The **FKTN** gene is located at 9q31.2 and encodes a 461-amino acid transmembrane protein. **FKTN** is localized to the cis-Golgi compartment, where it may be involved in the glycosylation of alpha-dystroglycan in skeletal muscle. **FKTN** is a glycosyltransferase and plays a role in brain development. **FKTN** mutations are passed on in an autosomal recessive fashion. Defects in this gene cause Fukuyama-type congenital muscular dystrophy (FCMD), Walker-Warburg syndrome (WWS), limb-girdle muscular dystrophy type 2M (LGMD2M), and dilated cardiomyopathy type 1X (CMD1X). Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for **FKTN** mutations. Individuals are tested by DNA sequencing of the coding exons of the **FKTN** 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 Fukuyama-type congenital muscular dystrophy (FCMD), Walker-Warburg syndrome (WWS), limb-girdle muscular dystrophy type 2M (LGMD2M), and dilated cardiomyopathy type 1X (CMD1X).

### METHODOLOGY

Genomic DNA is analyzed for **FKTN** mutations by DNA sequencing of the coding exons of the **FKTN** 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>Index Case (Male or Female)</th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>$900 per sample</td>
<td>81405</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 1-9 of **FKTN**.

### SPECIMEN REQUIREMENTS

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