FTSJ1-related Intellectual Disability: *FTSJ1* Gene Sequencing

| Test Code: TK | Turnaround time: 4 weeks | CPT Codes: 81479 x1 |

**Condition Description**

Mutations or absence of the *FTSJ1* gene (Xp11.23) have been associated with non-syndromic X-linked intellectual disability. In one family, affected members had nonprogressive intellectual disability noted during childhood, and several demonstrated aggressive behavior. The intellectual disability in tested members of the family was moderate to severe; none of the patients was able to read, write, or solve simple arithmetic problems. In another family of three affected brothers, two had moderate and one had severe intellectual handicap. The oldest patient had autistic behavior that lessened after age 5 years, as well as delayed speech and motor development. The two younger brothers also had seizures; one had flat nasal bridge and shortened distal phalanges. This family had a deletion of the region which included the *FTSJ1* gene. Female carriers often show no symptoms due to the complete inactivation of the aberrant X chromosome.

For patients with suspected *FTSJ1*-related intellectual disability, 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.

Click [here](#) for the OMIM summary on this condition.

**Genes**

*FTSJ1*

**Indications**

This test is indicated for:

- Confirmation of a clinical/biochemical diagnosis of *FTSJ1*-related intellectual disability
- Carrier testing in adult females with a family history of *FTSJ1*-related intellectual disability

**Methodology**

Full Gene Sequencing: PCR amplification of 12 exons contained in the *FTSJ1* gene is performed on patient genomic DNA. Direct sequencing of amplification products is performed in both the forward and reverse directions using automated fluorescence dideoxy sequencing methods. Patient gene sequences are compared to a normal reference sequence. Sequence variations are then classified as mutations, benign variants unrelated to disease, or variations of unknown clinical significance. Variants of unknown clinical significance may require further studies of the patient and/or family members. This assay does not interrogate the promoter region, deep intronic regions or other regulatory elements. Large deletions are not detected by this analysis.

**Detection**

Clinical Sensitivity: Unknown. Mutations in the promoter region, some mutations in the introns and other regulatory element mutations cannot be detected by this analysis. Large deletions will not be detected by this analysis. Results of molecular analysis should be interpreted in the context of the patient's biochemical phenotype.

Analytical Sensitivity: ~99%

**Specimen Requirements**

Submit only 1 of the following specimen types

* Preferred specimen type: Whole Blood

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) or ACD (yellow 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**

Submit copies of diagnostic biochemical test results with the sample, if appropriate. Contact the laboratory if further information is needed.

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**

- **Deletion/duplication analysis of the FTSJ1 gene** by CGH array is available for those individuals in whom sequence analysis is negative (TN).
- A CGH array-based test for deletion/duplication analysis of 64 different X-linked intellectual disability genes is available (OL).
- Custom diagnostic mutation analysis (KM) is available to family members if mutations are identified by targeted mutation testing or sequencing analysis.
- Prenatal testing is available to adult females who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.