



MONOGENSKE BOLESTI KOJE ISPITUJE VERAgene

BOLEST	GEN	KLASIFIKACIJA
3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	<i>HMGL</i>	MET
3-Methylcrotonyl-CoA Carboxylase Deficiency 1	<i>MCCC1</i>	MET
3-Methylcrotonyl-CoA Carboxylase Deficiency 2	<i>MCCC2</i>	MET
Abetalipoproteinemia	<i>MTTP</i>	DIG, NEUR, OPTH, HEM
Acyl-CoA Oxidase I Deficiency	<i>ACOX1</i>	NEUR
Aicardi-Goutières Syndrome	<i>SAMHD1</i>	NEUR
Alport Syndrome, X-Linked	<i>COL4A5</i>	REN, OPTH, HEAR
Alstrom Syndrome	<i>ALMS1</i>	OPTH, HEAR, REN, CARD
Andermann Syndrome	<i>SLC12A6</i>	MUSC, NEUR
Aromatase Deficiency	<i>CYP19A1</i>	SD
Arthrogryposis Mental Retardation Seizures	<i>SLC35A3</i>	MET
Asparagine Synthetase Deficiency	<i>ASNS</i>	NEUR
Aspartylglycosaminuria	<i>AGA</i>	MET, NEUR
Autosomal Recessive Polycystic Kidney Disease	<i>PKHD1</i>	REN
Bardet-Biedl Syndrome (BBS1-related)	<i>BBS1</i>	OPTH, MET, END
Bardet Biedl Syndrome (BBS12-related)	<i>BBS12</i>	OPTH
Beta Thalassemia	<i>HBB</i>	HEM
Biotinidase Deficiency	<i>BTD</i>	MET
Canavan Disease	<i>ASPA</i>	NEUR
Carpenter Syndrome	<i>RAB23</i>	SKEL
Choreacanthocytosis	<i>VPST3A</i>	NEUR
Choroideremia, X-Linked	<i>CHM</i>	OPTH
Citrin Deficiency	<i>SLC25A13</i>	MET
Combined Oxidative Phosphorylation Deficiency 3	<i>TSM</i>	NEUR, MET, CARD
Congenital Disorder of Glycosylation, Type 1A (PMM2-related)	<i>PMM2</i>	MET
Congenital Neutropenia (HAX1-related)	<i>HAX1</i>	IMM
Crigler Najjar Syndrome, Type I	<i>UGT1A1</i>	MET
Cystic Fibrosis *	<i>CFTR</i>	RESP, DIG
Factor XI Deficiency	<i>F11</i>	HEM
Familial Dysautonomia	<i>IKBKAP</i>	NEUR
Fanconi Anemia, Type C	<i>FANCC</i>	IMM
Fanconi Anemia, Type G	<i>FANCG</i>	HEM
Gaucher Disease	<i>GBA</i>	NEUR, HEP, CARD
Glutaric Acidemia, Type 2A	<i>ETFA</i>	MET
Glycine Encephalopathy (GLDC-related)	<i>GLDC</i>	MET
Glycogen Storage Disease, Type 1A	<i>G6PC</i>	MET
Glycogen Storage Disease, Type 1B	<i>SLC37A4</i>	MET
Glycogen Storage Disease, Type 3	<i>AGL</i>	MET
Glycogen Storage Disease, Type 7	<i>PFKM</i>	MET
GRACILE Syndrome	<i>BCS1L</i>	MET
Hereditary Fructose Intolerance	<i>ALDOB</i>	MET
Homocystinuria, Type cbIE	<i>MTRR</i>	MET
Hydrolethalus Syndrome	<i>HYLS1</i>	NEUR, CARD
Inclusion Body Myopathy, Type 2	<i>GNE</i>	MUSC
Isovaleric Acidemia	<i>IVD</i>	MET
Joubert Syndrome, Type 2	<i>TMEM216</i>	NEUR
Junctional Epidermolysis Bullosa, Herlitz Type	<i>LAMC2</i>	SKIN
Lamellar Ichthyosis, Type 1	<i>TGM1</i>	MET
Leber Congenital Amaurosis (LCA5-related)	<i>LCA5</i>	OPTH
Leigh Syndrome, French-Canadian Type	<i>LRPPRC</i>	NEUR, MUSC
Leukoencephalopathy with Vanishing White Matter	<i>EIF2B5</i>	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 cb1C	MMACHC	MET
Methylmalonic Aciduria and Homocystinuria, Type cb1D	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	KARDIOLOŠKE	DIG	PROBAVNE	END	ENDOKRINOLOŠKE	HEAR	SLUŠNE	HEM	HEMATOLOŠKE
HEP	JETRENE	IMM	IMUNOLOŠKE	INF	NEPLODNOST	MET	METABOLIČKE	MUSC	MIŠIČNE
NEUR	NEUROLOŠKE	OPTH	OFTALMOLOŠKE	REN	BUBREŽNE	RESP	RESPIRATORNE	SD	SPOLNI RAZVOJ
SKEL	KOŠTANE	SKIN	KOŽNE						

Bolesti mogu biti klasificirane na više načina. Ovdje korištena klasifikacija utemeljena je na najčešćim i najizraženijim simptomima kojima se pojedina bolest manifestira.

Rezultate i moguće daljnje korake treba uvijek razmatrati u kontekstu ostalih kliničkih kriterija i u neposrednoj suradnji s vašim liječnikom. Kada dobiveni rezultati ukazuju na visok rizik preporučuje se genetsko savjetovanje.