Congenital Stationary Night Blindness: Deletion/Duplication Panel

**Test Code:** MD133  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1, 81479 x1

### Condition Description

Congenital stationary night blindness (CSNB) commonly refers to a non-progressive disorder with impaired night vision, reduced visual acuity, myopia, and defective dark adaptation (scotopic ERG defect) with usually normal appearance of the retina on fundus examination. CSNB may be inherited in an autosomal recessive, autosomal dominant, or X-linked manner.

References:

- OMIM
- GeneReviews

### Genes

- CABP4
- CACNA1F
- CACNA2D4
- GNAT1
- GPR179
- GRM6
- LRIT3
- NYX
- PDE6B
- RBP4
- RHO
- SAG
- SLC24A1
- TRPM1

### Indications

This test is indicated for:

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

### Methodology

**Deletion/Duplication:** 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**

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: DNA, Isolated

**Specimen Requirements:**

- Microtainer
- 3µg

Isolation using the PerkinElmer™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

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.
• Congenital Stationary Night Blindness: Sequencing Panel.