MAYO CLINIC LABORATORIES

Test Definition: 2B6Q

Cytochrome P450 2B6 Genotype, Varies

Overview

Useful For

Aiding in determining therapeutic strategies for drugs that are metabolized by cytochrome P450 (CYP) 2B6

Providing information relevant to efavirenz and sertraline, as well as other medications metabolized by CYP2B6

Determining the genotype if genotype-phenotype discord is encountered clinically after testing with a less comprehensive genotyping method has occurred

Identifying a genotype when required for drug trials and research protocols

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
_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

Testing Algorithm

For cord blood specimens that have an accompanying maternal blood specimen, maternal cell contamination studies will be performed at an additional charge.

Special Instructions

- Informed Consent for Genetic Testing
- Pharmacogenomic Association Tables
- Informed Consent for Genetic Testing (Spanish)

Method Name

Real Time-Polymerase Chain Reaction (RT-PCR) with Allelic Discrimination Analysis

NY State Available

Yes

Specimen

Specimen Type
Varies

Ordering Guidance
Testing is available as the single gene assay (this test) and as a part of a psychotropic pharmacogenomics panel.

If genotype testing for psychotropic medications is requested, order PSYQP / Psychotropic Pharmacogenomics Gene Panel, Varies.

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

Submit only 1 of the following specimens:

Specimen Type: Whole blood
Container/Tube: Lavender top (EDTA) or yellow top (ACD)
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 are 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, 1 mL
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 a Therapeutics Test Request (T831) with the specimen.

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

The cytochrome P450 (CYP) family of enzymes is a group of oxidative/dealkylating enzymes localized in the microsomes of many tissues including the intestines and liver. CYP2B6 is wholly or partially responsible for the metabolism of several commonly prescribed drugs.

The *CYP2B6* gene is highly variable with over 38 named alleles. This gene can have multiple sequence variations. Alleles thought to have an impact upon CYP2B6 enzyme function at the time that this test was developed are included in this test (see Table). Individuals without a detectable gene alteration will be reported as *CYP2B6*1/*1,* but it is possible that these individuals harbor unknown variants that may impact metabolism. In addition, some individuals have genes that are hybrids of *CYP2B6* and the *CYP2B7* pseudogene. The frequency of these hybrids is unknown, and this assay does not test for these hybrids.

Star allele genotypes are called based on the Pharmacogene Variation Consortium definitions (1) *CYP2B6* genotype results are used to predict metabolizer phenotypes based on the Clinical Pharmacogenetics Implementation Consortium published guidelines.(2,3) A *CYP2B6* phenotype is predicted based upon the number of functional, partially functional, and nonfunctional alleles present in a sample. In rare instances where alleles with unknown function are present in a homozygous or compound heterozygous state, an unknown phenotype occurs and a range is provided.

Several medications act as substrates of CYP2B6. CYP2B6-metabolized medications with published guidelines for the use of *CYP2B6* genotype to guide medication use include efavirenz and sertraline.(2-4) While methadone is known to be metabolized by CYP2B6, the data do not consistently demonstrate that variation in *CYP2B6* clinically impacts methadone efficacy or toxicity and a guideline suggests that no change in dosing based on *CYP2B6* genotype is required.(5) Bupropion, ketamine, and nevirapine are also known to be metabolized by CYP2B6, but no dosing guidelines are available. Other enzymes may be involved in the metabolism of these drugs.

There is a variable degree of substrate specificity exhibited by *CYP2B6* alleles on these medications. This means that the same allele (ie, *6) may not metabolize all substrates at exactly the same rate.

Drugs that are metabolized by CYP2B6 may require dosage adjustment based on the individual patient's genotype. For example, patients who are poor metabolizers may require much lower than usual doses to achieve optimal response in the case of drugs that are inactivated by the CYP2B6 enzyme. Alternatively, patients who are ultrarapid metabolizers may benefit from increased doses in the case of drugs that are inactivated by CYP2B6 enzyme. In the absence of clear guidance from the US Food and Drug Administration on dosing for various metabolizer phenotypes, patients with either ultrarapid or poor metabolism may benefit by switching to comparable alternate medications that are not primarily metabolized by CYP2B6 or by therapeutic drug monitoring where applicable.

Table. Function of Individual Star Alleles

Enzyme function	**Examples of *CYP2B6* star alleles**
Normal function	*1, *5
Increased function	*4
Decreased function	*6, *7, *9, *19, *20, *26, *36
No or null function	*8, *12, *13, *16 (also known as *18.002), *18, *38
Uncertain function	*11, *14, *15, *22, *27, *35

Reference Values

An interpretive report will be provided.

Interpretation

An interpretive report will be provided.

The genotype, with associated star alleles, is assigned using standard allelic nomenclature as published by the Pharmacogene Variation (PharmVar) Consortium.(1)

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

Rare variants may be present that could lead to false-negative or false-positive results.

Samples may contain donor DNA if obtained from patients who received non-leukocyte reduced 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, 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 this gene.

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

Clinical Reference

1. PharmVar. Pharmacogene Variation Consortium. Updated March 3, 2021. Accessed May 12, 2025. Available at www.pharmvar.org/
2. Clinical Pharmacogenetics Implementation Consortium (CPIC). Accessed May 12, 2025. Available at https://cpicpgx.org/
3. Desta Z, Gammal RS, Gong L, et al. Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for CYP2B6 and Efavirenz-Containing Antiretroviral Therapy. Clin Pharmacol Ther. 2019;106(4):726-733. doi:10.1002/cpt.1477
4. Bousman CA, Stevenson JM, Ramsey LB, et al. Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for *CYP2D6, CYP2C19, CYP2B6, SLC6A4,* and *HTR2A* Genotypes and Serotonin Reuptake Inhibitor Antidepressants. Clin Pharmacol Ther. 2023;114(1):51-68
5. Robinson KM, Eum S, Desta Z, et al. Clinical Pharmacogenetics Implementation Consortium Guideline for *CYP2B6* Genotype and Methadone Therapy. Clin Pharmacol Ther. 2024;116(4):932-938

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 DNA section 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)

PDF Report
No

Day(s) Performed
Varies

Report Available
3 to 8 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
81479

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
2B6Q	CYP2B6 Genotype, V	72511-9

Result ID	Test Result Name	Result LOINC® Value
610082	CYP2B6 Genotype	72882-4
610083	CYP2B6 Phenotype	79720-9
610566	CYP2B6 Activity Score	104666-3

610084	Interpretation	69047-9
610085	Additional Information	48767-8
610086	Method	85069-3
610087	Disclaimer	62364-5
610088	Reviewed by	18771-6