Voltage-gated sodium channels are transmembrane glycoprotein complexes composed of a large alpha subunit and one or more regulatory beta subunits. They are responsible for the generation and propagation of action potentials in neurons and muscle. *SCN3B* gene encodes one member of the sodium channel beta subunit gene family, and influences the inactivation kinetics of the sodium channel. This gene contains six exons (1 noncoding) spanning 26 kb of genomic distance that has been mapped to chromosome 11q23.3. Two alternatively spliced variants, encoding the same protein, have been identified. Defects in this gene cause Brugada syndrome type 7 (BRS7). *SCN3B* mutations demonstrate autosomal dominant inheritance with a broad range of clinical severity both within and between families.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for *SCN3B* mutations. Individuals are tested by DNA sequencing of the coding exons of the *SCN3B* gene. We strongly recommend initial testing of a clearly affected individual, if available, in order to provide the greatest test sensitivity and clearest interpretation of results for subsequent family members. Genetic counseling is recommended for all individuals.

### REASONS FOR REFERRAL

- Molecular confirmation of Brugada syndrome type 7 (BRS7).

### METHODOLOGY

Genomic DNA is analyzed for *SCN3B* mutations by DNA sequencing of the coding exons of the *SCN3B* gene, as well as the exon/intron junctions and a portion of the 5’ and 3’ untranslated regions. Patient DNA is sequenced in both the forward and reverse orientations. If a mutation is identified, additional family members are analyzed only for the familial mutation by automatic fluorescent DNA sequencing.

### SERVICE FEES

<table>
<thead>
<tr>
<th></th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<tbody>
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<td>Index Case (Male or Female)</td>
<td>$750 per sample</td>
<td>81404</td>
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<tr>
<td>Additional Family Members</td>
<td>$300 per sample; known familial mutation only</td>
<td>81403</td>
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### SENSITIVITY

DNA Sequencing Analysis: Approximately 99% detection of mutations in the five coding exons of *SCN3B*.

### SPECIMEN REQUIREMENTS

- **Blood (preferred):** EDTA (purple-top) tubes: *Adult:* 5 cc  *Child:* 5 cc  *Infant:* 2-3 cc
- **Tissue:** Frozen (preferred), RNA*Later*
- **Other Body Fluids and Formalin-fixed, Paraffin-embedded Tissue:** Call to inquire

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