

Full name:	Lysosomal Storage (64 genes) - VUB
Description:	https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6393649/table/mgg3527-tbl-0001/?report=objectonly (version 1) https://www.brightcore.be/gene-panels/mendeliome---lysosomal-storage-disease-(lsd)---v11
Type of panel:	<u>Custom panel</u>
Version number:	Version 11
Laboratory:	<u>Centrum Medische Genetica - UZ Brussel VUB</u>
Created:	16 Jul 2019 - 10:51
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Related Diseases

- Adult Krabbe disease
- Chronic visceral acid sphingomyelinase deficiency
- GM1 gangliosidosis type 1
- GM1 gangliosidosis type 2
- GM1 gangliosidosis type 3
- Hurler-Scheie syndrome
- Infantile Krabbe disease
- Infantile neurovisceral acid sphingomyelinase deficiency
- Late-infantile/juvenile Krabbe disease
- Metachromatic leukodystrophy, adult form
- Metachromatic leukodystrophy, juvenile form
- Metachromatic leukodystrophy, late infantile form
- Mucopolidosis type II
- Mucopolidosis type III alpha/beta
- Mucopolidosis type III gamma
- Mucopolysaccharidosis type 2, attenuated form

- Mucopolysaccharidosis type 2, severe form
- Mucopolysaccharidosis type 4A
- Mucopolysaccharidosis type 4B
- Mucopolysaccharidosis type 7
- Niemann-Pick disease type C, adult neurologic onset
- Niemann-Pick disease type C, juvenile neurologic onset
- Niemann-Pick disease type C, late infantile neurologic onset
- Niemann-Pick disease type C, severe early infantile neurologic onset
- Niemann-Pick disease type C, severe perinatal form
- Sandhoff disease, adult form
- Sandhoff disease, infantile form
- Sandhoff disease, juvenile form
- Sanfilippo syndrome type A
- Sanfilippo syndrome type B
- Sialidosis type 1
- Tay-Sachs disease, B variant, adult form
- Tay-Sachs disease, B variant, infantile form
- Tay-Sachs disease, B variant, juvenile form
- Tay-Sachs disease, B1 variant

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ACD</u>	100.00	0	No comment
<u>ADCY5</u>	94.52	0	No comment
<u>AGA</u>	100.00	0	No comment
<u>ARSA</u>	100.00	0	No comment
<u>ASAH1</u>	99.94	0	No comment
<u>CHIT1</u>	100.00	0	No comment
<u>CLN3</u>	100.00	0	No comment

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CLN5</u>	99.94	0	No comment
<u>CLN6</u>	90.35	0	No comment
<u>CLN8</u>	100.00	0	No comment
<u>CTNS</u>	100.00	0	No comment
<u>CTSA</u>	100.00	0	No comment
<u>CTSC</u>	100.00	0	No comment
<u>CTSD</u>	99.45	0	No comment
<u>CTSK</u>	100.00	0	No comment
<u>DGUOK</u>	100.00	0	No comment
<u>FBXL4</u>	100.00	0	No comment
<u>FUCA1</u>	100.00	0	No comment
<u>GAA</u>	100.00	0	No comment
<u>GALC</u>	98.44	0	No comment
<u>GALNS</u>	96.18	0	No comment
<u>GBA1</u>	100.00	0	No comment
<u>GLA</u>	100.00	0	No comment
<u>GLB1</u>	100.00	0	No comment
<u>GM2A</u>	100.00	0	No comment
<u>GNPTAB</u>	100.00	0	No comment
<u>GNPTG</u>	94.69	0	No comment

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<u>GNS</u>	100.00	0	No comment
<u>GUSB</u>	100.00	0	No comment
<u>HEXA</u>	100.00	0	No comment
<u>HEXB</u>	99.82	0	No comment
<u>HGSNAT</u>	93.95	0	No comment
<u>HYAL1</u>	100.00	0	No comment
<u>IDS</u>	100.00	0	No comment
<u>IDUA</u>	88.67	0	No comment
<u>LAMP2</u>	100.00	0	No comment
<u>LIPA</u>	100.00	0	No comment
<u>MAN2B1</u>	100.00	0	No comment
<u>MANBA</u>	99.97	0	No comment
<u>MCOLN1</u>	97.27	0	No comment
<u>MFSD8</u>	100.00	0	No comment
<u>MGME1</u>	100.00	0	No comment
<u>MPV17</u>	100.00	0	No comment
<u>NAGA</u>	100.00	0	No comment
<u>NAGLU</u>	87.38	0	No comment
<u>NEU1</u>	100.00	0	No comment
<u>NPC1</u>	98.40	0	No comment

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<u>NPC2</u>	100.00	0	No comment
<u>OCRL</u>	99.77	0	No comment
<u>POLG</u>	100.00	0	No comment
<u>PPT1</u>	100.00	0	No comment
<u>PSAP</u>	100.00	0	No comment
<u>RRM2B</u>	100.00	0	No comment
<u>SGSH</u>	99.99	0	No comment
<u>SLC17A5</u>	100.00	0	No comment
<u>SLURP1</u>	85.73	0	No comment
<u>SMPD1</u>	100.00	0	No comment
<u>STS</u>	100.00	0	No comment
<u>SUCLA2</u>	100.00	0	No comment
<u>SUCLG1</u>	95.74	0	No comment
<u>SUMF1</u>	100.00	0	No comment
<u>TK2</u>	95.90	0	No comment
<u>TWINK</u>	100.00	0	No comment
<u>TYMP</u>	91.72	0	No comment