FANCE (Fanconi Anemia, Complementation Group E) encodes a part of the Fanconi anemia (FA) nuclear complex which is essential for protection against chromosome breakage. FANCE is required for the nuclear accumulation of FANCC and provides a critical bridge between the FA complex and FANCD2. Human FANCE gene contains 10 exons mapped within chromosome 6p21.31. Mutations in the FANCE gene can cause Fanconi anemia complementation group E. 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. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for FANCE mutations. Individuals are tested by automatic fluorescent DNA sequencing of the coding exons of the FANCE 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 complementation group E.

**METHODOLOGY**

Genomic DNA is analyzed for FANCE mutations by DNA sequencing of the coding exons of the FANCE 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**

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<tr>
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<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<tr>
<td>Index Case (Male or Female)</td>
<td>$1,000 per sample</td>
<td>81405</td>
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<tr>
<td>Additional Family Members</td>
<td>$300 per sample; known familial mutation only</td>
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**SENSITIVITY**

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-10 of FANCE.

**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