

## Ciliopathies: Deletion/Duplication Panel

**Test Code:** DCIL1

**Turnaround time:** 2 weeks

**CPT Codes:** 81406 x1, 81403 x1, 81405 x1

### Condition Description

The ciliopathies are a group of disorders caused by mutations in genes that encode proteins involved in the formation and function of cilia. Cilia are microtubule-based, hair-like cytoplasmic extensions that extend from the cell surface. The cilium is a highly conserved organelle that is structurally complex with approximately 1000 different recognized polypeptides.

Cilia can be classified as either motile cilia or primary cilia (often called sensory cilia). Motile cilia, sometimes referred to as flagella, are typically found on epithelia cells that line the brain ventricles, oviducts, and respiratory tract. They can appear in bundles of 200-300 and can create movement of the extracellular fluid. Primary cilia are found on the surface of almost all cell types. They sense a wide variety of extracellular signals and transmit them to the interior of the cell. They are critical for developmental and physiological functions. Recent research suggests that motile cilia can be chemosensory as well.

Cilia are a component of almost all cells, so defects in the cilia can lead to conditions that have features involving multiple organ systems, such as renal disease, cerebral anomalies, and retinal degeneration. Additional features include diabetes, skeletal dysplasia, obesity, and congenital fibrocystic diseases of the pancreas and liver; however, the specific phenotype depends on the specific cilia involved.

Diseases tested by the panel include primary ciliary dyskinesia, nephronophthisis, Senior-Loken syndrome, Leber congenital amaurosis, Meckel-Gruber syndrome, Joubert and related syndromes, Bardet-Biedl syndrome, and many others. Please refer to the below list for all genes on the ciliopathies panel.

References:

- Ferkol and Leigh, (2011), *J Pediatr*, 160:366-371.
- Hildebrandt *et al.*, (2011), *New Engl J Med*, 364:1533-1543.
- Ware *et al.*, (2011), *Proc Am Thorac Soc*, 8:444-450.
- Waters and Beales, (2011), *Pediatr Nephrol*, 26:1039-1056.

### Genes

[ADGRV1](#), [AHI1](#), [AIPL1](#), [ARL13B](#), [ARL6](#), [ATXN10](#), [B9D1](#), [B9D2](#), [BBS1](#), [BBS10](#), [BBS12](#), [BBS2](#), [BBS4](#), [BBS5](#), [BBS7](#), [BBS9](#), [CC2D2A](#), [CCDC28B](#), [CCDC39](#), [CCDC40](#), [CDH23](#), [CEP290](#), [CFTR](#), [CLRN1](#), [CRB1](#), [CRX](#), [DNAAF1](#), [DNAAF2](#), [DNAH11](#), [DNAH5](#), [DNAI1](#), [DNAI2](#), [DNAL1](#), [DYNC2H1](#), [EVC](#), [EVC2](#), [FOXH1](#), [GLIS2](#), [GUCY2D](#), [HYLS1](#), [IFT43](#), [IFT80](#), [IMPDH1](#), [INVS](#), [IQCB1](#), [KCNJ13](#), [KIF7](#), [LCA5](#), [LRAT](#), [MKKS](#), [MKS1](#), [MYO7A](#), [NEK1](#), [NEK8](#), [NME8](#), [NODAL](#), [NPHP1](#), [NPHP3](#), [NPHP4](#), [OFD1](#), [PCARE](#), [PCDH15](#), [PKD2](#), [PKHD1](#), [RD3](#), [RDH12](#), [RPE65](#), [RPGR](#), [RPGRI1](#), [RPGRI1L](#), [RSPH4A](#), [RSPH9](#), [SDCCAG8](#), [SPATA7](#), [TCTN1](#), [TCTN2](#), [TMEM216](#), [TMEM67](#), [TOPORS](#), [TRIM32](#), [TSC1](#), [TSC2](#), [TTC21B](#), [TTC8](#), [TULP1](#), [UMOD](#), [USH1C](#), [USH1G](#), [USH2A](#), [VHL](#), [WDPCP](#), [WDR19](#), [WDR35](#), [WHRN](#), [XPNPEP3](#), [ZIC3](#)

### Indications

This test is indicated for:

- Individuals with a suspected ciliopathy.

### 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 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: 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/ $\mu$ L 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.

**Related Tests**

- Individual gene sequencing and deletion/duplication analysis is available for some genes on this panel.
- A comprehensive Eye Disorders Panel is also available.
- Custom diagnostic mutation analysis (KM) is available to family members if mutations are identified by targeted mutation testing or sequencing analysis.
- Ciliopathies: Sequencing Panel.