Senior-Loken Syndrome: Deletion/Duplication Panel

**Test Code:** MD235  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1, 81405 x1

### Condition Description
Senior-Loken syndrome is an autosomal recessive retinal-renal syndrome with clinical features of nephronophthisis and Leber congenital amaurosis (LCA).

### References:
- OMIM
- GeneReviews

### Genes
- CEP290
- INVS
- IQCB1
- NPHP1
- NPHP3
- NPHP4
- SDCCAG8

### Indications
This test is indicated for:
- Confirmation of a clinical diagnosis of Senior-Loken syndrome.
- Carrier testing in adults with a family history of Senior-Loken 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

#### 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

Disclaimer: This information is confidential and subject to change without notice. It may not be reproduced in whole or part unless authorized in writing by an authorized EGL representative.
- Eye Disorders: Comprehensive Sequencing and Deletion/Duplication Panels.
- Senior-Loken Syndrome: Sequencing Panel.