MAYO CLINIC

1-800-533-1710

Expanded Pancreatitis Gene Panel

PANGP

PATIENT NAME				ORDER NUMBER
PANGP, ABNORMAL1				**P409000293**
PATIENT ID X100498720	DATE OF BIRTH 10/30/1990	AGE 34 Y	SEX Male	REQUESTED BY PROVIDER UNKNOWN
COLLECTED 5/9/2025, 8:00 AM	RECEIVED 5/12/2025, 3:04 PM	REPORTED 5/13/2025, 4:04 PM		
The collected, received, and reported dates and times on the report are in the time zone of the performing location. 7028846 MCL RochesterCampus Rochester MN 55901				CLIENT ORDER NUMBER X100498720 CLIENT MRN C7028846-0006407

TEST DESCRIPTION

Evaluation of 9 genes associated with an increased risk for pancreatitis

SPECIMEN

WB Whole Blood

RESULT SUMMARY

Pathogenic Variant Detected

RESULT

Gene (Transcript)	Variant	Zygosity	Classification
TRPV6 (NM_018646.6)	c.1168C>T p.Arg390Cys (p.R390C) Chr7(GRCh37):g.142573295G>A	heterozygous	PATHOGENIC

The following PATHOGENIC variant was detected:
TRPV6 (NM_018646.6), chr7(GRCh37):g.142573295G>A, c.1168C>T, p.Arg390Cys (p.R390C), heterozygous

No additional reportable variants were detected within all other tested genes. See the Genes Analyzed section for a complete list of genes evaluated by this assay.

CFTR Intron 9 poly-T alleles: 7T/9T (c.1210-12T[7]/c.1210-12T[9])

INTERPRETATION

TRPV6 c.1168C>T (p.Arg390Cys), PATHOGENIC
The heterozygous c.1168C>T (p.Arg390Cys) missense variant in the TRPV6 gene (MIM:606680) is an established pathogenic variant. Pathogenic variants in the TRPV6 gene have been associated with early-onset nonalcoholic chronic pancreatitis and autosomal recessive transient neonatal hyperparathyroidism (1). In some cases, TRPV6-related conditions do not follow a classic dominant or recessive inheritance pattern but are caused by a combination of multiple genetic variants in one or more genes, as well as environmental triggers. Therefore, the identified TRPV6 variant may predispose this individual to pancreatitis, but additional genetic or environmental factors may also contribute to disease presentation and/or penetrance. Clinical correlation is recommended.

Pancreatitis is a multifactorial condition that may involve environmental and/or genetic factors. This result should be interpreted in the context of clinical findings, family history, and other laboratory testing.

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CFTR Intron 9 poly T tract alleles (5T-11, 12 or 13TG/7T/9T) are polymorphic variants. The 5T-11TG, 7T and 9T alleles are considered benign. The 5T-12TG and 5T-13TG alleles are considered clinically significant only when detected in the homozygous state or with a pathogenic CFTR variant.

Consultation with a genetics professional is recommended for interpretation of this result and to determine whether reproductive risk assessment and familial testing may be of benefit to this family.

FOLLOW-UP OPTION(S)
Familial Testing:
If relevant, genetic testing for family members is available by ordering FMTT / Familial Mutation, Targeted Testing for the specific variant(s) detected. Please contact the laboratory at 1-800-533-1710 or the online test catalog at www.mayocliniclabs.com for information about FMTT.

ALTERNATE NOMENCLATURE
The intron 9 poly T alleles were previously known as intron 8 poly T alleles, using historic intron/exon numbering.

