Helix Factor V Leiden and Factor II (Prothrombin) Variant Test

Item	Description
Test Name	Helix Factor V Leiden and Factor II (Prothrombin) Variant Test
Test Type	Target Analysis
Catalog Number	FVFT1
Procedure Code	H00123-6 (Helix)
Test Description	Helix Factor V Leiden and Factor II (Prothrombin) Variant Test is a diagnostic test that analyzes one variant in *F5* and one variant in *F2* associated with factor V Leiden-related thrombophilia or prothrombin-related thrombophilia.
Genes Tested	F5: NM_000130.5:c.1601G>A (p.Arg534Gln) and F2: NM_000506.5:c.*97G>A
Genetics Information	This test utilizes next-generation sequencing to detect single nucleotide variants.
Indications For Testing	A relevant personal and/or family history suggestive of factor V Leiden-related thrombophilia or prothrombin-related thrombophilia.
Clinical Descriptions	This panel includes targeted variants that have an established association with factor V Leiden-related thrombophilia or prothrombin-related thrombophilia. The targeted variants were specifically selected for their established association with these conditions. Detection of these variants may facilitate increased early-detection and prevention, as well as helping to identify at-risk family members who can pursue genetic testing and preventive measures.
Conditions	Factor V Leiden Thrombophilia (F5) Prothrombin thrombophilia (F2)
Interpretation	All detected variants are evaluated according to American College of Medical Genetics and Genomics recommendations. Variants are classified based on known, predicted, or possible pathogenicity; however, this test only reports pathogenic and likely pathogenic variants along with interpretive comments detailing the evidence applied towards classification. Variants of uncertain significance are not reported.
Reclassification of Variants	Helix does not systematically review their variant database looking for classification changes. Helix will review the classification of previously reported variants upon request of the ordering physician/provider. Ordering physicians/providers may contact Helix Customer Support or their Dedicated Advisor and request a review of the variant classification to be performed. At the discretion of the laboratory director, the frequency of reclassification requests may be limited to once per year, no earlier than 12 months after initial variant interpretation has been performed.
Variant Evaluation	Variant classification is performed using the guidelines set forth by the American College of Medical Genetics and Genomics and the Association for Molecular Pathology, with modifications as suggested by domain specific Expert Panels of the Clinical genome Resource (ClinGen) when available. Variant pathogenicity is categorized as benign, likely benign, variant of uncertain significance (VUS), likely pathogenic, or pathogenic.
Turnaround Time	7 to 24 days
Available in NY State	No

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Item	Description
Test Classification	This test was developed, and its performance characteristics determined, by Helix, Inc. in a manner consistent with CLIA requirements. This test has not been cleared or approved by the US Food and Drug Administration.
Performing Laboratory Information	CLIA Laboratory Number: 05D2117342 Laboratory Hours of Operation: Monday-Saturday (7AM-10:30PM PST) Address: 10170 Sorrento Valley Road, Suite 100, San Diego, CA 92121 Helix Customer Service: (844) 211-2070 Email: support@helix.com
Regulatory Information	CLIA Complexity: High Test Classification: Non-Waived/ Laboratory Developed Test
CLIA Category	Chemistry / Routine Chemistry

Methods & Limitations for Helix Factor V Leiden and Factor II (Prothrombin) Variant Test

Extracted DNA is enriched for targeted regions and then sequenced using the Helix Exome+ (R) assay on an Illumina DNA sequencing system. Data is then aligned to a modified version of GRCh38. Small variant calling is completed using a customized version of Sentieon's DNAseq software, augmented by a proprietary small variant caller for difficult variants. Interpretation is based upon guidelines published by the American College of Medical Genetics and Genomics (ACMG) and the Association for Molecular Pathology (AMP) or their modification by ClinGen Variant Curation Expert Panels when available. Interpretation is limited to the variant indicated on the report. Helix variant classifications include pathogenic, likely pathogenic, variant of uncertain significance (VUS), likely benign, and benign. Only variants classified as pathogenic and likely pathogenic are included in the report. All reported variants are confirmed through secondary manual inspection of DNA sequence data or orthogonal testing. Risk estimations and management guidelines included in this report are based on analysis of primary literature and recommendations of applicable professional societies, and should be regarded as approximations.

Based on validation studies, this assay delivers > 99% sensitivity and specificity for single nucleotide variants. The results of a genetic test may be influenced by various factors, including bone marrow transplantation, blood transfusions, or in rare cases, hematolymphoid neoplasms.

Gene Specific Notes:
*F2: analysis is limited to NM_000506.5:c.*97G>A.*
F5: analysis is limited to NM_000130.5:c.1601G>A (p.Arg534Gln).

Disclaimer:
This test was developed and validated by Helix, Inc. This test has not been cleared or approved by the United States Food and Drug Administration (FDA). The Helix laboratory is accredited by the College of American Pathologists (CAP) and certified under the Clinical Laboratory Improvement Amendments (CLIA #: 05D2117342) to perform high-complexity clinical tests. This test is used for clinical purposes. It should not be regarded as investigational or for research.

Targeted Genes & Methodology for Helix Factor V Leiden and Factor II (Prothrombin) Variant Test

The following applies to the Helix Factor V Leiden Variant and Factor II (Prothrombin) Variant Test. Testing is performed by next-generation sequencing to evaluate for the presence of a single variant. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from November 2024 to the present. For questions regarding genes, reference transcripts, or specific regions covered, contact Helix Customer Service at (844) 211-2070.

Genomic Build: GRCh38
Catalog Number: FVFT1

Gene	Transcript	Additional Evaluations
F2	NM_000506.5	–
F5	NM_000130.5	–