FANCD2 (Fanconi Anemia, Complementation Group D2) encodes a members of the Fanconi anemia complementation group D2. This protein is monoubiquinated in response to DNA damage, required for maintenance of chromosomal stability and plays a role in preventing breakage and loss of missegregating chromatin at the end of cell division, particularly after replication stress. Human FANCD2 gene contains 43 exons maped within chromosome 3p25.3. Mutations in the FANCD2 gene can cause Fanconi anemia Complementation Group D2. Fanconi anemia is a genetically heterogeneous recessive disorder characterized by cytogenetic instability, hypersensitivity to DNA crosslinking agents, increased chromosomal breakage, and defective DNA repair. Characteristic clinical features include developmental abnormalities in major organ systems, early-onset bone marrow failure, and a high predisposition to cancer. Malformations were frequent in FA-D2 patients, and hematologic manifestations appeared earlier and progressed more rapidly when compared with all other patients (FA-non-D2). Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for FANCD2 mutations. Individuals are tested by automatic fluorescent DNA sequencing of the coding exons of the FANCD2 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 the diagnosis of CHD and Fanconi anemia (FA).

### METHODOLOGY

Genomic DNA is analyzed for FANCD2 mutations by DNA sequencing of the coding exons of the FANCD2 gene, as well as the exon/intron junctions and a portion of the 5’ and 3’ untranslated region. 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>
</tr>
</thead>
<tbody>
<tr>
<td>Index Case (Male or Female)</td>
<td>$1,500 per sample</td>
<td>81407</td>
</tr>
<tr>
<td>Additional Family Members</td>
<td>$300 per sample; known familial mutation only</td>
<td>81403</td>
</tr>
</tbody>
</table>

### SENSITIVITY

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 2-43 of FANCD2.

### SPECIMEN REQUIREMENTS

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

Contact Information:
- John Welsh Cardiovascular Diagnostic Laboratory • Section of Cardiology • Department of Pediatrics
- Baylor College of Medicine • 1102 Bates Avenue, Suite 480.02 • Houston, TX 77030
- PHONE: (832) 824-4155 • FAX: (832) 825-5159 • E-MAIL: yuxinf@bcm.edu
- Web Site: [www.bcm.edu/pediatrics/welsh](http://www.bcm.edu/pediatrics/welsh)