Test Definition: PGXQP

Focused Pharmacogenomics Panel, Varies

Overview

Useful For

Preemptive or reactive genotyping of patients for pharmacogenomic purposes

Providing an assessment for genes with strong drug-gene associations

Assisting in the management of patients with complex medication regimens

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
CULFB	Fibroblast Culture for Genetic Test	Yes	No
CULAF	Amniotic Fluid Culture/Genetic Test	Yes	No
2D61Z	CYP2D6 Full Gene Sequence	No, (Bill Only)	No
2D62Z	CYP2D6 GEN CYP2D6-2D7 Hybrid	No, (Bill Only)	No
2D63Z	CYP2D6 GEN CYP2D7-2D6 Hybrid	No, (Bill Only)	No
2D64Z	CYP2D6 Nonduplicated Gene	No, (Bill Only)	No
2D65Z	CYP2D6 5' Gene DUP/MLT	No, (Bill Only)	No
2D66Z	CYP2D6 3' Gene DUP/MLT	No, (Bill Only)	No
_STR1	Comp Analysis using STR (Bill only)	No, (Bill only)	No
_STR2	Add'l comp analysis w/STR (Bill Only)	No, (Bill only)	No
MATCC	Maternal Cell Contamination, B	Yes	No

Genetics Test Information

This test includes targeted testing to evaluate the following genes:
CYP1A2, CYP2C9, CYP2C19, CYP2D6, CYP3A4, CYP3A5, SLCO1B1, VKORC1, CYP4F2, and rs12777823.

CYP2D6 testing is done in 2 tiers when needed. Tier 1 uses a polymerase chain reaction (PCR)-based 5'-nuclease assay to determine the variants present. All samples also have copy number determined by PCR-based 5'-nuclease assay. Testing in tier 1 allows for the detection of all common *CYP2D6* variants (eg, **2, *3, *4, *5, *6, *7, *8, *9, *10, *17, *29, *35, *41, *59*) and rarer alleles such as **11*, **12, *14*, **15,* and *114. Duplications and multiplications of alleles are also identified. Unitary and tandem *CYP2D7-2D6* (**13*) alleles and *CYP2D6-2D7* (eg, **4N*, **36*, and **68*) alleles can also be

detected. Tier 2 testing involves sequencing using fluorescent dye-terminator chemistry and is only done if an ambiguous phenotype results from tier 1 testing. Approximately 3% of samples require tier 2 testing. Reflex sequencing is limited to the minimum number of reactions that are required to define the patient's genotype.

Testing Algorithm

If a specimen requires follow-up for *CYP2D6*, then reflex testing will be performed as appropriate at an additional charge.

For cord blood specimens, maternal cell contamination testing may be performed at an additional charge.

For more information see *CYP2D6* Comprehensive Cascade Testing Algorithm.

Special Instructions

- Informed Consent for Genetic Testing
- CYP2D6 Comprehensive Cascade Testing Algorithm
- Pharmacogenomic Association Tables
- Informed Consent for Genetic Testing (Spanish)

Method Name

Real Time Polymerase Chain Reaction (RT-PCR) with Allelic Discrimination Analysis/PCR followed by DNA Sequencing, when appropriate

NY State Available

Yes

Specimen

Specimen Type

Varies

Specimen Required

Patient Preparation: A previous hematopoietic stem cell transplant from an allogenic donor will interfere with testing. For information about testing patients who have received a hematopoietic stem cell transplant, call 800-533-1710.

Submit only 1 of the following specimens:

Specimen Type: Whole blood
Container/Tube: Lavender top (EDTA)
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. **Do not aliquot**.
3. Whole blood collected postnatal from an umbilical cord is also acceptable. See Additional Information.

Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated 4 days/Frozen 4 days

Additional Information:

1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed.
2. To ensure minimum volume and concentration of DNA is met, the requested volume must be submitted. Testing may be canceled if DNA requirements are inadequate.
3. For postnatal umbilical cord whole blood specimens, maternal cell contamination studies are recommended to ensure test results reflect that of the patient tested. A maternal blood specimen is required to complete maternal cell contamination studies. Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on both the cord blood and maternal blood specimens under separate order numbers.

Specimen Type: Saliva
Patient Preparation: Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.
Supplies:
DNA Saliva Kit High Yield (T1007)
Saliva Swab Collection Kit (T786)
Container/Tube:
Preferred: High-yield DNA saliva kit
Acceptable: Saliva swab
Specimen Volume: 1 Tube if using T1007 or 2 swabs if using T786
Collection Instructions: Collect and send specimen per kit instructions.
Specimen Stability Information: Ambient (preferred) 30 days/Refrigerated 30 days
Additional Information: Saliva specimens are acceptable but not recommended. Due to lower quantity/quality of DNA yielded from saliva, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing.

