

Dihydropyrimidine Dehydrogenase Genotype, Varies

Test ID: DPYDQ

Useful for:

Identifying individuals with genetic variants in DPYD who are at increased risk of toxicity when prescribed 5-fluorouracil (5-FU) or capecitabine chemotherapy treatment

Genetics Information:

This is a pharmacogenomics test associated with 5-fluorouracil and capecitabine drug sensitivity. Biallelic variation in the DPYD gene is also associated with dihydropyrimidine dehydrogenase (DPD) deficiency. Individuals who have variations identified in the DPYD may benefit from genetic consultation.

Methods:

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

Advisory:

This test does not detect or report variants other than the *2A, *7, *8, *10, *13, rs67376798, rs75017182, and rs115232898 alleles. Sequencing of the full gene is also available for detection of additional variants as well as the alleles listed: order DPYDG / Dihydropyrimidine Dehydrogenase, DPYD Full Gene Sequencing, Varies.

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 tier 2 sequencing and will stop after tier 1 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 75 ng/mcL.
2. Include concentration and volume on tube.

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

Cautions:

Rare genetic variants may be present that could lead to false-negative or false-positive results. Other variants in the primer binding regions can affect the testing, and ultimately, the genotype assessment made.

Samples may contain donor DNA if obtained from patients who received heterologous blood transfusions or allogeneic blood or marrow 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 blood or marrow transplantation, a pretransplant DNA specimen is recommended for testing.

Dihydropyrimidine dehydrogenase (DPYD) genetic test results in patients who have undergone liver transplantation may not accurately reflect the patient's DPYD status.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Large deletions or rearrangements are not detected by this assay, and these may affect DPYD protein expression and their impact on fluoropyrimidine related side effects and tumor response.

This test is not designed to provide specific dosing or drug selection 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.

CPT Code:

81232

Day(s) Setup: Monday through Friday (not reported
on Saturday or Sunday)

Analytic Time: 3 days

Questions

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