Barth Syndrome is rare and the prevalence is unknown as the disease is thought to be under diagnosed (1). The TAZ gene codes for taffazin, a protein important for normal cardiolipin production and mitochondrial function. Cardiolipin is an important component of the inner mitochondrial membrane and is closely associated with the electron transport chain (2). Mutations in the TAZ gene can result in cardiomyopathy (dilated cardiomyopathy and/or left ventricular non-compaction), neutropenia, skeletal myopathy, growth deficiency, and 3-methylglutaconic aciduria. Female carriers of TAZ mutations do not have features of Barth syndrome. The TAZ gene contains 11 exons and is located at chromosome Xq28.

The majority of individuals with a clinical diagnosis of Barth syndrome have TAZ mutations (3, 4). TAZ mutations are inherited in an X-linked recessive manner. Approximately 2/3 of males with Barth have a confirmed or suspected family history of the condition (4).

**Indication**

TAZ testing is utilized to confirm a diagnosis of Barth syndrome in patients with clinically evident disease. Barth syndrome should be considered when males present with cardiomyopathy, especially when associated with left ventricular noncompaction, neutropenia, skeletal muscle weakness, or family history suggestive of X-linked inheritance. We recommend testing the most clearly affected individual in the family whenever possible.
Methodology:

All 11 exons of the TAZ gene, as well as the exon/intron boundaries and a portion of untranslated regions of the gene are amplified by PCR. Genomic DNA sequences from both forward and reverse directions are obtained by automatic fluorescent detection using an ABI PRISM® 3730 DNA Analyzer. Sequence variants different from National Center for Biotechnology Information GenBank references are further evaluated for genetic significance. If a mutation is identified, a known familial mutation analysis will be available for additional family members.

Sensitivity & Accuracy:

Greater than 98.5% of the mutations in exon 1-11 of TAZ are detectable by sequence based methods. Sequencing does not detect deletions or duplications in carrier females but may detect these changes in affected males. Mutations in TAZ account for the majority of cases of Barth syndrome.

References:


Specimen:

Peripheral blood in EDTA tube
Adult: 3-5mL
Child: 3-5mL
Infant: 1-3mL
For other specimen types, please contact Amy Shikany at 513-803-3317

Turnaround Time:

Full Mutation Analysis 2-4 weeks
Known Mutation Analysis 1-2 weeks

CPT Codes:

Full Gene Sequencing 81406
Additional Family Members 81403