Helix Pharmacogenomics (PGx) Level A Panel

Item	Description
Test Name	Helix Pharmacogenomics (PGx) Level A Panel
Test Type	Pharmacogenomics
Catalog Number	PLVA1
Procedure Code	H01225-1 (Helix)
Test Description	This test evaluates a patient's metabolizer status for 15 genes, which can aid in appropriate medication selection and dosing to reduce risk of side effects and treatment failure for cancer, cardiovascular disease, pain, mental health and other conditions. All results in this test are based on CPIC Level A guidance, but this test is not comprehensive for all CPIC Level A guidance.
Genes Tested	*ABCG2, CYP2C9, CYP2C19, CYP2D6, CYP3A5, CYP4F2, DPYD, HLA-B, IFNL3, IFNL4, MT-RNR1, NUDT15, SLCO1B1, TPMT, VKORC1* Genes with CPIC Level A guidance that are not included in this test include: CACNA1S, CFTR, CYP2B6, HLA-A*31:01, HLA-B*58:01, G6PD, RYR1, or UGT1A1.
Genetics Information	This test utilizes next-generation sequencing to determine star alleles or genotypes and metabolizer status or activity for *ABCG2, CYP2C9, CYP2C19, CYP2D6, CYP3A5, CYP4F2, DPYD, HLA-B, IFNL3, IFNL4, MT-RNR1, NUDT15, SLCO1B1, TPMT, VKORC1*
Indications For Testing	Patients for whom medication from the following list are being considered, have been ineffective, or caused side effects: Abacavir, Amikacin, Amitriptyline, Atomoxetine, Atorvastatin, Azathioprine, Capecitabine, Carbamazepine, Celecoxib, Citalopram, Clopidogrel, Codeine, Escitalopram, Fluorouracil, Flurbiprofen, Fluvastatin, Gentamicin, Ibuprofen, Kanamycin, Lansoprazole, Lornoxicam, Lovastatin, Meloxicam, Mercaptopurine, Nortriptyline, Omeprazole, Ondansetron, Oxcarbazepine, Pantoprazole, Paroxetine, Peginterferon alfa-2a, Peginterferon alfa-2b, Piroxicam, Pitavastatin, Plazomicin, Pravastatin, Rosuvastatin, Sertraline, Simvastatin, Streptomycin, Tacrolimus, Tamoxifen, Tenoxicam, Thioguanine, Tobramycin, Tramadol, Tropisetron, Voriconazole, Vortioxetine, and Warfarin
Clinical Descriptions	Many medications are impacted by specific variations in known genes. These genetic variations can contribute to medication efficacy and risk for side effects. Pharmacogenomic testing to guide prescribing, paired with clinical monitoring can impact outcomes for patients by reducing the time to therapeutic effect and reducing the risk of side effects.
Disease States	Cardiovascular, neurovascular, mental health, gastrointestinal, cancer, pain and many other conditions.
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 and all reported drug-gene interactions are assigned to CPIC Level A in this test.
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.

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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 and meet Level A priority for this test. Variant classifications are based on the combination of alleles found in the given individual and data set forth by CPIC. Variants of unknown significance on drug metabolism are not reported.
Turnaround Time - Standard	Typically 7 to 10 days
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

Methods & Limitations for Helix Pharmacogenomics (PGx) Level A Panel

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 CPIC.

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:
ABCG2: rs2231142; *CYP2C9*: *1-*61; *CYP2C19*: *1-*19, *22-*26, *28-*39; *CYP2D6*: *1-*15, *4N, *17-*65, *68-*75, *81, *83-*114; *CYP3A5*: *1, *3, *6-*9; *CYP4F2*: *1,*3; *DPYD*: *1, *2A, *3, *4, *6-*8, *9B, *10-*13, HapB3, rs150036960, rs72549310, rs80081766, rs150385342, rs141462178, rs200562975, rs2297595, rs139834141, rs6670886, rs115232898, rs72549308, rs72549307, rs45589337, rs146356975, rs150437414, rs145112791, rs201018345, rs183385770, rs143154602, rs72549305, rs75017182, rs140602333, rs143815742, rs61622928, rs200064537, rs764666241, rs142512579, rs186169810, rs72975710, rs144395748, rs57918000, rs199549923, rs72549304, rs111858276, rs138391898, rs148994843, rs190951787, rs142619737, rs201615754, rs59086055, rs138616379, rs145773863, rs147601618, rs17376848, rs3918289, rs55971861, rs138545885, rs137999090, rs145548112, rs146529561, rs60511679, rs112766203, rs56005131, rs199634007, rs200687447, rs60139309, rs201035051, rs55674432, rs147545709, rs67376798, rs141044036, rs145529148, rs72547602, rs72547601, rs202144771, rs139459586, rs140114515, rs148799944, rs114096998; *HLA-B**15:02, *57:01; I*FNL3 / IFNL4*: rs12979860; *MT-RNR1*: rs267606617, rs267606618, rs267606619; *NUDT15*: *1-*20; *SLCO1B1*: *1-*16, *19, *20, *23-*34, *36-*44, *47-*49; *TPMT*: *1, *2, *3A, *3B, *3C, *4-*44; *VKORC1*: rs9923231. Sensitivity may be reduced for the *CYP2D6**13 allele.

