosomal recessive 1A	*GJB2*	AR/DD
Deafness, digenic, GJB2/GJB3	*GJB3*	AR/DD
Deafness, digenic GJB2/GJB6	*GJB6*	AR/DD
Deafness, autosomal recessive 2	*MYO7A*	AR
Deafness, autosomal dominant 11	*MYO7A*	AD
Deafness, autosomal recessive 3	*MYO15A*	AR
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	*SLC26A4*	AR
Deafness, autosomal recessive 6	*TMIE*	AR
Deafness, autosomal recessive 7	*TMC1*	AR
Deafness, autosomal recessive 8/10	*TMPRSS3*	AR
Deafness, autosomal recessive 9	*OTOF*	AR
Deafness, autosomal recessive 12	*CDH23*	AR
Deafness, autosomal recessive 15	*GIPC3*	AR
Deafness, autosomal recessive 16	*STRC*	AR
Deafness, autosomal recessive 18B	*OTOG*	AR
Deafness, autosomal recessive 21	*TECTA*	AR
Deafness, autosomal recessive 24	*RDX*	AR
Deafness, autosomal dominant 20/26	*ACTG1*	AD
Deafness, autosomal recessive 29	*CLDN14*	AR
Deafness, autosomal dominant 9	*COCH*	AD
Deafness, autosomal recessive 36	*ESPN*	AR
Deafness, neurosensory, without vestibular involvement, autosomal dominant	*ESPN*	AR
Deafness, autosomal recessive 35	*ESRRB*	AR
Deafness, autosomal dominant 10	*EYA4*	AD
Deafness, autosomal recessive 25	*GRXCR1*	AR
Deafness, autosomal recessive 42	*ILDR1*	AR
Deafness, autosomal dominant 2A	*KCNQ4*	AD
Deafness, autosomal recessive 67	*LHFPL5*	AR
Deafness, autosomal recessive 77	*LOXHD1*	AR
Deafness, autosomal recessive 63	*LRTOMT*	AR
Deafness, autosomal recessive 49	*MARVELD 2*	AR
Deafness, autosomal recessive 30	*MYO3A*	AR
Deafness, autosomal dominant 22	*MYO6*	AD
Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy	*MYO6*	AD

Disorders	Genes	Inheritance
Deafness, autosomal recessive 22	*OTOA*	AR
Deafness, autosomal recessive 84B	*OTOGL*	AR
Deafness, X-linked 2	*POU3F4*	XLR
Deafness, autosomal dominant 15/52	*POU4F3*	AD
Deafness, autosomal recessive 28	*TRIOBP*	AR
Deafness, X-linked 4	*SMPX*	XLD
Deafness, autosomal recessive 79	*TPRN*	AR
Deafness, aminoglycoside-induced	*MT-RNR1*	.
Usher syndrome, type 2A	*USH2A*	AR

Mitochondrial Disorders (3)

Disorders	Genes	Inheritance
MELAS syndrome	*MT-TL1*	.
Leigh syndrome	*MT-ND5*	.
Multiple mitochondrial dysfunctions syndrome 7	*GCSH*	AR

Urologic Diseases (2)

Disorders	Genes	Inheritance
Hemolytic uremic syndrome, atypical, susceptibility to, 3	*CFI*	AD
Hemolytic uremic syndrome, atypical, susceptibility to, 5	*C3*	AD

Respiratory Disease (1)

Disorders	Genes	Inheritance
Cystic fibrosis	*CFTR*	AR

Note: AD: Autosomal dominant; AR: Autosomal recessive; XL: X-linked; XLD: X-linked dominant; XLR: X-linked recessive; DD: Digenic dominant; SMo: Somatic mosaicism; SMu: Somatic mutation

(Global) www.xcelomglobal.com | marketing@xcelom.com | Xcelom Limited is a subsidiary of Berry Genomics responsible for international business.