MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Expanded Pancreatitis Gene Panel

The following applies to PANGP / Expanded Pancreatitis Gene Panel. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 10 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select noncoding 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 July 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
CASR	NM_000388.4	-	-
CEL	NM_001807.5	-	Sequence and CNV analysis in exons 1, 8, 9, 11 are not performed
CFTR	NM_000492.4	Poly T tract; TG repeat region for 5T alleles only; deletion/duplication analysis; chr7:g. 117179040AGAAT>A (c.870-1113_870-1110del); chr7:g.117199500G>A (c.1393-18G>A); chr7:g.117227784T>A (c.1585-9T>A); chr7:g.117227785G>A (c.1585-8G>A); chr7:g.117218381A>G (c.1585-9412A>G); chr7:g.117229521A>G (c.1680-886A>G); chr7:g.117229524A>G (c.1680-883A>G); chr7:g.117229530G>T (c.1680-877G>T); chr7:g.117246713T>G (c.2909-15T>G); chr7:g.117250260A>T (c.2989-313A>T); chr7:g.117251609A>G (c.3140-26A>G); chr7:g.117251619T>A (c.3140-16T>A); chr7:g.117266272C>G (c.3469-1304C>G); chr7:g.117267864A>G (c.3717+40A>G); chr7:g.117280015C>T (c.3718-2477C>T); chr7:g.117288374A>G (c.3874-4522A>G)	CNV analysis in exon 13 is not performed
CLDN2	NM_020384.4	-	-
CPA1	NM_001868.4	-	-
CTRC	NM_007272.3	-	-
PRSS1	NM_002769.5	-	-
SPINK1	NM_003122.4	chr5:g.147211193G>A (c.-53C>T)	-
TRPV6	NM_018646.6	-	-