Dihydropyrimidine Dehydrogenase (DPYD) Genotyping

Disease Overview
The DPYD gene (1p21.3) encodes the enzyme dihydropyrimidine dehydrogenase (DPYD) which plays a critical role in the metabolism of fluoropyrimidine drugs, such as 5-Fluorouracil (5-FU) and capecitabine. Mutations in the gene encoding this enzyme may result in decreased activity and can thereby cause severe toxicity.

Uses for Test
- Estimate genetic risk of dose-related toxicity with 5-fluorouracil (5-FU) and capecitabine.
- Identify genotypes shown to have a drug-gene variant relationship.
- Pharmacogenomic orders may be reviewed by a pharmacist for clinical appropriateness prior to test completion if clinical data is available.

Therapeutic Implications
To estimate genetic risk of abnormal drug metabolism for drugs metabolized by DPYD.

Treatment Guidelines
The Clinical Pharmacogenetics Implementation Consortium (CPIC) has published dosing guidelines for DPYD genotypes: https://cpicpgx.org/

Test Interpretation
- Clinical sensitivity: drug dependent
- Analytical sensitivity/specificity: > 99%

Results
A detailed report is provided. This report is reviewed and signed out by a Laboratory Director. No mutations detected is predictive of *1 functional enzyme.

Test Limitations
- Only the targeted DPYD variants will be detected.
- Lack of detection of the targeted DPYD variants does not exclude a risk for toxicity, nor predict degree of responsiveness. Diagnostic errors can occur due to rare sequence variations.
- Drug metabolism, efficacy and risk for toxicity may be affected by genetic and non-genetic factors that are not evaluated by this test.
- This result does not replace the need for therapeutic drug or clinical evaluation and monitoring.

Related Tests
- Multiple genes can be involved in drug metabolism, drug activation and drug action on the target tissue. Additional genotyping tests are available for CYP2D6, CYP2C19, CYP2C9, VKORC1, SLC01B1, IFNL3, CYP4F2, CYP3A4, SLC22A1, TPMT, and CYB5A as individual tests or as a PGx Panel.
- The panel includes a comprehensive medication report based on the genotypes detected.
- Therapeutic drug monitoring and/or metabolic ratios may be useful for evaluating the pharmacokinetics of a particular drug, for a particular patient.

Sample Requirements
Collection
- Lavender-top tube (EDTA)
- All specimens should be sent in the original container and should not be aliquoted to another tube
- The specimen submitted should only be used for this testing and should not be shared with any other testing that would also utilize this specimen type

Specimen
- Whole blood, preferred Volume: 2 mL to 4 mL
  (1mL minimum)

Stability
- Room temp – 72 hours
- Refrigerated – 7 days
- Frozen – 7 days
- Not affected by hemolysis
- Not affected by lipemia

Tests Involved
- CPT code: 81232
- Lab Test ID: LB0149

Test Schedule
- Set up Monday to Friday
- Turn Around Time: 5-7 days

Additional information
- These tests are available through the Sanford Imagenetics program. Contact Sanford Laboratories at (605) 328-5464 or (800) 522-2561 for questions regarding this testing.