Ciliopathies: Deletion/Duplication Panel

Test Code: DCIL1
Turnaround time: 2 weeks
CPT Codes: 81403 x1, 81405 x1, 81406 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 epithelial 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:

Genes

ADGRV1, AHI1, AIP1L1, ARL13B, ARL6, ATXN10, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CCDC28B, CCDC39, CCDC40, CDH23, CEP290, CFT1, CLRN1, CRB1, CRX, DAAF1, DAAF2, DAAH1, DAAH5, DAAH6, DAAH12, DNL1, DYNC1H1, EVC, EVC2, FOXH1, GLI1, GLI2, GUCY2D, HPS1, IFT14, IFT30, IFTD1, INVS, IGF1R, KCNJ1, KIF7, LCAS, LRAT, MKKS, MKS1, MYO7A, NEK1, NEK8, NME8, NOdal, NPHP1, NPHP3, NPHP4, OP1D1, PCAR1, PCDAH15, PKD2, PKHD1, RASD1, RPE65, RPGR, RPGRIP1, RPGRIP1L, RPGRIP1L1, RSPH4A, RSPH8, SDC2CA58, SPATA7, TCTN1, TCTN2, TMEM216, TMEM73, TOPORS, TRIM32, TSC1, TSC2, TTC21B, TTC5, TULP1, UMOD, USH1G, USH1J, USH2A, VHL, WDPCP, WDR19, 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. 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

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.
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\textsuperscript{TM} 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

- 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.