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 (CNV) 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 October 2025 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 | Additional Evaluations | Technical Limitations |
|---|---|---|---|
| *AGA* | NM_000027.4 | - | - |
| *ARSA* | NM_000487.6 | - | - |
| *ARSB* | NM_000046.5 | - | - |
| *ASAH1* | NM_177924.5 | - | CNVs may not be detected in exon 4 |
| *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 | chr17:g.78078341T>A (c.-32-13T>A); chr17:g.78090422A>G (c.2190-345A>G) | - |
| *GALC* | NM_000153.4 | - | CNVs may not be detected in exon 6 |
| *GALNS* | NM_000512.5 | - | - |
| *GBA* | NM_000157.4 | - | - |
| *GFAP* | NM_002055.5 | - | - |
| *GLA* | NM_000169.2 | chrX:g.100654735C>T (c.640-801G>A); chrX:g.100654764A>G (c.640-830T>C); chrX:g.100654793G>A (c.640-859C>T) | - |
| *GLB1* | NM_000404.4 | - | - |
| *GM2A* | NM_000405.5 | - | - |

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# Targeted Genes and Methodology Details for Lysosomal Storage Disease Gene Panel (continued)

| Gene | Reference Transcript | Additional Evaluations | Technical Limitations |
|---|---|---|---|
| *GNPTAB* | NM_024312.5 | - | CNVs may not be detected in exon 4 |
| *GNPTG* | NM_032520.5 | - | - |
| *GNS* | NM_002076.4 | - | CNVs may not be detected in exons 2 and 7 |
| *GRN* | NM_002087.3 | - | - |
| *GUSB* | NM_000181.4 | - | - |
| *HEXA* | NM_000520.6 | - | - |
| *HEXB* | NM_000521.4 | - | CNVs may not be detected in exon 4 |
| *HGSNAT* | NM_152419.3 | - | - |
| *HYAL1* | NM_153281.1 | - | - |
| *IDS* | NM_000202.8 | chrX:g.148586802-148586978del (c.-311_-135del177); chrX:g.148578704C>T (c.709-657G>A); chrX:g.148568762 (c.1007-133A>G); IDS/IDSP1 inversion | - |
| *IDUA* | NM_000203.5 | chr4:g.980786T>C (c.-87T>C) | - |
| *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* | NM_000271.5 | chr18:g.21132700C>T (c.1554-1009G>A) | - |
| *NPC2* | NM_006432.4 | - | - |
| *PANK2* | NM_153638.3 | - | - |
| *PPT1* | NM_000310.3 | - | - |
| *PSAP* | NM_002778.4 | chr10:g.73583679G>T (c.777+1915C>A) | - |
| *SGSH* | NM_000199.5 | - | - |
| *SLC17A5* | NM_012434.5 | - | - |
| *SMPD1* | NM_000543.5 | - | - |
| *SUMF1* | NM_182760.4 | - | - |
| *TPP1* | NM_000391.4 | - | - |

| Effective Date | Version | Synopsis of Test Change |
|---|---|---|
| 10/2025 | V2 | Added IDS/IDSP1 inversion detection for the *IDS* gene |