VERAgene®
comprehensive NIPT

# MONOGENIC DISEASES TESTED BY **VERAgene**

| DISEASE | GENE | CLASSIFICATION | SEVERITY |
|---|---|---|---|
| 3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency | *HMGCL* | MET | Severe |
| 3-Methylcrotonyl-CoA Carboxylase Deficiency 1 | *MCCC1* | MET | Severe |
| 3-Methylcrotonyl-CoA Carboxylase Deficiency 2 | *MCCC2* | MET | Severe |
| Abetalipoproteinemia | *MTTP* | DIG, NEUR, OPTH, HEM | Severe |
| Acyl-CoA Oxidase I Deficiency | *ACOX1* | NEUR | Very severe |
| Aicardi-Goutières Syndrome | *SAMHD1* | NEUR | Severe |
| Alport Syndrome, X-Linked | *COL4A5* | REN, OPTH, HEAR | Severe |
| Alstrom Syndrome | *ALMS1* | OPTH, HEAR, REN, CARD | Severe |
| Andermann Syndrome | *SLC12A6* | MUSC, NEUR | Severe |
| Aromatase Deficiency | *CYP19A1* | SD | Moderate |
| Arthrogryposis Mental Retardation Seizures | *SLC35A3* | MET | Severe |
| Asparagine Synthetase Deficiency | *ASNS* | NEUR | Very severe |
| Aspartylglycosaminuria | *AGA* | MET, NEUR | Severe |
| Autosomal Recessive Polycystic Kidney Disease | *PKHD1* | REN | Severe |
| Bardet-Biedl Syndrome (BBS1-related) | *BBS1* | OPTH, MET, END | Severe |
| Bardet Biedl Syndrome (BBS12-related) | *BBS12* | OPTH | Severe |
| Beta Thalassemia | *HBB* | HEM | Very severe |
| Biotinidase Deficiency | *BTD* | MET | Severe |
| Canavan Disease | *ASPA* | NEUR | Severe |
| Carpenter Syndrome | *RAB23* | SKEL | Severe |
| Choreacanthocytosis | *VPS13A* | NEUR | Moderate |
| Choroideremia, X-Linked | *CHM* | OPTH | Severe |
| Citrin Deficiency | *SLC25A13* | MET | Moderate |
| Combined Oxidative Phosphorylation Deficiency 3 | *TSFM* | NEUR, MET, CARD | Very severe |
| Congenital Disorder of Glycosylation, Type 1A (PMM2-related) | *PMM2* | MET | Severe |
| Congenital Neutropenia (HAX1-related) | *HAX1* | IMM | Severe |
| Crigler Najjar Syndrome, Type I | *UGT1A1* | MET | Very severe |
| Cystic Fibrosis * | *CFTR* | RESP, DIG | Very severe |
| Factor XI Deficiency | *F11* | HEM | Severe |
| Familial Dysautonomia | *IKBKAP* | NEUR | Moderate |
| Fanconi Anemia, Type C | *FANCC* | IMM | Severe |
| Fanconi Anemia, Type G | *FANCG* | HEM | Severe |
| Gaucher Disease | *GBA* | NEUR, HEP, CARD | Severe |
| Glutaric Acidemia, Type 2A | *ETFA* | MET | Moderate |
| Glycine Encephalopathy (GLDC-related) | *GLDC* | MET | Very severe |
| Glycogen Storage Disease, Type 1A | *G6PC* | MET | Moderate |
| Glycogen Storage Disease, Type 1B | *SLC37A4* | MET | Moderate |
| Glycogen Storage Disease, Type 3 | *AGL* | MET | Severe |
| Glycogen Storage Disease, Type 7 | *PFKM* | MET | Severe |
| GRACILE Syndrome | *BCS1L* | MET | Very severe |
| Hereditary Fructose Intolerance | *ALDOB* | MET | Moderate |
| Homocystinuria, Type cblE | *MTRR* | MET | Severe |
| Hydrolethalus Syndrome | *HYLS1* | NEUR, CARD | Very severe |
| Inclusion Body Myopathy, Type 2 | *GNE* | MUSC | Moderate |
| Isovaleric Acidemia | *IVD* | MET | Severe |
| Joubert Syndrome, Type 2 | *TMEM216* | NEUR | Severe |
| Junctional Epidermolysis Bullosa, Herlitz Type | *LAMC2* | SKIN | Severe |
| Lamellar Ichthyosis, Type 1 | *TGM1* | MET | Moderate |
| Leber Congenital Amaurosis (LCA5-related) | *LCA5* | OPTH | Severe |
| Leigh Syndrome, French-Canadian Type | *LRPPRC* | NEUR, MUSC | Severe |
| Leukoencephalopathy with Vanishing White Matter | *EIF2B5* | NEUR | Severe |

* VERAgene tests for mutations that cause the classic Cystic Fibrosis phenotype.

