

## Jalili Syndrome: *CNNM4* Gene Deletion/Duplication

**Test Code:** DCNNM

**Turnaround time:** 2 weeks

**CPT Codes:** 81228 x1

### Condition Description

Jalili syndrome is an autosomal recessive condition characterized by visual impairment and amelogenesis imperfecta (abnormal enamel). In families with Jalili syndrome, all affected individuals had significant visual impairment, starting in infancy or early childhood and progressing with age, and abnormal enamel of the primary and secondary dentitions and were susceptible to rapid posteruptive failure due to hypomineralization. The first clinical sign of visual impairment is often nystagmus. Photophobia and achromatopsia are also present in individuals with Jalili syndrome. Pathogenic variants in the *CNNM4* gene (2q11.2) cause Jalili syndrome.

For patients with suspected Jalili syndrome, sequence analysis is recommended as the first step in pathogenic variant identification. For patients in whom pathogenic variants are not identified by full gene sequencing, deletion/duplication analysis is appropriate.

References:

- OMIM #607805: *CNNM4* gene
- OMIM #217080: Jalili syndrome
- Doucette et al. (2013), *Ophthalmic Genet*, 34:19-29.
- Parry et al. (2009), *Am J Hum Genet*, 84:266-273.

### Genes

[CNNM4](#)

### Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of Jalili syndrome in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of Jalili syndrome 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 pathogenic variants. 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: 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.

#### 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.

## 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 *CNNM4* 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.
- Prenatal testing is available only for known familial mutations to individuals who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.