

Lizoszómális tárolási betegségek (Lysosomal storage disorders) génpanel v:2025-05-23

Panel tartalma 151 gén:

ABCC8, ABCD1, ABHD5, ACOX1, ACY1, ADAMTS10, ADAMTSL2, ADSL, AGA, ALDH7A1, AMACR, AMT, ANTXR2, AP3B1, ARG1, ARSA, ARSB, ARSG, ASAH1, ASPA, ATG5, ATP13A2, ATP6AP1, BLOC1S3, BLOC1S6, BTBD, CHIT1, CLCN5, CLCN7, CLN3, CLN5, CLN6, CLN8, COL11A2, COL2A1, CTNS, CTSA, CTSC, CTSD, CTSF, CTSK, DHCR7, DNAJC5, DNM1L, DPYD, DTNBP1, DYM, ETFA, ETFB, ETFDH, FAR1, FBN1, FH, FIG4, FOLR1, FUCA1, GAA, GALC, GALNS, GAMT, GBA, GCDH, GLA, GLB1, GLDC, GM2A, GNE, GNPAT, GNPTAB, GNPTG, GNS, GPC3, GRN, GUSB, HEXA, HEXB, HGSNAT, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, HSD17B4, HYAL1, IDS, IDUA, KCTD7, KMT2D, L2HGDH, LAMA2, LAMP2, LDB3, LIPA, LTBP2, LYST, MAN1B1, MAN2B1, MANBA, MCOLN1, MFSD8, MLPH, MMP14, MOCS1, MOCS2, MYO5A, MYOT, NAGA, NAGLU, NEU1, NPC1, NPC2, NPR2, OCRL, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGK1, PHYH, PNPLA2, PPT1, PSAP, QDPR, RAB27A, RAI1, SCARB2, SCP2, SGSH, SLC17A5, SLC25A15, SLC46A1, SLC9A6, SMPD1, SUMF1, SUOX, TCF4, TPP1, TRIM37, VPS16, VPS33A

Forrás:

Lysosomal storage disorder (Version 3.3)

<https://panelapp.genomicsengland.co.uk/panels/529/>

Invitae Comprehensive Lysosomal Storage Disorders Panel

<https://www.invitae.com/en/physician/tests/06170/>

Lysosomal Storage Disorders Panel

<https://www.preventiongenetics.com/testInfo?val=Lysosomal+Storage+Disorders+Panel>

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