MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Phenylalanine Disorders Gene Panel

The following applies to PHEGP/Phenylalanine Disorders 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]
DDC	NM_000790.4
DNAJC12	NM_021800.3
GCH1[b,c]	NM_000161.3
PAH	NM_000277.3
PCBD1	NM_000281.4
PTS	NM_000317.3
QDPR	NM_000320.3
SLC18A2	NM_003054.6
SPR	NM_003124.5
TH	NM_199292.3

[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.