Results are based on CPIC Level A guidelines for:
Abacavir, *HLA-B**57:01; Amikacin, *MT-RNR1*; Amitriptyline, *CYP2C19*, *CYP2D6*; Atomoxetine, *CYP2D6*; Atorvastatin, *SLCO1B1*; Azathioprine, *NUDT15*, *TPMT*; Capecitabine, *DPYD*; Carbamazepine, *HLA-B**15:02; Celecoxib, *CYP2C9*; Citalopram, *CYP2C19*; Clopidogrel, *CYP2C19*; Codeine, *CYP2D6*; Escitalopram, *CYP2C19*; Fluorouracil, *DPYD*; Flurbiprofen, *CYP2C9*; Fluvastatin, *CYP2C9*, *SLCO1B1*; Gentamicin, *MT-RNR1*; Ibuprofen, *CYP2C9*; Kanamycin, *MT-RNR1*; Lansoprazole, *CYP2C19*; Lornoxicam, *CYP2C9*; Lovastatin, *SLCO1B1*; Meloxicam, *CYP2C9*; Mercaptopurine, *NUDT15*, *TPMT*; Nortriptyline, *CYP2D6*; Omeprazole, *CYP2C19*; Ondansetron, *CYP2D6*; Oxcarbazepine, *HLA-B**15:02; Pantoprazole, *CYP2C19*; Paroxetine, *CYP2D6*; Peginterferon alfa-2a, *IFNL3 / IFNL4*; Peginterferon alfa-2b, IFNL3 / IFNL4; Piroxicam, *CYP2C9*; Pitavastatin, *SLCO1B1*; Plazomicin, *MT-RNR1*; Pravastatin, *SLCO1B1*; Rosuvastatin, *ABCG2*, *SLCO1B1*; Sertraline, *CYP2C19*; Simvastatin, *SLCO1B1*; Streptomycin, *MT-RNR1*; Tacrolimus, *CYP3A5*; Tamoxifen, *CYP2D6*; Tenoxicam, *CYP2C9*; Thioguanine, NUDT15, TPMT; Tobramycin, *MT-RNR1*; Tramadol, *CYP2D6*; Tropisetron, *CYP2D6*; Voriconazole, *CYP2C19*; Vortioxetine, *CYP2D6*; Warfarin, *CYP2C9*, *CYP4F2*, *VKORC1*.

Methods & Limitations for Helix Pharmacogenomics (PGx) Level A Panel

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.

Targeted Genes & Methodology for Helix Pharmacogenomics (PGx) Level A Panel

The following applies to the Helix Pharmacogenomics (PGx) CPIC A Panel. 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 September 2025 to the present. For questions regarding genes, reference transcripts, or specific regions covered, contact Helix Customer Service at (844) 211-2070.

Catalog Number: PLVA1

Gene	Reportable Range
ABCG2	rs2231142
CYP2C9	*1-*61
CYP2C19	*1-*19, *22-*26, *28-*39
CYP2D6	*1-*15, *4N, *17-*65, *68-*75, *81, *83-*114
CYP3A5	*1, *3, *6-*9
CYP4F2	*1,*3
DPYD	*1, *2A, *3, *4, *6-*8, *9B, *10-*13, HapB3, rs150036960, rs72549310, rs80081766, rs150385342, rs141462178, rs200562975, rs2297595, rs139834141, rs6670886, rs115232898, rs72549308, rs72549307, rs45589337, rs146356975, rs150437414, rs145112791, rs201018345, rs183385770, rs143154602, rs72549305, rs75017182, rs140602333, rs143815742, rs61622928, rs200064537, rs764666241, rs142512579, rs186169810, rs72975710, rs144395748, rs57918000, rs199549923, rs72549304, rs111858276, rs138391898, rs148994843, rs190951787, rs142619737, rs201615754, rs59086055, rs138616379, rs145773863, rs147601618, rs17376848, rs3918289, rs55971861, rs138545885, rs137999090, rs145548112, rs146529561, rs60511679, rs112766203, rs56005131, rs199634007, rs200687447, rs60139309, rs201035051, rs55674432, rs147545709, rs67376798, rs141044036, rs145529148, rs72547602, rs72547601, rs202144771, rs139459586, rs140114515, rs148799944, rs114096998
HLA-B	*15:02, *57:01
IFNL3 / IFNL4	rs12979860
MT-RNR1	rs267606617, rs267606618, rs267606619
NUDT15	*1-*20
SLCO1B1	*1-*16, *19, *20, *23-*34, *36-*44, *47-*49
TPMT	*1, *2, *3A, *3B, *3C, *4-*44
VKORC1	rs9923231