MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Lysosomal Storage Disease Gene Panel

The following applies to LSDGP/Lysosomal Storage Disease Gene Panel. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 30 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select non-coding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from February 2021 to the present. This document is intended to highlight additional evaluations for variants of high clinical interest as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, genes or regions covered, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript[a]
AGA	NM_000027.4
ARSA	NM_000487.6
ARSB	NM_000046.5
ASAH1[b]	NM_177924.5
ATP13A2	NM_022089.4
CHIT1	NM_003465.3
CLN3	NM_001042432.1
CLN5	NM_006493.4
CLN6	NM_017882.3
CLN8	NM_018941.4
CTNS	NM_001031681.2
CTSA	NM_000308.3
CTSD	NM_001909.5
CTSF	NM_003793.4
CTSK	NM_000396.4
DNAJC5	NM_025219.3
FUCA1	NM_000147.4
GAA	NM_000152.5
GALC[b]	NM_000153.4
GALNS	NM_000512.5
GBA	NM_000157.4
GFAP	NM_002055.5
GLA[b]	NM_000169.2
GLB1	NM_000404.4
GM2A	NM_000405.5
GNPTAB[b]	NM_024312.5
GNPTG	NM_032520.5
GNS[b]	NM_002076.4

Gene	Reference Transcript[a]
GRN	NM_002087.3
GUSB	NM_000181.4
HEXA	NM_000520.6
HEXB[b]	NM_000521.4
HGSNAT	NM_152419.3
HYAL1	NM_153281.1
IDS	NM_000202.8
IDUA	NM_000203.5
KCTD7	NM_153033.4
LAMP2	NM_002294.3
LIPA	NM_000235.4
MAN2B1	NM_000528.4
MANBA	NM_005908.4
MCOLN1	NM_020533.3
MFSD8	NM_152778.3
NAGA	NM_000262.3
NAGLU	NM_000263.4
NEU1	NM_000434.4
NPC1[b]	NM_000271.5
NPC2	NM_006432.4
PANK2	NM_153638.3
PPT1	NM_000310.3
PSAP	NM_002778.4
SGSH	NM_000199.5
SLC17A5	NM_012434.5
SMPD1	NM_000543.5
SUMF1	NM_182760.4
TPP1	NM_000391.4

[a] Reference transcript numbers may be updated due to transcript re-versioning. Always refer to the final patient report for gene transcript information referenced at the time of testing.

[b] There are regions of this gene that cannot be effectively analyzed for the presence of copy number variants.

To verify if a specific region/exon/variant is covered by this assay, contact a laboratory genetic counselor at 800-533-1710.