| DISEASE | GENE | CLASSIFICATION | SEVERITY |
|---|---|---|---|
| Leydig Cell Hypoplasia [Luteinizing Hormone Resistance] | *LHCGR* | SD | Moderate |
| Limb Girdle Muscular Dystrophy, Type 2E | *SGCB* | MUSC | Severe |
| Lipoamide Dehydrogenase Deficiency [Maple Syrup Urine Disease, Type 3] | *DLD* | MET | Severe |
| Lipoprotein Lipase Deficiency | *LPL* | MET | Moderate |
| Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency | *HADHA* | MET | Severe |
| Lysinuric Protein Intolerance | *SLC7A7* | MET | Severe |
| Maple Syrup Urine Disease, Type 1B | *BCKDHB* | MET | Severe |
| Methylmalonic Acidemia (MMAA-related) | *MMAA* | MET | Very severe |
| Methylmalonic Aciduria, Type mut(0) | *MMUT* | MET | Severe |
| Methylmalonic Aciduria and Homocystinuria, Type cblC | *MMACHC* | MET | Severe |
| Methylmalonic Aciduria and Homocystinuria, Type cblD | *MMADHC* | MET | Severe |
| Mucopolysaccharidosis, Type II [Hunter Syndrome], X-Linked | *IDS* | RESP, CARD | Very severe |
| Mucopolysaccharidosis, Type IIIC [Sanfilippo C] | *HGSNAT* | MET, NEUR, OPTH | Severe |
| Multiple Sulfatase Deficiency | *SUMF1* | MET | Very severe |
| Myotubular Myopathy, X-Linked | *MTM1* | MUSC | Severe |
| Navajo Neurohepatopathy [MPV17-related Hepatocerebral Mitochondrial DNA Depletion Syndrome] | *MPV17* | NEUR | Severe |
| Neuronal Ceroid Lipofuscinosis (CLN8-related) | *CLN8* | NEUR | Very severe |
| Neuronal Ceroid Lipofuscinosis (MFSD8-related) | *MFSD8* | NEUR | Very severe |
| Neuronal Ceroid Lipofuscinosis (TPP1-related) | *TPP1* | NEUR | Very severe |
| Nijmegen Breakage Syndrome | *NBN* | NEUR | Severe |
| Omenn Syndrome (RAG2-related) | *RAG2* | IMM | Very severe |
| Ornithine Aminotransferase Deficiency | *OAT* | OPTH | Moderate |
| Ornithine Translocase Deficiency [Hyperornithinemia-Hyperammonemia -Homocitrullinuria (HHH) Syndrome] | *SLC25A15* | MET | Severe |
| Pendred Syndrome | *SLC26A4* | HEAR, END | Moderate |
| Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX1-related) | *PEX1* | MET | Severe |
| Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX2-related) | *PEX2* | MET | Severe |
| Phenylketonurea | *PAH* | MET | Very severe |
| Pontocerebellar Hypoplasia, Type 1A | *VRK1* | NEUR, MUSC | Very severe |
| Pontocerebellar Hypoplasia, Type 2D | *SEPSECS* | NEUR | Very severe |
| Pontocerebellar Hypoplasia, Type 2E | *VPS53* | NEUR | Very severe |
| Primary Ciliary Dyskinesia (DNAH5-related) | *DNAH5* | RESP, INF | Moderate |
| Primary Ciliary Dyskinesia (DNAI1-related) | *DNAI1* | RESP, INF | Moderate |
| Primary Hyperoxaluria, Type 3 | *HOGA1* | REN, MET | Moderate |
| Pycnodysostosis | *CTSK* | MET | Severe |
| Pyruvate Dehydrogenase Deficiency (PDHB-Related) | *PDHB* | NEUR, MET | Severe |
| Retinal Dystrophy (RLBP1-related) [Bothnia Retinal Dystrophy] | *RLBP1* | OPTH | Severe |
| Retinitis Pigmentosa 25 (EYS-related) | *EYS* | OPTH | Severe |
| Retinitis Pigmentosa 59 (DHDDS-related) | *DHDDS* | OPTH | Severe |
| Sanfilippo Syndrome, Type D [Mucopolysaccharidosis IIID] | *GNS* | MET | Severe |
| Severe Combined Immunodeficiency, Type Athabaskan | *DCLRE1C* | IMM | Very severe |
| Severe Combined Immunodeficiency, X-Linked | *IL2RG* | IMM | Very severe |
| Sickle-Cell Disease | *HBB* | HEM | Very severe |
| Sjögren-Larsson Syndrome | *ALDH3A2* | MET | Severe |
| Steroid-Resistant Nephrotic Syndrome | *NPHS2* | REN | Severe |
| Stuve-Wiedemann Syndrome | *LIFR* | SKEL | Severe |
| Tay-Sachs Disease | *HEXA* | MET | Very severe |
| Usher Syndrome, Type 1F | *PCDH15* | HEAR | Moderate |
| Usher Syndrome, Type 3 | *CLRN1* | HEAR, OPTH | Moderate |
| Wolman Disease | *LIPA* | MET, HEP | Severe |

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|---|---|---|---|---|---|---|---|---|---|
| CARD | CARDIAC | DIG | DIGESTIVE | END | ENDOCRINE | HEAR | HEARING | HEM | HEMATOLOGICAL |
| HEP | HEPATIC | IMM | IMMUNOLOGICAL | INF | INFERTILITY | MET | METABOLIC | MUSC | MUSCULAR |
| NEUR | NEUROLOGICAL | OPTH | OPTHALMOLOGICAL | REN | RENAL | RESP | RESPIRATORY | SD | SEXUAL DEVELOPMENT |
| SKEL | SKELETAL | SKIN | SKIN | | | | | | |

A disease may be classified into several types. The classification listed is based on the most common symptoms associated with each condition. Degree of severity of a condition can vary and depends on the specific mutation, signs and symptoms.

*Results and possible next steps should always be considered in the context of other clinical criteria and should be fully discussed with your healthcare provider. Genetic counseling is recommended when a high risk result is received.*

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