

Lizoszómális tárolási betegségek (Lysosomal storage disorders) génpanel v:2022.02.09.

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, CLN2 (TPP1), 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, TRIM37, VPS16, VPS33A.

Forrás:

Lysosomal storage disorder (Version 1.76)

<https://panelapp.genomicsengland.co.uk/panels/529/> (Megtekintve: 2022.02.09.)

Lysosomal Storage Diseases (LSD) Panel

<https://www.centogene.com/science/centopedia/lysosomal-storage-diseases-lsd-panel.html>

(Megtekintve: 2022.02.09.)

Lysosomal Disorders Panel

<https://www.genedx.com/tests/detail/lysosomal-disorder-panel-991>

(Megtekintve: 2022.02.09.)

Invitae Comprehensive Lysosomal Storage Disorders Panel

<https://www.invitae.com/en/physician/tests/06170/> (Megtekintve: 2022.02.09.)

Lysosomal Storage Disorders Panel

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

(Megtekintve: 2022.02.09.)