RODINA - PRE PÁR

ZOZNAM TESTOVANÝCH OCHORENÍ

Zoznam testovaných ochorení je rozšírenou verziou služby RODINA určenej pre jednotlivca zostavenej podľa medzinárodných odporúčaní (ACMG), ktorá zahŕňa závažné ochorenia so **zvýšenou frekvenciou prenášačov** v populácii. Možnosť kombinovať výsledky testov oboch partnerov však pridáva možnosť sledovať nielen ochorenia vyskytujúce sa v populácii s vyššou frekvenciou, ale aj tie menej časté.

Zoznam ochorení sa môže meniť z dôvodu jeho aktualizácie podľa súčasných vedeckých, klinických poznatkov a technických obmedzení použitej sekvenačnej metódy.

TESTOVANÉ DEDIČNÉ OCHORENIA	DEDIČNOSŤ
Abetalipoproteinemia	AR
Acrodermatitis enteropathica	AR
Acyl-CoA dehydrogenase, medium chain, deficiency of	AR
Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR
Adrenal hypoplasia, congenital	XL
Agammaglobulinemia, X-linked 1	XL
Achondrogenesis Ib	AR
Achromatopsia 3	AR
Aicardi Goutieres syndrome 2	AR
Aicardi Goutieres syndrome 4	AR
Aicardi Goutieres syndrome 5	AR
Albinism, oculocutaneous, type IA / type IB	AR
Alkaptonuria	AR
Alpha-methylacetoacetic aciduria	AR
Alpha-thalassemia/impaired intellectual development syndrome	XL
Alstrom syndrome	AR
Anterior segment dysgenesis 6, multiple subtypes	AR
Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	AR
Argininemia	AR
Argininosuccinate aciduria	AR
Arts syndrome	XL
Aspartylglucosaminuria	AR
Ataxia-telangiectasia	AR
Atelosteogenesis, type II	AR
Atransferrinemia	AR
Auditory neuropathy, autosomal recessive, 1	AR
Bardet-Biedl syndrome 1	AR
Bardet-Biedl syndrome 2	AR
Bardet-Biedl syndrome 4	AR
Bardet-Biedl syndrome 5	AR
Bardet-Biedl syndrome 6	AR
Bardet-Biedl syndrome 7	AR
Bardet-Biedl syndrome 9	AR
Bardet-Biedl syndrome 10	AR
Bardet-Biedl syndrome 12	AR

ACMG - American College of Medical Genetics; **AR** - autozómovo recesívna dedičnosť, **XL** - dedičnosť viazaná na pohlavný chromozóm X

Autor fotky na pozadí: Surendra Basnet (pexels.com)

TESTOVANÉ DEDIČNÉ OCHORENIA	DEDIČNOSŤ
Bardet-Biedl syndrome 13	AR
Bartter syndrome, type 1	AR
Bartter syndrome, type 2	AR
Becker muscular dystrophy	XL
Biotinidase deficiency	AR
Bjornstad syndrome	AR
Bloom syndrome	AR
Brittle cornea syndrome 1	AR
Canavan disease	AR
Carbamoylphosphate synthetase I deficiency	AR
Cardiomyopathy, dilated, 1X	AR
Carnitine-acylcarnitine translocase deficiency	AR
Carnitine deficiency, systemic primary	AR
Cerebral creatine deficiency syndrome 2	AR
Cerebrotendinous xanthomatosis	AR
Ceroid lipofuscinosis, neuronal, 3	AR
Ceroid lipofuscinosis, neuronal, 5	AR
Ceroid lipofuscinosis, neuronal, 7	AR
Ceroid lipofuscinosis, neuronal, 8	AR
Citrullinemia type 1	AR
COACH syndrome 1	AR
COACH syndrome 2	AR
Cohen syndrome	AR
Combined cellular and humoral immune defects with granulomas	AR
Combined immunodeficiency, severe, B cell-negative	AR
Combined immunodeficiency, X-linked, moderate / severe	XL
Combined immunodeficiency 104, severe	AR
Congenital bilateral absence of vas deferens	AR
Congenital disorder of glycosylation, type Ia	AR
Congenital disorder of glycosylation, type Ic	AR
Corneal endothelial dystrophy, autosomal recessive / Corneal endothelial dystrophy and perceptive deafness	AR
Cortical dysplasia, complex, with other brain malformations 14A, (bilateral frontoparietal)	AR
CPT deficiency, hepatic, type IA	AR
CPT II deficiency, infantile / lethal neonatal	AR
Cystic fibrosis	AR
D-bifunctional protein deficiency	AR
De la Chapelle dysplasia	AR
Deafness, autosomal recessive 1A	AR
Deafness, autosomal recessive 1B	AR
Deafness, autosomal recessive 2	AR
Deafness, autosomal recessive 3	AR
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	AR
Deafness, autosomal recessive 9	AR
Deafness, autosomal recessive 18A	AR
Deafness, autosomal recessive 23	AR
Deafness, autosomal recessive 49	AR
Deafness, digenic GJB2/GJB6	AR, DD
Deafness, X-linked 1	XL
Dent disease 2	XL

