NIPD GENETICS

VERAgene® comprehensive NIPT

Листа на 100 моногенетски болести

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

* VERAgene врши тестирање за мутации на гени кои предизвикуваат класичен фенотип на Цистична фиброза.

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

CARD	КАРДИОЛОШКИ	DIG	ДИГЕСТИВНИ	END	ЕНДОКРИНОЛОШКИ	HEAR	СЛУХ	HEM	ХЕМАТОЛОШКИ
HEP	ХЕПАТАЛНИ	IMM	ИМУНОЛОШКИ	INF	НЕПЛОДНОСТ	MET	МЕТАБОЛНИ	MUSC	МУСКУЛНИ
NEUR	НЕВРОЛОШКИ	OPTH	ОФТАЛМОЛОШКИ	REN	БУБРЕЖНИ	RESP	РЕСПИРАТОРНИ	SD	СЕКСУАЛЕН РАЗВОЈ
SKEL	СКЕЛЕТНИ	SKIN	КОЖНИ						

Болеста може да се класифицира во неколку видови. Наведената класификација се заснова на најчестите симптоми поврзани со секоја состојба. Степенот на сериозноста на состојбата може да варира и зависи од специфичната мутација, знаци и симптоми.

Резултатите и можните следни чекори секогаш треба да се разгледуваат во контекст на другите клинички критериуми и треба целосно да се дискутираат со вашиот доктор. Генетско советување се препорачува кога ќе се добие резултат со висок ризик.

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