

## Focused Pharmacogenomics Panel, Varies

**Test ID:** PGXQP

**Useful for:**

Preemptive or reactive genotyping of patients for pharmacogenomic purposes

Providing an assessment for genes with strong drug-gene associations

**Genetics 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) 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.

**Testing Algorithm:**

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

**Reflex Tests:**

Test ID	Reporting Name	Available Separately	Always Performed
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

**Methods:**

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

**Reference Values:**

An interpretive report will be provided.

**Specimen Stability Information:**

Specimen Type	Temperature	Time
Varies	Varies	

## Specimen Requirements:

Multiple genotype tests can be performed on a single specimen after a single extraction.

## 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 specimen in original tube.

**Specimen Stability Information:** Ambient (preferred) 9 days/Refrigerated 30 days

**Specimen Type:** Saliva

**Patient Preparation:** Patient should not eat, drink smoke, or chew gum 30 minutes prior to collection.

**Supplies:** Saliva Swab Collection Kit (T786)

**Specimen Volume:** 1 swab

**Collection Instructions:** Collect and send specimen per kit instructions.

**Additional Information:** Due to lower concentration of DNA yielded from saliva, testing cannot proceed to reflex testing for 2D6 sequencing and will stop after initial testing is complete.

**Specimen Stability Information:** Ambient 30 days

**Specimen Type:** DNA

**Container/Tube:** 2 mL screw top tube

**Specimen Volume:** 100 mcL (microliters)

**Collection Instructions:**

1. The preferred volume is 100 mcL at a concentration of 50 ng/mcL.
2. Include concentration and volume on tube.

**Specimen Stability Information:** Frozen (preferred)/Ambient/Refrigerated

## Cautions:

Samples may contain donor DNA if obtained from patients who received non-leukoreduced blood transfusions or allogeneic hematopoietic stem cell transplantation. Results from samples 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

.

**CPT Code:**

0029U

0071U (if appropriate)

0072U (if appropriate)

0073U (if appropriate)

0074U (if appropriate)

0075U (if appropriate)

0076U (if appropriate)

**Day(s) Setup:** Monday through Friday

**Analytic Time:** 3 days

**Questions**

Contact your Laboratory Technologist Resource Coordinator Heather Flynn Gilmer at 800-533-1710.

