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
- RDH5
- RH0
- 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.

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

**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.

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.
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

- Eye Disorders: Comprehensive Sequencing and Deletion/Duplication Panels.
- Congenital Stationary Night Blindness: Sequencing Panel.