Usher Syndrome: Deletion/Duplication Panel

**Test Code:** MD237  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1, 81406 x1

### Condition Description

Usher syndrome is a disorder consisting of retinitis pigmentosa (RP) and congenital hearing loss, ranging from partial or profound. Several clinical subtypes exist. Usher syndrome type I is generally characterized by profound congenital hearing loss with no vestibular function and early onset RP. Usher syndrome type II is generally characterized by mild to severe pre-lingual hearing loss with intact vestibular function and adolescent or adult onset RP. Usher syndrome type III is characterized by progressive post-lingual hearing loss, variable vestibular impairment, and late onset RP.

### References:
- OMIM
- GeneReviews
- Emory and Rimoin’s Principles and Practice of Medical Genetics, 5th Edition

### Genes

ABHD12, ADGRV1, CDH23, CIB2, CLRN1, HARS, MYO7A, PCDH15, USH1C, USH1G, USH2A, WHRN

### Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of Usher syndrome.
- Carrier testing in adults with a family history of Usher 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. 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**

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

Specimen Requirements:
- In EDTA (purple top) tube:
  - Infants (2 years): 3-5 ml
  - Older Children & Adults: 5-10 ml
- Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

#### Type: Isolated DNA

Specimen Requirements:
- In microtainer: 10 ug
- Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.
Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of 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**

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