PGX CYP2B6 Genotyping

CPT Code(s): 81479

Service Code (IU Health): 53102257

Ordering Recommendation: Information about genotype may be used as an aid to clinicians in determining therapeutic strategy and treatment dose for therapeutics that are metabolized by CYP2B6.

Synonyms: CYP2B6, efavirenz, methadone

Methodology: realtime polymerase chain reaction (PCR) and microarray analysis

Performed: weekly

Reported: 7-10 days

Specimen Requirements

Patient Preparation: None required for whole blood; Saliva: no eating, drinking, smoking or chewing gum 30 min. prior to collection

Collect: Preferred: Lavender (EDTA); Acceptable: Yellow (ACD Solution A or B), Green (Heparin), Saliva (Oragene collection device)

Specimen Volume: Blood: 3 mL whole blood (minimum 1 mL); Saliva: 2 mL

Storage/Transport: Refrigerated/Room temperature

Unacceptable Conditions: Grossly hemolyzed or clotted

Remarks:

Stability: 1 month refrigerated; 1 month frozen
Reference Interval: by report

Interpretive Data

Characteristics: Impaired drug metabolism causing adverse drug reactions or lack of therapeutic response. Drugs metabolized by CYP2B6 include: efavirenz, methadone

Inheritance: autosomal recessive

Cause: CYP2B6 allelic variants

Penetrance: Drug dependent

Clinical sensitivity: unknown

Analytical sensitivity and specificity: 99%

Limitations: Only the targeted CYP2B6 variants will be detected. Mutations or variants in other genes will not be detected. Although rare, false positive or false negative results may occur. All results should be interpreted in context of clinical findings, relevant history, and other laboratory data.

References: http://www.pharmgkb.org/gene/PA123

Variants Tested

<table>
<thead>
<tr>
<th>Allele</th>
<th>variant</th>
<th>dbSNP</th>
<th>Predicted enzyme activity</th>
</tr>
</thead>
<tbody>
<tr>
<td>*1</td>
<td>Assumed when no variant detected</td>
<td></td>
<td>Normal</td>
</tr>
<tr>
<td>*6</td>
<td>c.516G&gt;T; rs3745274</td>
<td></td>
<td>Decreased function</td>
</tr>
<tr>
<td>*18</td>
<td>c.983T&gt;C rs28399499</td>
<td></td>
<td>Decreased expression</td>
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