Helix Pharmacogenomics (PGx) Proton Pump Inhibitors CYP2C19 Test

Item	Description
Test Name	Helix Pharmacogenomics (PGx) Proton Pump Inhibitors CYP2C19 Test
Test Type	Pharmacogenomics
Catalog Number	PPPI1
Procedure Code	H00624-1
Test Description	This test evaluates a patient's metabolizer status for *CYP2C19*, which can aid in appropriate prescription and dosing to reduce risk of side effects and treatment failure with proton pump inhibitors.
Genes Tested	*CYP2C19*
Genetics Information	This test utilizes next-generation sequencing to determine star alleles and metabolizer status for *CYP2C19* and is used to determine drug considerations for omeprazole, lansoprazole, pantoprazole, and dexlansoprazole.
Indications For Testing	Patients for whom omeprazole, lansoprazole, pantoprazole, and dexlansoprazole are being considered, have been ineffective or caused side effects.
Clinical Descriptions	Proton pump inhibitors are prescribed to patients with gastroesophageal reflux disease (GERD), peptic ulcer disease and other conditions related to dysfunctional gastric acid secretion. Certain *CYP2C19* metabolizer types may reduce efficacy of PPIs, while others may lead to increased risk for adverse events.
Disease States	GERD, stomach or duodenal ulcers, H. pylori infection, or other conditions causing stomach acid irritation.
Interpretation	All detected variants are evaluated according to the Clinical Pharmacogenetics Implementation Consortium (CPIC). Variants are classified based on known, predicted, or possible impact on drug metabolism.
Reclassification Of Variants	Helix reviews relevant pharmacogenomic guidelines as published by the FDA, CPIC and PharmGKB at least annually. In the event of updated guidance, past results will be assessed. In the event of report revisions, updated results are communicated to providers prior to results being uploaded to the EHR, and patients are notified through the EHR patient portal.
Variant Evaluation	Variant classification is performed using data provided by CPIC whenever available. When CPIC data is not available, a thorough literature search is performed to evaluate whether specific alleles have known and established impact on drug metabolism. Recommendations and interpretation for dosage and prescription are based on guidelines set forth by CPIC, the Food and Drug Administration (FDA), and the Pharmacogenomics Knowledgebase (PharmGKB). Variants are classified as having impact on drug metabolism with the following tiers: poor metabolizer, intermediate metabolizer, normal metabolizer, and ultrarapid metabolizer. These classifications are based on the combination of alleles found in the given individual and data set forth by CPIC and the other entities mentioned above. Variants of unknown significance on drug metabolism are not reported.
Turnaround Time - Standard	Typically 7 to 10 days

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Item	Description
Turnaround Time - Requery (SOQO®)	Typically ≤ 5 minutes
Available In NY State	Yes
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

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Methods & Limitations for Helix Pharmacogenomics (PGx) Proton Pump Inhibitors CYP2C19 Test

Data were generated from extracted DNA using the validated Helix Exome+ assay by the Helix clinical laboratory. The Exome+ assay is based on target enrichment followed by next generation sequencing using paired end reads on an Illumina DNA sequencing system. Star alleles were determined using a proprietary algorithm which performs variant calling and then determines star allele solutions based on a combination of defining SNPs and exon-level copy number.

Metabolizer status was determined based on star allele solutions according to CPIC guidelines, with the following exceptions: (1) metabolizer status was set as Indeterminate if a novel nonsense or truncating novel mutation was observed within the gene, (2) metabolizer status was set as Indeterminate if the combination of defining SNPs and copy number suggested a novel star allele solution, and (3) if more than two copies of a gene were detected then metabolizer status was set as Indeterminate. Drug/gene considerations were limited to guidelines published by FDA, CPIC, or PharmGKB.

Phasing could not be performed for genotypes, and therefore in some cases the star allele solution could not be disambiguated between two or more equally likely possibilities. In these cases, if the metabolizer status was the same regardless of possible star allele solutions, the more common star allele solution was provided along with the metabolizer status. If the metabolizer status was different for the equally-likely star allele solutions, the star alleles were reported as Unknown and the metabolizer status was considered Indeterminate.

All samples were sequenced and interpreted in Helix's CLIA-certified (#05D2117342) and CAP-accredited (#9382893) laboratory in San Diego, California. These tests have not been cleared or approved by the U.S. Food and Drug Administration.

The reportable range includes the following results:
CYP2C19: *1-*19, *22-*26, *28-*39.

Results are based on:
Dexlansoprazole, *CYP2C19* (PGKB 1A); Lansoprazole, *CYP2C19* (PGKB 1A; CPIC A); Omeprazole, *CYP2C19* (PGKB 1A; CPIC A); Pantoprazole, *CYP2C19* (FDA Section 1; CPIC A).

Disclaimer:
The interpretations and drug considerations provided by Helix are intended solely for use by a medical professional and do not constitute medical advice by Helix. All treatment decisions and diagnoses remain the full responsibility of the treating provider. Results included in this report are based on the guidelines published by the FDA and CPIC, and do not account for other factors that may impact drug response, such as environment, medical conditions, drug-drug interactions, or additional genetic variants. Helix is not responsible or liable for any errors, omissions, or ambiguities in the interpretation or use of the results of this report. Administration of any medication listed in this report requires careful therapeutic monitoring regardless of the drug considerations outlined in this report. All dates and times displayed are Pacific Time and may vary from the dates and times for Collection, Order and Report for the providers/patients.

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Targeted Genes & Methodology for Helix Pharmacogenomics (PGx) Proton Pump Inhibitors CYP2C19 Test

The following applies to the Helix Pharmacogenomics (PGx) Proton Pump Inhibitors CYP2C19 Test. Next-generation sequencing is performed to test for the presence of star allele solutions in the genes analyzed, according to the reportable range listed.

This list is current from January 2025 to the present. For questions regarding genes, reference transcripts, or specific regions covered, contact Helix Customer Service at (844) 211-2070.

Catalog Number: PPPI1

Gene	Reportable Range
CYP2C19	*1- *19, *22- *26, *28- *39

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