The DTNA gene encodes alpha dystrobrevin (DTNA) which is arrayed transversely in sarcomere A-bands and binds myosin heavy chain in thick filaments and titin in elastic filaments. The DTNA gene contains 22 exons and spans around 180 kb genomic distance that was mapped to chromosome 18q12.1. Defects in this gene are the cause of left ventricular noncompaction (LVNC), which is rare congenital cardiomyopathy which results from the failure of myocardial development during embryogenesis. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for DTNA mutations. Individuals are tested by DNA sequencing of the coding exons of the DTNA 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 left ventricular noncompaction (LVNC).

METHODOLOGY

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

Direct and Institutional Billing

Index Case (Male or Female) $1300 per sample  
Additional Family Members $300 per sample; Known familial mutation only

CPT Codes

81406

81403

SENSITIVITY

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons of DTNA.

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