PGX CYP2C9 Genotyping

CPT Code(s): 81227

Service Code (IU Health): 53102208

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 CYP2C9.

Synonyms: CYP2C9, phenytoin, warfarin, glyburide, ibuprofen

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 CYP2C9 include: phenytoin, warfarin, glyburide, ibuprofen.

Inheritance: autosomal recessive

Cause: CYP2C9 allelic variants

Incidence: The carrier frequency is approximately 20% in Caucasians, 5% in African Americans, and 4% in Asians.

Penetrance: Drug dependent

Clinical sensitivity: unknown

Analytical sensitivity and specificity: 99%

Limitations: Only the targeted CYP2C9 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.


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>*2</td>
<td>c.430C&gt;T</td>
<td>rs1799853</td>
<td>Decreased function</td>
</tr>
<tr>
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<td>rs28371686</td>
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<tr>
<td></td>
<td>Variant</td>
<td>Reference SNP</td>
<td>Function</td>
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<td>---</td>
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<td>c.818delA</td>
<td>rs9332131</td>
<td>Non-functional</td>
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<td>c.449G&gt;A</td>
<td>rs7900194</td>
<td>Decreased function</td>
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<td>c.1003C&gt;T</td>
<td>rs28371685</td>
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