Specimen Type: Extracted DNA
Container/Tube:
Preferred: Screw Cap Micro Tube, 2 mL with skirted conical base
Acceptable: Matrix tube, 1mL
Collection Instructions:
1. The preferred volume is at least 100 mcL at a concentration of 75 ng/mcL.
2. Include concentration and volume on tube.
Specimen Stability Information: Frozen (preferred) 1 year/Ambient/Refrigerated
Additional Information: DNA must be extracted in a CLIA-certified laboratory or equivalent and must be extracted from a specimen type listed as acceptable for this test (including applicable anticoagulants). Our laboratory has experience with Chemagic, Puregene, Autopure, MagnaPure, and EZ1 extraction platforms and cannot guarantee that all extraction methods are compatible with this test. If testing fails, one repeat will be attempted, and if unsuccessful, the test will be reported as failed and a charge will be applied. If applicable, specific gene regions that were unable to be interrogated due to DNA quality will be noted in the report.

Forms

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:
-Informed Consent for Genetic Testing (T576)
-Informed Consent for Genetic Testing-Spanish (T826)
2. If not ordering electronically, complete, print, and send 1 of the following forms with the specimen:
-Neurology Specialty Testing Client Test Request (T732)

-Therapeutics Test Request (T831)
-Cardiovascular Test Request (T724)
-Kidney Transplant Test Request

Specimen Minimum Volume
See Specimen Required

Reject Due To
All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Clinical & Interpretive

Clinical Information
This panel provides a comprehensive analysis for multiple genes with strong drug phenotype associations. Each sample is tested for specific variations with known functional impact. Pharmacogenomic data for the following specific variants are reviewed and reported (if present):
-*CYP1A2 *1F*, *1K*, *6*, and *7*
-*CYP2C9 *2*, *3*, *4*, *5*, *6*, *8*, *9*, *11*, *12*, *13*, *14*, *15*, *16*, *17*, *18*, *25*, *26*, *28*, *30*, *33*, and *35*
-*CYP2C19 *2*, *3*, *4*, *5*, *6*, *7*, *8*, *9*, *10*, *17*, and *35*
-*CYP2D6 *2*, *3*, *4*, *4N*, *5*, *6*, *7*, *8*, *9*, *10*, *11*, *12*, *13*, *14A* (now known as *114*), *14B* (now known as *14*), *15*, *17*, *29*, *35*, *36*, *41*, *59, *68*, and *CYP2D6* gene duplication; additional *CYP2D6* variants may be detected through the reflex testing process
-*CYP3A4 *8*, *11*, *12*, *13*, *16*, *17*, *18*, *22*, and *26*
-*CYP3A5 *3*, *6*, *7*, *8*, and *9*
-*CYP4F2 *3*
-rs12777823G>A
-*SLCO1B1* rs4149056 (*5*)
-*VKORC1* c. -1639G>A, c.85G>T, c.106G>T, c.121G>T, c.134T>C, c.172A>G, c.196G>A, c.358C>T, and c.383T>G

Based on the results of each assay, a genotype is assigned, and a phenotype is predicted for each gene. Assessment of multiple genes may assist the ordering clinician with personalized drug recommendations, avoidance of adverse drug reactions, and optimization of drug treatment.

Reference Values
An interpretive report will be provided.

Interpretation
An interpretive report will be provided, which focuses on only drugs and genes with published pharmacogenomic practice guidance by the Clinical Pharmacogenetics Implementation Consortium, other professional organizations, or where strong US Food and Drug Administration guidance has been issued in drug labels.

For additional information regarding pharmacogenomic genes and their associated drugs, see Pharmacogenomic Associations Tables. This resource also includes information regarding enzyme inhibitors and inducers, as well as potential alternate drug choices.

Cautions

Specimens may contain donor DNA if obtained from patients who received non-leukocyte reduced blood transfusions or allogeneic hematopoietic stem cell transplantation. Results from specimens obtained under these circumstances may not accurately reflect the recipient's genotype. For individuals who have received blood transfusions, the genotype usually reverts to that of the recipient within 6 weeks. For individuals who have received allogeneic hematopoietic stem cell transplantation, a pretransplant DNA specimen is recommended for testing. Genetic test results in patients who have undergone liver transplantation may not accurately reflect the patient's genetic status for the genes on this panel.

This test is not designed to provide specific dosing recommendations and is to be used as an aid to clinical decision making only. Results should be used along with other clinical and laboratory data. Drug-label guidance should be used when dosing patients with medications regardless of the predicted phenotype.