AR - autozómovo recesívna dedičnosť, **XL** - dedičnosť viazaná na pohlavný chromozóm X, **DD** - digénová dedičnosť
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TESTOVANÉ DEDIČNÉ OCHORENIA	DEDIČNOSŤ
Desbuquois dysplasia 1	AR
Diabetes mellitus, neonatal, with congenital hypothyroidism	AR
Diarrhea 1, secretory chloride, congenital	AR
Diastrophic dysplasia / Diastrophic dysplasia, broad bone-platyspondylic variant	AR
Dihydrolipoamide dehydrogenase deficiency	AR
Disordered steroidogenesis due to cytochrome P450 oxidoreductase	AR
Distal renal tubular acidosis 2 with progressive sensorineural hearing loss	AR
Donnai–Barrow syndrome	AR
Duchenne muscular dystrophy	XL
Dyskeratosis congenita, X-linked	XL
Ectodermal dysplasia 1, hypohidrotic, X-linked	XL
Emery-Dreifuss muscular dystrophy 1, X-linked	XL
Epidermolysis bullosa, junctional 1A, intermediate / junctional 1B, severe	AR
Epidermolysis bullosa, junctional 2A, intermediate / junctional 2B, severe / junctional 2C, laryngoonychocutaneous	AR
Epidermolysis bullosa, junctional 3A, intermediat / junctional 3B, severe	AR
Epilepsy, early-onset, 4, vitamin B6-dependent	AR
Epilepsy, progressive myoclonic 3, with or without intracellular inclusions	AR
Epiphyseal dysplasia, multiple, 4	AR
Epiphyseal dysplasia, multiple, 7	AR
Escobar syndrome	AR
Ethylmalonic encephalopathy	AR
Fabry disease / Fabry disease, cardiac variant	XL
Fanconi anemia, complementation, group A	AR
Fanconi anemia, complementation, group C	AR
Fanconi anemia, complementation, group G	AR
Fatty liver, acute, of pregnancy	AR
Fetal akinesia deformation sequence 2	AR
Fetal akinesia deformation sequence 3	AR
Fraser syndrome 1	AR
Fraser syndrome 3	AR
Fructose intolerance, hereditary	AR
Galactokinase deficiency with cataracts	AR
Galactose epimerase deficiency	AR
Galactosemia	AR
Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset	AR
Glutaric acidemia type I	AR
Glutaric acidemia type IIA	AR
Glutaric acidemia type IIB	AR
Glutaric acidemia type IIC	AR
Glycine encephalopathy 1	AR
Glycine encephalopathy 2	AR
Glycogen storage disease Ia	AR
Glycogen storage disease Ib / Ic	AR
Glycogen storage disease II	AR
Glycogen storage disease IIIa / IIIb	AR
GM1-gangliosidosis, type I / type II / type III	AR
GM2-gangliosidosis, several forms	AR
Gout, PRPS-related	XL
GRACILE syndrome	AR

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TESTOVANÉ DEDIČNÉ OCHORENIA	DEDIČNOSŤ
Heimler syndrome 1	AR
Heimler syndrome 2	AR
HELLP syndrome, maternal, of pregnancy	AR
Hemochromatosis, type 2A	AR
Hemophagocytic lymphohistiocytosis, familial, 3	AR
Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease	AR
Hemophilia B	XL
Hermansky-Pudlak syndrome 1	AR
Hermansky-Pudlak syndrome 2	AR
Hermansky-Pudlak syndrome 4	AR
HMG-CoA lyase deficiency	AR
Homocystinuria, B6-responsive and nonresponsive types	AR
Homocystinuria-megaloblastic anemia, cbl E type	AR
Hydrocephalus, congenital, X-linked	XL
Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	AR
Hyperoxaluria, primary, type 1	AR
Hyperphenylalaninemia, BH4-deficient, A	AR
Hyperphenylalaninemia, BH4-deficient, C	AR
Charcot-Marie-Tooth disease, type 4K	AR
Charcot-Marie-Tooth disease, X-linked recessive, 5	XL
Cholesteryl ester storage disease	AR
Ichthyosis, congenital, autosomal recessive 1	AR
Ichthyosis, congenital, autosomal recessive 4A / 4B (harlequin)	AR
Ichthyosis, congenital, autosomal recessive 5	AR
Immunodeficiency, X-linked, with hyper-IgM	XL
Infantile neuroaxonal dystrophy 1	AR
Insensitivity to pain, congenital, with anhidrosis	AR
Intellectual disability-hypotonic facies syndrome, X-linked	XL
Isolated growth hormone deficiency, type III, with agammaglobulinemia	XL
Isovaleric acidemia	AR
Johanson-Blizzard syndrome	AR
Joubert syndrome 2	AR
Joubert syndrome 3	AR
Joubert syndrome 5	AR
Joubert syndrome 6	AR
Joubert syndrome 7	AR
Joubert syndrome 9	AR
Joubert syndrome 28	AR
Krabbe disease	AR
Leber congenital amaurosis 5	AR
Leber congenital amaurosis 8	AR
Leber congenital amaurosis 10	AR
Leukoencephalopathy with vanishing white matter with or without ovarian failure, 2 / 5	AR
LCHAD deficiency	AR
LIG4 syndrome	AR
Lipoid adrenal hyperplasia	AR
Liver failure, transient infantile	AR
Lowe syndrome	XL
Lysinuric protein intolerance	AR

