MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Peroxisomal Disorder Gene Panel

The following applies to PDGP / Peroxisomal Disorder 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 to providers 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]
ABCD1	NM_000033.4
ABCD3	NM_002858.4
ACOX1	NM_004035.7
ACOX3	NM_003501.3
AGPS	NM_003659.4
AMACR	NM_014324.6
CAT	NM_001752.4
DNM1L	NM_012062.5
GNPAT[b]	NM_014236.4
HSD17B4	NM_001199291.3
PEX1	NM_000466.3
PEX10	NM_153818.1
PEX11B	NM_003846.3
PEX12	NM_000286.3

Gene	Reference Transcript[a]
PEX13	NM_002618.4
PEX14	NM_004565.3
PEX16	NM_057174.2
PEX19	NM_002857.3
PEX2	NM_000318.3
PEX26	NM_017929.6
PEX3	NM_003630.3
PEX5	NM_001131023.1
PEX6	NM_000287.4
PEX7	NM_000288.4
PHYH	NM_006214.4
SCP2[b]	NM_002979.5
SUGCT[b,c]	NM_024728.2
TRIM37	NM_015294.6

[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.

[c] There are regions of this gene that cannot be effectively amplified for sequencing as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences.

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