For additional information, see the following tests:
-1A2Q / Cytochrome P450 *1A2* Genotype, Varies
-2C9QT / Cytochrome P450 *2C9* Genotype, Varies
-2C19R / Cytochrome P450 *2C19* Genotype, Varies
-2D6Q / Cytochrome P450 *2D6* Comprehensive Cascade, Varies
-3A4Q / Cytochrome P450 *3A4* Genotype, Varies
-3A5Q / Cytochrome P450 *3A5* Genotype, Varies
-SLC1Q / Solute Carrier Organic Anion Transporter Family Member 1B1 (*SLCO1B1*) Genotype, Statin, Varies
-WARSQ / Warfarin Response Genotype, Varies

Clinical Reference

1. Ji Y, Skierka JM, Blommel JH, et al. Preemptive pharmacogenomic testing for precision medicine: A comprehensive analysis of five actionable pharmacogenomic genes using next-generation DNA sequencing and a customized CYP2D6 genotyping cascade. J Mol Diagn. 2016;18(3):438-445. doi:10.1016/j.jmoldx.2016.01.003
2. Samwald M, Xu H, Blagec K, et al. Incidence of exposure of patients in the United States to multiple drugs for which pharmacogenomic guidelines are available. PLoS One. 2016;11(10):e0164972. doi:10.1371/journal.pone.0164972
3. Clinical Pharmacogenetic Implementation Committee (CPIC): Genes-Drugs. CPIC; Accessed April 4, 2025. Available at https://cpicpgx.org/genes-drugs/
4. Pharmacogenomics Knowledgebase (PharmGKB). Accessed April 4, 2025. Available at www.pharmgkb.org/
5. Crews KR, Monte AA, Huddart R, et al. Clinical Pharmacogenetics Implementation Consortium Guideline for CYP2D6, OPRM1, and COMT Genotypes and Select Opioid Therapy. Clin Pharmacol Ther. 2021;110(4):888-896. doi:10.1002/cpt.2149

Performance

Method Description

Genomic DNA is extracted from whole blood or saliva. Genotyping for each allele is performed using a polymerase chain

reaction (PCR)-based 5'-nuclease assay. Fluorescently labeled detection probes anneal to the target DNA. PCR is used to amplify the section of DNA that contains the variant. If the detection probe is an exact match to the target DNA, the 5'-nuclease polymerase degrades the probe, the reporter dye is released from the effects of the quencher dye, and a fluorescent signal is detected. Genotypes are assigned based on the allele-specific fluorescent signals that are detected.(Unpublished Mayo method)

CYP2D6 Copy Number Assay:
This assay utilizes a duplex real-time PCR, which includes 1 copy number probe and a reference assay per reaction. Each copy number probe detects the genomic sequence of interest and the reference assay detects a sequence that is known to be present in 2 copies in a diploid genome. This assay uses three probes which detect copy number in the promoter, intron 6, and exon 9 of the *CYP2D6* gene. Relative quantitation is used to determine the relative copy number of the target of interest in a genomic DNA (gDNA) sample normalized to10 ng/mcL for each probe. Each probe is normalized to the known copy number of the reference sequence and compared to a calibrator sample with known copies of the target sequence included with each run.(Package insert: Taqman Copy Number Assays. Applied Biosystems; Revision D, 02/2019)

2D6 Sequencing Assays (Tier 2, as needed):
The *CYP2D6* allele of interest is amplified by PCR. The PCR product is then purified and sequenced in both directions using fluorescent dye-terminator chemistry. Sequencing products are separated on an automated sequencer and trace files analyzed for variations in the exons and intron/exon boundaries of all 9 exons using mutation detection software and visual inspection.(Unpublished Mayo method)

PDF Report
Supplemental RE

Day(s) Performed
Varies

Report Available
3 to 14 days

Specimen Retention Time
Whole blood: 28 days (if available); Saliva: 30 days (if available); Extracted DNA: 3 months

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification

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

CPT Code Information

81418
0071U (if appropriate)
0072U (if appropriate)
0073U (if appropriate)
0074U (if appropriate)
0075U (if appropriate)
0076U (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
PGXQP	Focused Pharmacogenomics Panel, V	82118-1

Result ID	Test Result Name	Result LOINC® Value
610185	CYP1A2 Genotype	72884-0
610186	CYP1A2 Phenotype	94254-0
610187	CYP2C19 Genotype	57132-3
610188	CYP2C19 Phenotype	79714-2
610570	CYP2C19 Activity Score	104667-1
610189	CYP2C9 Genotype	46724-1
610190	CYP2C9 Phenotype	79716-7
610571	CYP2C9 Activity Score	104668-9
610191	CYP2D6 Genotype	40425-1
610192	CYP2D6 Phenotype	79715-9
610572	CYP2D6 Activity Score	104669-7
610193	CYP3A4 Genotype	81139-8
610194	CYP3A4 Phenotype	81145-5
610195	CYP3A5 Genotype	81140-6
610196	CYP3A5 Phenotype	79717-5
610197	SLCO1B1 Genotype	93412-5
610198	SLCO1B1 Phenotype	79722-5
610199	Warfarin CYP2C9 Genotype	46724-1
610201	Warfarin VKORC1 Resistance Genotype	50722-8
610200	Warfarin VKORC1 Promoter Genotype	50722-8
614000	Warfarin CYP2C9 and VKORC1 Promoter Phenotype	54451-0
610202	Warfarin CYP4F2 *3 Genotype	93197-2
610203	Warfarin rs12777823 Genotype	93198-0

610204	Interpretation	69047-9
610205	Additional Information	48767-8
610207	Disclaimer	62364-5
610208	Reviewed by	18771-6
610206	Method	85069-3