Fucosidosis: *FUCA1* Gene Deletion/Duplication

**Test Code:** DFUCA  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1

### Condition Description

Fucosidosis is an autosomal recessive lysosomal storage disorder due to deficient activity of the alpha-1-fucosidase enzyme. When this enzyme is deficient, a build-up of fucosyl-glycolipids, glycopeptides, and oligosaccharides occurs in various tissues. Major features of fucosidosis include neurodegeneration with progressive motor and mental deterioration. Additional features include muscle wasting and dystrophy, growth retardation, spasticity, contractures, recurrent infections, seizures, coarse features, dysostosis multiplex, angiokeratoma corporis diffusum, ocular abnormalities, visceromegaly, and hearing loss. Fucosidosis has a wide continuous clinical spectrum; however, all of the features are progressive and ultimately lead to an early death. The disease may take a rapid course with death occurring in infancy or it may be more mild, with death occurring in adulthood.

Mutations in the *FUCA1* gene (1p36.11) cause fucosidosis. Missense, nonsense, and splice-site mutations have been reported as well as small deletions, large deletions, insertions, and duplications. Willems *et al.* (1999) report 79 out of 80 mutations in 40 patients.

For patients with suspected fucosidosis, sequence analysis is recommended as the first step in mutation identification. For patients in whom mutations are not identified by full gene sequencing, deletion/duplication analysis is appropriate.

### References:

- OMIM #612280: *FUCA1* gene
- OMIM #230000: Fucosidosis

### Genes

**FUCA1**

### Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of fucosidosis in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of fucosidosis in whom sequence analysis was negative.

### Methodology

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region.

### Detection

Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

### Specimen Requirements

*Submit only 1 of the following specimen types*

**Type: DNA, Isolated**

**Specimen Requirements:**
- Microtainer
- 3µg Isolation using the Perkin Elmer™ Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

**Specimen Collection and Shipping:**
- Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

**Type: Whole Blood (EDTA)**

**Specimen Requirements:**
- EDTA (Purple Top)
- Infants and Young Children (2 years of age to 10 years old): 3-5 ml
- Older Children & Adults: 5-10 ml
- Autopsy: 2-3 ml unclotted cord or cardiac blood

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Specimen Collection and Shipping:
Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

Special Instructions
Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside of EGL Genetics, please submit a copy of the sequencing report with the test requisition.

Related Tests
- Sequence analysis of the *FUCA1* gene is available and is required before deletion/duplication analysis.
- Custom diagnostic mutation analysis (KM) is available to family members if mutations are identified by targeted mutation testing or sequencing analysis.