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TESTOVANÉ DEDIČNÉ OCHORENIA	DEDIČNOSŤ
Macular dystrophy with central cone involvement	AR
Mannosidosis, alpha-, types I and II	AR
Maple syrup urine disease, type Ia	AR
Maple syrup urine disease, type Ib	AR
Maple syrup urine disease, type II	AR
MASA syndrome	XL
McKusick-Kaufman syndrome	AR
Meckel syndrome 1	AR
Meckel syndrome 2	AR
Meckel syndrome 3	AR
Meckel syndrome 4	AR
Meckel syndrome 5	AR
Meckel syndrome 6	AR
Megalencephalic leukoencephalopathy with subcortical cysts 1	AR
Menkes disease	XL
Metachromatic leukodystrophy	AR
Methylmalonic aciduria and homocystinuria, cblC type	AR
Methylmalonic aciduria, mut(0) type	AR
Methylmalonic aciduria, vitamin B12-responsive, cblA type	AR
Methylmalonic aciduria, vitamin B12-responsive, cblB type	AR
Microcephalic osteodysplastic primordial dwarfism, type II	AR
Mitochondrial complex I deficiency, nuclear type 20	AR
Mitochondrial complex III deficiency, nuclear type 1	AR
Mitochondrial complex IV deficiency, nuclear type 1	AR
Mitochondrial complex IV deficiency, nuclear type 2	AR
Mitochondrial complex IV deficiency, nuclear type 5	AR
Mitochondrial DNA depletion syndrome 1 (MNGIE type)	AR
Mitochondrial trifunctional protein deficiency 1	AR
Mitochondrial trifunctional protein deficiency 2	AR
Mucolipidosis type II alpha/beta / type III alpha/beta	AR
Mucolipidosis III gamma	AR
Mucolipidosis IV	AR
Mucopolysaccharidosis type Ih / Ih/s / Is	AR
Mucopolysaccharidosis type II	XL
Mucopolysaccharidosis type IIIA (Sanfilippo A)	AR
Mucopolysaccharidosis type IIID	AR
Mucopolysaccharidosis type IVA	AR
Mucopolysaccharidosis type IVB (Morquio)	AR
Mucopolysaccharidosis VII	AR
Multiple pterygium syndrome, lethal type	AR
Multiple sulfatase deficiency	AR
Muscular dystrophy, limb-girdle, autosomal recessive 3	AR
Muscular dystrophy, limb-girdle, autosomal recessive 4	AR
Muscular dystrophy, limb-girdle, autosomal recessive 5	AR
Muscular dystrophy-dystroglycanopathy, type A, 1 (congenital with brain and eye anomalies) / type B, 1 (congenital with impaired intellectual development) / type C, 1 (limb-girdle)	AR
Muscular dystrophy-dystroglycanopathy, type A, 2 (congenital with brain and eye anomalies) / type B, 2 (congenital with impaired intellectual development) / type C, 2 (limb-girdle)	AR
Muscular dystrophy-dystroglycanopathy, type A, 3 (congenital with brain and eye anomalies) / type B, 3 (congenital with impaired intellectual development) / type C, 3 (limb-girdle)	AR

