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, angio-keratoma 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.

Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

### 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

* **Preferred specimen type:** Whole Blood

**Type: Whole Blood**

Specimen Requirements:

- In EDTA (purple top) tube: Infants (2 years): 3-5 ml  
  Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

**Type: Saliva**
Specimen Requirements:

Oragene™ Saliva Collection kit (available through EGL) used according to manufacturer instructions.

Specimen Collection and Shipping: Store sample at room temperature. Ship sample within 5 days of collection at room temperature with overnight delivery.

**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.