NIPD GENETICS

VERAgene®

comprehensive NIPT

MONOGENIC DISEASES TESTED BY **VERAgene**

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

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

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

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.

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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