Congenital Central Hypoventilation Syndrome: Deletion/Duplication Panel

**Test Code:** MD244  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1, 81403 x1

### Condition Description

Congenital central hypoventilation syndrome (CCHS) is a disorder of respiratory and autonomic regulation typically presenting in newborns. It is characterized by hypoventilation with monotonous respiratory rates and shallow breathing. Some individuals present with altered development of neural crest-derived structures and/or tumors of neural crest origin (neuroblastoma, ganglioneuroma and ganglioneuroblastoma). A milder later-onset presentation of the syndrome can be seen in toddlers, children and adults.

### References:
- Genereviews
- OMIM

### Genes

ASCL1, BDNF, EDN3, GDNF, HOXA1, PHOX2B, RET

### Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of congenital central hypoventilation 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 Analysis:** Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. 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.
Related Tests

- Congenital Central Hypoventilation Syndrome: Sequencing Panel
- Pulmonary Disease: Comprehensive Sequencing Panel
- Bronchiectasis: Sequencing Panel
- Cystic Lung Disease: Sequencing Panel
- Pulmonary Fibrosis and Hermansky-Pudlak Syndrome: Sequencing Panel
- Pulmonary Hypertension: Sequencing Panel