TvojaDNA

RODINA - PRE JEDNOTLIVCA

ZOZNAM TESTOVANÝCH OCHORENÍ

Zoznam ochorení bol zostavený podľa medzinárodných odporúčaní (ACMG) pre testovanie statusu prenášačstva závažných **dedičných ochorení** s dôrazom na **zvýšenú frekvenciu prenášačov** vybraných ochorení v populácii, kvôli ktorej rastie riziko "stretu" dvoch prenášačov pri plánovaní rodiny.

Výnimkou sú ochorenia viazané na pohlavný chromozóm X. Frekvencia prenášačok v tomto prípade nehrá úlohu pri výbere ochorenia. Dôležitým kritériom bola závažnosť ochorenia a fakt, že v prípade potvrdeného statusu prenášačstva u ženy vzniká až 50% riziko narodenia chlapca s daným ochorením.

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

TESTOVANÉ DEDIČNÉ OCHORENIA	DEDIČNOSŤ	FREKVENCIA PRENÁŠAČOV V POPULÁCII*
Acyl-CoA dehydrogenase, medium chain, deficiency of	AR	≥1:50
Adrenal hypoplasia, congenital	XL	_
Achondrogenesis Ib	AR	1/150 - 1/200
Achromatopsia 3	AR	1/100 - 1/150
Aicardi Goutieres syndrome 2	AR	1/150 - 1/200
Alpha-methylacetoacetic aciduria	AR	1/150 - 1/200
Argininosuccinate aciduria	AR	1/150 - 1/200
Aspartylglucosaminuria	AR	1/100 - 1/150
Atelosteogenesis, type II	AR	_
Bardet-Biedl syndrome 1	AR	1/150 - 1/200
Bardet-Biedl syndrome 2	AR	1/100 - 1/150
Becker muscular dystrophy	XL	_
Biotinidase deficiency	AR	1/150 - 1/200
Canavan disease	AR	≥1:50
Cardiomyopathy, dilated, 1X	AR	≥1:50
Cerebrotendinous xanthomatosis	AR	1/100 - 1/150
Congenital bilateral absence of vas deferens	AR	_
Congenital disorder of glycosylation, type Ia	AR	≥1:50
CPT II deficiency, infantile / lethal neonatal	AR	1/50 - 1/100
Cystic fibrosis	AR	≥1:50
De la Chapelle dysplasia	AR	_
Deafness, autosomal recessive 1A	AR	≥1:50
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	AR	≥1:50
Deafness, autosomal recessive 23	AR	1/100 - 1/150
Diastrophic dysplasia / Diastrophic dysplasia, broad bone-platyspondylic variant	AR	_
Duchenne muscular dystrophy	XL	_

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

* **Zdroj:** Gregg AR, Aarabi M, Klugman S, Leach NT, Bashford MT, Goldwaser T, Chen E, Sparks TN, Reddi HV, Rajkovic A, Dungan JS; ACMG Professional Practice and Guidelines Committee. Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2021 Oct;23(10):1793-1806. doi: 10.1038/s41436-021-01203-z. Epub 2021 Jul 20. Erratum in: Genet Med. 2021 Aug 27;: PMID: 34285390; PMCID: PMC8488021.

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

TESTOVANÉ DEDIČNÉ OCHORENIA	DEDIČNOSŤ	FREKVENCIA PRENÁŠAČOV V POPULÁCII*
Epiphyseal dysplasia, multiple, 4	AR	1/150 - 1/200
Fabry disease / Fabry disease, cardiac variant	XL	_
Fanconi anemia, complementation, group C	AR	1/50 - 1/100
Fructose intolerance, hereditary	AR	1/50 - 1/100
Galactosemia	AR	1/100 - 1/150
Glycogen storage disease Ia	AR	1/50 - 1/100
Glycogen storage disease Ib / Ic	AR	1/100 - 1/150
Glycogen storage disease II	AR	1/50 - 1/100
GM2-gangliosidosis, several forms	AR	_
Hemophilia B	XL	_
Homocystinuria, B6-responsive and nonresponsive types	AR	1/150 - 1/200
Hydrocephalus, congenital, X-linked	XL	_
Hyperoxaluria, primary, type 1	AR	1/100 - 1/150
Joubert syndrome 2	AR	1/100 - 1/150
Joubert syndrome 3	AR	1/50 - 1/100
Joubert syndrome 5	AR	1/50 - 1/100
Leber congenital amaurosis 10	AR	1/50 - 1/100
Maple syrup urine disease, type Ib	AR	1/50 - 1/100
MASA syndrome	XL	_
Meckel syndrome 2	AR	1/100 - 1/150
Meckel syndrome 4	AR	_
Megalencephalic leukoencephalopathy with subcortical cysts 1	AR	1/150 - 1/200
Metachromatic leukodystrophy	AR	1/150 - 1/200
Methylmalonic aciduria and homocystinuria, cblC type	AR	1/100 - 1/150
Methylmalonic aciduria, mut(0) type	AR	1/50 - 1/100
Mucolipidosis type II alpha/beta / type III alpha/beta	AR	1/100 - 1/150
Mucolipidosis IV	AR	1/100 - 1/150
Mucopolysaccharidosis type Ih / Ih/s / Is	AR	1/150 - 1/200
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	≥1:50
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	1/150 - 1/200
Myasthenic syndrome, congenital, 4B, fast-channel / 4C, associated with acetylcholine receptor deficiency	AR	1/50 - 1/100
Nephrotic syndrome, type 1	AR	≥1:50
Niemann-Pick disease, type A / type B	AR	1/50 - 1/100
Ornithine transcarbamylase deficiency	XL	_
Pendred syndrome	AR	≥1:50
Phenylketonuria	AR	≥1:50
Polycystic kidney disease 4, with or without hepatic disease	AR	1/150 - 1/200
Pontocerebellar hypoplasia, type 6	AR	1/150 - 1/200

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TESTOVANÉ DEDIČNÉ OCHORENIA	DEDIČNOSŤ	FREKVENCIA PRENÁŠAČOV V POPULÁCII*
Retinitis pigmentosa 39	AR	_
Retinitis pigmentosa 74	AR	1/100 - 1/150
Retinoschisis	XL	_
Senior-Loken syndrome 6	AR	_
Smith-Lemli-Opitz syndrome	AR	≥1:50
Tay-Sachs disease	AR	≥1:50
Thrombophilia 8, X-linked, due to factor IX defect	XL	_
Thrombosis, hyperhomocysteinemic	AR	_
Trichothiodystrophy 1, photosensitive	AR	≥1:50
Tyrosinemia, type I	AR	1/100 - 1/150
Usher syndrome, type 1F	AR	1/100 - 1/150
Usher syndrome, type 2A	AR	1/50 - 1/100
Vitamin D-dependent rickets, type I	AR	1/150 - 1/200
VLCAD deficiency	AR	1/150 - 1/200
Wilson disease	AR	≥1:50
Xeroderma pigmentosum, group C	AR	≥1:50
Xeroderma pigmentosum, group D	AR	_

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