

## Vizsgált genetikai betegségek listája

105 betegség, 458 mutáció, 925 próba

ABCC8-Related Hyperinsulinism	Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency
Achromatopsia	Hurler Syndrome
Alkaptonuria	Hypophosphatasia, Autosomal Recessive
Alpha-1 Antitrypsin Deficiency	Inclusion Body Myopathy 2
Alpha-Mannosidosis	Isovaleric Acidemia
Andermann Syndrome	Joubert Syndrome 2
ARSACS	Krabbe Disease
Aspartylglycosaminuria	Limb-Girdle Muscular Dystrophy Type 2D
Ataxia With Vitamin E Deficiency	Limb-Girdle Muscular Dystrophy Type 2E
Ataxia-Telangiectasia	Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency
Autosomal Recessive Polycystic Kidney Disease	Maple Syrup Urine Disease Type 1B
Bardet-Biedl Syndrome, BBS1-Related	Maple Syrup Urine Disease Type 3
Bardet-Biedl Syndrome, BBS10-Related	Medium Chain Acyl-CoA Dehydrogenase Deficiency
Beta Thalassemia <b>ACOG</b>	Megalencephalic Leukoencephalopathy With Subcortical Cysts
Biotinidase Deficiency	Metachromatic Leukodystrophy
Bloom Syndrome <b>ACMG</b>	*MTHFR Deficiency
Canavan Disease <b>ACMG ACOG</b>	Mucopolidosis IV <b>ACMG</b>
Carnitine Palmitoyltransferase IA Deficiency	Muscle-Eye-Brain Disease
Carnitine Palmitoyltransferase II Deficiency	NEB-Related Nemaline Myopathy
Cartilage-Hair Hypoplasia	Niemann-Pick Disease Type C
Choroideremia	Niemann-Pick Disease, SMPD1-Associated <b>ACMG</b>
Citrullinemia Type 1	Nijmegen Breakage Syndrome
CLN3-Related Neuronal Ceroid Lipofuscinosis	Northern Epilepsy
CLN5-Related Neuronal Ceroid Lipofuscinosis	Pendred Syndrome
Cohen Syndrome	PEX1-Related Zellweger Syndrome Spectrum
Congenital Disorder of Glycosylation Type Ia	Phenylalanine Hydroxylase Deficiency
Congenital Disorder of Glycosylation Type Ib	Polyglandular Autoimmune Syndrome Type 1
Congenital Finnish Nephrosis	Pompe Disease
Costeff Optic Atrophy Syndrome	PPT1-Related Neuronal Ceroid Lipofuscinosis
Cystic Fibrosis <b>ACMG ACOG</b>	Primary Carnitine Deficiency
Cystinosis	Primary Hyperoxaluria Type 1
D-Bifunctional Protein Deficiency	Primary Hyperoxaluria Type 2
*Factor V Leiden Thrombophilia	PROP1-Related Combined Pituitary Hormone Deficiency
Factor XI Deficiency	*Prothrombin Thrombophilia
Familial Dysautonomia <b>ACMG ACOG</b>	Pseudocholinesterase Deficiency
Familial Mediterranean Fever	Pycnodysostosis
Fanconi Anemia Type C <b>ACMG</b>	Rhizomelic Chondrodysplasia Punctata Type 1
Galactosemia	Salla Disease
Gaucher Disease <b>ACMG</b>	Segawa Syndrome
GJB2-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness	Short Chain Acyl-CoA Dehydrogenase Deficiency
*Glucose-6-Phosphate Dehydrogenase Deficiency	Sickle Cell Disease <b>ACOG</b>
Glutaric Acidemia Type 1	Sjogren-Larsson Syndrome
Glycogen Storage Disease Type Ia	Smith-Lemli-Opitz Syndrome
Glycogen Storage Disease Type Ib	Spinal Muscular Atrophy <b>ACMG</b>
Glycogen Storage Disease Type III	Steroid-Resistant Nephrotic Syndrome
Glycogen Storage Disease Type V	Sulfate Transporter-Related Osteochondrodysplasia
GRACILE Syndrome	Tay-Sachs Disease <b>ACMG ACOG</b>
Hereditary Fructose Intolerance	TPP1-Related Neuronal Ceroid Lipofuscinosis
Hereditary Thymine-Uraciluria	Tyrosinemia Type I
Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related	Usher Syndrome Type 1F
Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related	Usher Syndrome Type 3
Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related	Very Long Chain Acyl-CoA Dehydrogenase Deficiency
Hexosaminidase A Deficiency	Wilson Disease
*HFE-Associated Hereditary Hemochromatosis	X-Linked Juvenile Retinoschisis

**ACOG** = Az Amerikai Szülészeti-Nőgyógyászati Társaság javasolja a szűrését**ACMG** = Az Amerikai Orvosgenetikai Szakmai Kollégium javasolja a szűrését

\* - Csak a beküldő orvos kérésére része a tesztnek.