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.

**Genes**

ACVR2B, ADGRV1, AH11, APL1, ARH13B, ARL6, ATXN10, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CDDC2B, CCDC40, CDH23, CEP164, CEP290, CEP41, CFTR, CLRN1, CLN5, COL2A1, CRX, DNAF1, DNAF2, DNAF3, DNAH11, DNAH5, DNAI1, DNAI2, DNL1, DYNC2H1, EVC, EVC2, FOXH1, GDF1, GLI2, GUCY2D, HYL5, ITF4, ITF5, IMPDH1, INVS, IQCB1, KCNJ13, KIF7, LCA5, LEFTY2, LRAT, MKKS, MKS1, MYO7A, NEK1, NEK8, NKB2-5, NME8, NODAL, NPHP1, NPHP3, NPHP4, OFD1, PCARE, PCDH15, PKD2, PKHD1, R3D, RPE65, RPRG, RPRGIP1, RPRGIP1L, RSPH4A, RSPH9, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SPATA7, TCTN1, TCTN2, TME13B, TME216, TME231, TME237, TME6, TOPORS, TRIM32, TSC1, TSC2, T2C12, T2C8, TULP1, UMOD, USH1C, USH1G, USH2A, VHL, WDPCP, WDR19, WDR3, 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/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: Deletion/Duplication Panel.