

XLMR 58: *TSPAN7* Gene Deletion/Duplication

Test Code: DTSPA

Turnaround time: 2 weeks

CPT Codes: 81228 x1

Condition Description

Intellectual disability (ID) is a nonprogressive cognitive impairment affecting 1-3% of the Western population. It is estimated that up to 50% of moderate-severe cases have genetic causes and approximately 10% are due to X-linked intellectual disability disorders (XLID). XLID can be syndromic or nonsyndromic and is observed in all ethnic groups. More than 100 XLID syndromes have been described in the literature to date. Fragile X is the most common XLID syndrome (~1 in 4000 males) while others can be quite rare with only a few patients reported in the literature. Males can have moderate to severe intellectual disability depending on the syndrome, and carrier females can also be affected, but typically have milder clinical symptoms.

Zemni et al. (2000) described a female individual with mild intellectual disability and minor autistic features. She had a balanced translocation involving the X chromosome [46,X,t(X;2)(p11.2;p21.3)]. The *TSPAN7* gene, formerly known as the *TM4SF2* gene, is located at the breakpoint on the X chromosome. *TSPAN7* transcripts from this individual were barely detectable when compared with *TSPAN7* transcripts from controls. The *TSPAN7* gene is expressed in both the fetal and adult brain.

Zemni et al. identified two other mutations in the *TSPAN7* gene in two families with XLID. Both mutations were not found in 100 control chromosomes. Males and females can have mild to moderate intellectual disability.

References:

- Zemni et al. (2000). Nat Gen, 24:167-170.
- Maranduba et al. (2004). Am J Med Genet, 124A:413-415.
- [OMIM #300096](#): *TSPAN7* gene

Genes

[TSPAN7](#)

Indications

This test is indicated for:

- Confirmation of a clinical/biochemical diagnosis of XLMR 58 in individuals who have tested negative for sequence analysis.
- Carrier testing in adults with a family history of XLMR 58 who have tested negative for sequence analysis.

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

Specimen Collection and Shipping:

Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not 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

- Sequencing analysis of the *TSPAN7* gene is available and is required before deletion/duplication 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.
- X-Linked Intellectual Disability panels are available for 30, 60, and 90 genes.