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TESTOVANÉ DEDIČNÉ OCHORENIA	DEDIČNOSŤ
Muscular dystrophy-dystroglycanopathy, type A, 4 (congenital with brain and eye anomalies) / type B, 4 (congenital without impaired intellectual development) / type C, 4 (limb-girdle)	AR
Muscular dystrophy-dystroglycanopathy, type A, 5 (congenital with brain and eye anomalies) / type B, 5 (congenital with or without impaired intellectual development) / type C, 5 (limb-girdle)	AR
Myasthenic syndrome, congenital, 4B, fast-channel / 4C, associated with acetylcholine receptor deficiency	AR
Myasthenic syndrome, congenital, 5	AR
Myasthenic syndrome, congenital, 6, presynaptic	AR
Myasthenic syndrome, congenital, 10	AR
Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	AR
Myasthenic syndrome, congenital, 12, with tubular aggregates	AR
Myopathy, centronuclear, X-linked	XL
Nephronophthisis 1, juvenile	AR
Nephronophthisis 11	AR
Nephrotic syndrome, type 1	AR
Nephrotic syndrome, type 2	AR
Nephrotic syndrome, type 3	AR
Neu-Laxova syndrome 1	AR
Neurodegeneration with brain iron accumulation 2B	AR
Niemann-Pick disease, type A / type B	AR
Occipital horn syndrome	AR
Omenn syndrome	AR
Opitz GBBB syndrome	AR
Ornithine transcarbamylase deficiency	AR
Osteopetrosis, autosomal recessive 1	AR
Parkinson disease 14, autosomal recessive	AR
Pendred syndrome	AR
Peroxisome biogenesis disorder 1A (Zellweger) / 1B (NALD/IRD)	XL
Peroxisome biogenesis disorder 3A (Zellweger) / 3B	AR
Peroxisome biogenesis disorder 4A (Zellweger) / 4B	AR
Peroxisome biogenesis disorder 5A (Zellweger) / 5B	AR
Peroxisome biogenesis disorder 6A (Zellweger) / 6B	AR
Peroxisome biogenesis disorder 7A (Zellweger) / 7B	AR
Peroxisome biogenesis disorder 9B	AR
Perrault syndrome 1	AR
Phenylketonuria	AR
Phosphoglycerate dehydrogenase deficiency	AR
Phosphoribosylpyrophosphate synthetase superactivity	XL
Polycystic kidney disease 4, with or without hepatic disease	AR
Pontocerebellar hypoplasia, type 1B	AR
Pontocerebellar hypoplasia, type 6	AR
Prolidase deficiency	AR
Propionicacidemia	AR
Pseudohermaphroditism, male, with gynecomastia	AR
Pseudovaginal perineoscrotal hypospadias	AR
Pycnodysostosis	AR
Pyridoxamine 5'-phosphate oxidase deficiency	AR
Pyruvate carboxylase deficiency	AR
RENI syndrome	AR
Retinitis pigmentosa 12	AR
Retinitis pigmentosa 39	AR

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Retinitis pigmentosa 61	AR
Retinitis pigmentosa 74	AR
Retinitis pigmentosa 76	AR
Retinitis pigmentosa 93	AR
Retinoschisis	XL
Rhizomelic chondrodysplasia punctata, type 1	AR
Salla disease	AR
SCID, autosomal recessive, T-negative/B-positive type	AR
Segawa syndrome, recessive	AR
Senior-Loken syndrome 1	AR
Senior-Loken syndrome 6	AR
Short-rib thoracic dysplasia 3 with or without polydactyly	AR
Schimke immunoosseous dysplasia	AR
Sialic acid storage disorder, infantile	AR
Sjogren-Larsson syndrome	AR
Smith-Lemli-Opitz syndrome	AR
Spastic ataxia, Charlevoix-Saguenay type	AR
Spastic paraplegia 15, autosomal recessive	AR
Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome	AR
Surfactant metabolism dysfunction, pulmonary, 3	AR
Tay-Sachs disease	AR
Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	AR
Thiamine-responsive megaloblastic anemia syndrome	AR
Thrombocytopenia, congenital amegakaryocytic	AR
Thrombophilia 8, X-linked, due to factor IX defect	XL
Thrombosis, hyperhomocysteinemic	AR
Trichohepatoenteric syndrome 1	AR
Trichothiodystrophy 1, photosensitive	AR
Tumoral calcinosis, hyperphosphatemic, familial, 1	AR
Tyrosinemia, type I	AR
Tyrosinemia, type II	AR
Usher syndrome, type 1B	AR
Usher syndrome, type 1C	AR
Usher syndrome, type 1F	AR
Usher syndrome, type 2A	AR
Usher syndrome, type 2C	AR
Usher syndrome, type 3A	AR
Ventricular tachycardia, catecholaminergic polymorphic, 2	AR
Vitamin D-dependent rickets, type I	AR
VLCAD deficiency	AR
Wilson disease	AR
Wolcott-Rallison syndrome	AR
Wolman disease	AR
Xeroderma pigmentosum, group A	AR
Xeroderma pigmentosum, group C	AR
Xeroderma pigmentosum, group D	AR
Xeroderma pigmentosum, variant type	AR

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