REFERENCES:
1) GeneReviews: Pancreatitis Overview (https://www.ncbi.nlm.nih.gov/books/NBK190101/) (PMID: 24624459)

METHOD

Next generation sequencing (NGS) and/or Sanger sequencing was performed to test for the presence of variants in coding regions and intron/exon boundaries of the genes analyzed, as well as some other regions that have known pathogenic variants. NGS and/or a polymerase chain reaction (PCR)-based quantitative method was performed to test for the presence of deletions and duplications in the genes analyzed. The human genome reference GRCh37/hg19 build was used for sequence read alignment. At least 99% of the bases are covered at a read depth over 30X. Sensitivity is estimated at above 99% for single nucleotide variants, above 94% for indels less than 40 base pairs (bp), above 95% for deletions up to 75 bp and insertions up to 47 bp. See the Genes Analyzed field for a list of genes tested.

There may be regions of genes that cannot be effectively evaluated for sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences. Confirmation of select reportable variants was performed by alternate methodologies based on internal laboratory criteria. See www.mayocliniclabs.com (TEST ID PANGP) for details regarding genes with regions not routinely covered.

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GENES ANALYZED

CASR, CEL, CFTR, CLDN2, CPA1, CTRC, PRSS1, SPINK1 and TRPV6

DISCLAIMER

Clinical Correlations

The gene(s) analyzed by this test may have more than one associated phenotype or inheritance pattern. Additionally, the genetic variants detected may have reduced penetrance, variable expressivity, or different classifications based on disease state. Classification and reporting of the variants detected is based on the intended disease state of the test ordered. For more information regarding the phenotypic spectrum which may be involved for a specific gene, see OMIM (www.ncbi.nlm.nih.gov/omim) or GeneReviews (www.genereviews.org).

An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

If testing was performed because of a clinically significant family history it is often useful to first test an affected family member. Detection of a reportable variant(s) in an affected family member would allow for more informative testing of at risk individuals.

To discuss the availability of further testing options or for assistance in the interpretation of these results, Mayo Clinic Laboratory genetic counselors can be contacted at 1-800-533-1710.

Technical Limitations

Next generation sequencing may not detect all types of genomic variants. In rare cases, false negative or false positive results may occur. The depth of coverage may be variable for some target regions, but assay performance below the minimum acceptable criteria or for failed regions will be noted. The clinical phenotype observed in this individual and/or family may be due to genetic variants not targeted by this test. Given these limitations, negative results do not rule out the diagnosis of a genetic disorder. If a specific clinical disorder is suspected, evaluation by alternative methods can be considered.

There may be regions of genes that cannot be effectively evaluated for sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences. Confirmation of select reportable variants was performed by alternate methodologies based on internal laboratory criteria.

Additionally, low level mosaic variants may not be detected.

This test is not designed to differentiate between somatic and germline variants. If there is a possibility that any detected variant is somatic, additional testing may be necessary to clarify the significance of results.

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Genes may be added or removed based on updated clinical relevance. Please refer to the Targeted Genes and Methodology Details for the Expanded Pancreatitis Gene Panel, Varies in the Special Instructions section of the Test Catalog for the most up to date list of genes included in this test.

This test is validated to detect 95% of deletions up to 75 base pairs (bp) and insertions up to 47 bp. Deletions-insertions (delins) of 40 or more bp, including mobile element insertions, may be less reliably detected than smaller delins.

This analysis targets single and multi-exon deletions/duplications; however, in some instances, single exon resolution cannot be achieved due to isolated reduction in sequence coverage or inherent genomic complexity. Balanced structural rearrangements (such as translocations and inversions) may not be detected.

If the patient has had an allogeneic hematopoietic stem cell transplant or a recent blood transfusion, results may be inaccurate due to the presence of donor DNA. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Reclassification of Variants Policy
See www.mayocliniclabs.com (TEST ID PANGP) for information regarding the laboratory's policy for reclassification of variants.

Variant Evaluation
Variant curation is performed using published ACMG-AMP recommendations as a guideline. Other gene-specific guidelines may also be considered. Variants classified as benign or likely benign are not reported.

Results from in silico evaluation tools may change over time and should be interpreted with caution and professional clinical judgment.

TEST CLASSIFICATION
This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

RELEASED BY
LINDA HASADSRI

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