

## Joubert Syndrome: Deletion/Duplication Panel

**Test Code:** MD136

**Turnaround time:** 2 weeks

**CPT Codes:** 81405 x1, 81228 x1

### Condition Description

Joubert syndrome (JS) is an autosomal recessive multisystem disease characterized by cerebellar vermis hypoplasia with prominent superior cerebellar peduncles (resulting in the 'molar tooth sign', or MTS, on axial MRI), intellectual disability, hypotonia, irregular breathing pattern, and eye movement abnormalities. Some individuals with JS have retinal dystrophy and/or progressive renal failure characterized as nephronophthisis. The disorder in such patients is referred to as cerebellooculorenal syndrome, or CORS. Individuals with a mild form of JS have been shown to have a homozygous deletion of the NPHP1 gene identical, by mapping, to that in subjects with nephronophthisis alone. Please note, the *CEP164* gene is not included in the NGS panel at this time due to presence of at least one pseudogene. For clinicians that would like *CEP164* analysis in the event that all other genes test negative, we request that you contact EGL directly. Please note, the *TMEM138* and *TMEM231* genes are not included on the NGS panel at this time as these genes are only partially annotated in hg19. *TMEM138* and *TMEM231* will be re-evaluated with the release of hg20.

References:

- Daiger *et al.* (1998) *Invest Ophthalmol Vis Sci* 39:S295.
- OMIM
- GeneReviews

### Genes

[AHI1](#), [ARL13B](#), [CC2D2A](#), [CEP290](#), [CEP41](#), [CPLANE1](#), [KIF7](#), [NPHP1](#), [OFD1](#), [RPGRI1L](#), [TCTN1](#), [TCTN2](#), [TCTN3](#), [TMEM216](#), [TMEM237](#), [TMEM67](#), [TTC21B](#), [ZNF423](#)

### Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of Joubert syndrome.
- Carrier testing in adults with a family history of Joubert syndrome.

### Methodology

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region.

### Detection

**Deletion/Duplication:** 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

Please include fundus photographs, electroretinogram (ERG) findings, visual field findings, and visual acuity, if available, for expert review and clinical correlation with test results.

## Related Tests

- Joubert Syndrome: Sequencing Panel
- Eye Disorders: Comprehensive Sequencing Panel
- Eye Disorders: Deletion/Duplication Panel