TECHNICAL SPECIFICATIONS

Basic Pharmacogenetic Panel Testing

Disease Overview

How a patient processes and responds to medications is influenced by the genetic changes (variants) a person carries.

Pharmacogenetics (PGx) is the study of these gene variants related to a body's response to and interaction with many common prescription and over-the-counter medications. These gene variants are associated with a predicted drug response or drug disposition which may predispose a patient to risk of drug-related toxicity or lack of therapeutic benefit.

Genotyping identifies known gene changes (variants) that affect an individual's metabolism. This information can help to determine the optimal therapy and dosing in order to avoid ineffectiveness or intolerance or drugs in some individuals.

The goal of the PGx panel is to reduce the number of adverse drug reactions and identify non-responders who may benefit from a different medication or dosage, thus providing prophylactic guidance for drug and dose selection.

Uses for Test

- To estimate the risk of abnormal drug metabolism due to specific gene variants involving multiple drug classes; such as statins, specific psychologic and pain medications, and anticoagulants.
- To attempt to identify the cause of personal or family history of an adverse drug reaction or therapeutic failure for a large group of drugs and thereby guide drug and dose selection.
- The 11 following genes are included in this panel: *CYP2C9, VKORC1, SLCO1B1, TPMT, DPYD, CY2C19, CYP3A5, CYP2D6, CYP4F2, CYP2C cluster, IFNL3*.
- Pharmacogenomic orders may be reviewed by a pharmacist for clinical appropriateness prior to test completion if clinical data is available.

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

Methodology

Polymerase chain reaction (PCR)/allele-specific primer extension.

Tests Involved

- *CYP2C9, VKORC1, SLCO1B1, TPMT, DPYD, CY2C19, CYP3A5, CYP2D6, CYP4F2, CYP2C cluster, IFNL3*
- CPT code: 81479
- Lab Test ID: (LBOR0174)

Test Schedule

- Monday to Friday.
- Turn Around Time: 5-7 days.

Test Interpretation

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

Results

A detailed report is provided. This report is reviewed and signed out by the Laboratory Director. The major *CYP2C9, VKORC1, SLCO1B1, TPMT, DPYD, CY2C19, CYP3A5, CYP2D6, CYP4F2, CYP2C cluster,* and *IFNL3* genotypes will be identified and classified.
The panel includes a comprehensive medication report based on the genotypes detected.

Test Limitations

- Only the targeted genes and variants of the genes tested will be detected.
- Diagnostic errors can occur due to rare sequence variations.
- Risk of therapeutic failure or adverse reactions may be affected by genetic and nongenetic factors that are not detected by this test.
- This result does not replace the need for therapeutic drug or clinical monitoring.
- If the DNA is directly sent to the laboratory for analysis by an external laboratory, the Sanford Medical Genetics Laboratory takes no responsibility or liability for sample swaps or extraction errors occurring prior to receipt of the DNA sample by Sanford Medical Genetics Laboratory.

Related Tests

- Genotyping tests are available for each gene included in this panel: *CYP2C19, CYP2D6, CYP2C9, SLCO1B1, TPMT, CYP3A5, IFNL3, CYP2C9,VKORC1, CYP4F2, CYP2C cluster* and *DPY*D as individual tests.
- Therapeutic drug monitoring and/or metabolic ratios may be useful for evaluating the pharmacokinetics of a particular drug for a particular patient.

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.

References

CPIC: Clinical Pharmacogenetics Implementation Consortium (https://cpicpgx.org/)
Pharmacogenomics Knowledgebase (PharmGKB) (https://www.pharmgkb.org/)

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