In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic regions may also be analyzed, this assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and should not detect single or multi-exon deletions or duplications. Direct sequencing of the captured regions is performed using next generation sequencing. The patient's gene sequences are then compared to a standard reference sequence. Potentially causative variants and areas of low coverage are Sanger-sequenced. Sequence variations are classified as pathogenic, likely pathogenic, benign, likely benign, or variants of unknown significance. Variants of unknown significance may require further studies of the patient and/or family members.

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
ACVR2B, ADGRV1, AH11, AIP1, AR1L3B, AR1L6, ATXN10, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C2orf71, C5orf48, CC2D2A, CCDC28B, CCDC39, CCDC40, CDH23, CEP164, CEP290, CEP41, CFTR, CLRN1, CHB1, CRELD1, CRX, DNAAF1, DNAAF2, DNAF3, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DYNC2H1, EVC, EVC2, FOXH1, GDF1, GLIS2, GUCY2D, HVS1, IFT43, IFT80, IMPDH1, INVS, IQCB1, KCNJ13, KIF7, LCA5, LEFTY2, LRAT, MKKS, MKS1, MYO7A, NEK1, NEK8, NKC2-5, NME8, NODAL, NPHP1, NPHP3, NPHP4, OFD1, PCDH15, PKD2, PKHD1, RD3, RDH12, RPE65, RPRG, RPRGRI1, RPRGRI1L, RSPH4A, RSPH9, SCN11A, SCN1B1, SCN11G, SDCCAG8, SPATA7, TCTN1, TCTN2, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRIM32, TSC1, TSC2, TTC21B, TTC8, TULP1, UMOD, USH1C, USH1G, USH2A, VHL, WDPCP, WDR19, WDR35, WHRN, XPNPEP3, ZIC3, ZNF423

Indications
This test is indicated for:
- Individuals with a suspected ciliopathy.

Methodology

Next Generation Sequencing: In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic regions may also be analyzed, this assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and does not detect single or multi-exon deletions or duplications. Direct sequencing of the captured regions is performed using next generation sequencing. The patient's gene sequences are then compared to a standard reference sequence. Potentially causative variants and areas of low coverage are Sanger-sequenced. Sequence variations are classified as pathogenic, likely pathogenic, benign, likely benign, or variants of unknown significance. Variants of unknown significance may require further studies of the patient and/or family members.

Detection

Next Generation Sequencing: Clinical Sensitivity: Unknown. Mutations in the promoter region, some mutations in the introns and other regulatory element mutations cannot be detected by this analysis. Large deletions/duplications will not be detected by this analysis. Results of molecular analysis should be interpreted in the context of the patient's clinical/biochemical phenotype.

Analytical Sensitivity: ~99%.

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: 60 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/μl 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: Deletion/Duplication Panel.