FTSJ1-related Intellectual Disability: *FTSJ1* Gene Deletion/Duplication

| Test Code: TN |
| Turnaround time: 2 weeks |
| CPT Codes: 81405 x1 |

**Condition Description**

Mutations or absence of the *FTSJ1* (Xp11.23) gene 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.

Please 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 in an individual in whom sequencing analysis was negative.
- Carrier testing in adult females with a family history of *FTSJ1*-related intellectual disability in whom sequencing 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.

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delivery.

**Special Instructions**

Please submit copies of diagnostic biochemical test results along 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**

- **FTSJ1-Related Intellectual Disability: FTSJ1 Gene Sequencing (TK)** is required before deletion/duplication analysis.
- **X-Linked Mental Retardation: 64-Gene Deletion/Duplication (OL).**
- **Prenatal Custom Diagnostics** 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.