NBSure™

Newborn Genetic Screening 358-Gene Panel

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

Report Sections

Sections	Description
Main Report	Autosomal Dominant and X-linked Disorders P and LP variant Autosomal Recessive Disorders P/ LP + P/ LP and P/ LP + VUS* variant
Appendix	Additional Findings in Target Genes Findings partially related to the patient's clinical phenotype, but with insufficient genetic pathogenic evidence. Those require further clinical attention. (Applicable only to samples with provided neonatal phenotypes or biochemical/ tandem mass spectrometry abnormalities) Aneuploidies and Microdeletion/ Microduplication Syndromes • 37 microdeletion/ microduplication syndromes in 32 regions • 4 specific chromosomal aneuploidies: chr13，chr18，chr21，45XO
Other	The candidate variant table (targeted site detection information table) is available under special request

P: Pathogenic; LP: Likely Pathogenic; VUS: Variant of Uncertain Significance
X-Linked phenotype reported include X-linked, X-linked dominant and X-linked recessive
***** Only report the VUS with scoring > 4

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*

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

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

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

Disorders	Genes	Inheritance
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
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

Mitochondrial Disorders (3)

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

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

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

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
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

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.