

Helix Hereditary Hemochromatosis Targeted Test

Patient Name: Jane Doe	Specimen Type: WHOLE BLOOD	Provider Name: Client Client	Report Date: 06-15-2025
Date of Birth: 01-01-1990	Patient ID: 123456	Collection Date: 06-10-2025	
Sex Assigned at Birth: Female	Helix ID: Test12345	Order Date: 06-15-2025	

Note: This report is intended for use by a medical professional. Please discuss any adjustments to your medication with your treating provider.

Results NEGATIVE

No pathogenic or likely pathogenic variants were detected in the genes analyzed by this test.

Genetic test results should be interpreted in the context of an individual's personal medical and family history. Alteration to medical management is NOT recommended based solely on this result. Clinical correlation is advised.

Test Description

Helix Hereditary Hemochromatosis Targeted Test is a diagnostic test that analyzes three variants in HFE associated with hereditary hemochromatosis. The two variants C282Y (c.845G>A) and H63D (c.187C>G) are always reported, whereas the S65C (c.193A>T) variant will be reported only when it is observed as part of the C282Y/S65C genotype.

Genes Tested

HFE: NM_000410.4:c.845G>A (p.Cys282Tyr), c.187C>G (p.His63Asp), and c.193A>T (p.Ser65Cys)

Methods & Limitations

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: HFE: analysis is limited to NM_000410.4:c.845G>A (p.Cys282Tyr), c.187C>G (p.His63Asp), and c.193A>T (p.Ser65Cys).

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.

Report Signed By

Matt Ferber, Ph.D. FACMG

Helix's Sequence Once, Query Often® Model

When your provider orders a genetic test through Helix, we use our proprietary Sequence Once, Query Often® model to perform whole exome sequencing and analyze the specific genes related to the test. Helix securely stores your whole exome for future clinical use. With your permission, this allows your health care providers to order future medically necessary genetic tests from Helix without needing another sample. Instead, these tests are conducted through digital analysis of your stored genetic information.

To learn more about how Helix protects the privacy and security of your genetic information and learn more about your rights, please visit <https://www.helix.com/privacy-and-policy-highlights>.