

## Nephronophthisis Gene Sequencing Panel (Test #660)

**Brief Description of Clinical Features:** Nephronophthisis (NPH) is the most common genetic cause of progressive renal failure in children and young adults. NPH is characterized by polyuria, growth retardation and progressive deterioration of renal function with normal or slightly reduced kidney size (Hildebrandt et al., Nat Genet 17:149-153, 1997; Hildebrandt et al. J Am Soc Nephrol 20:23-35, 2009). Nephronophthisis when associated with Leber Congenital Amaurosis is known as Senior-Loken syndrome (SLS; OMIM# 609294) (Otto et al. Nat Genet 37:282-288, 2005; Hildebrandt et al. 2009).

NPH clinical features overlap with a group of diseases known as ciliopathies, which includes Meckel-Gruber Syndrome (MKS) (OMIM 249000), Joubert Syndrome (OMIM 213300), Bradet-Biedl Syndrome (BBS; OMIM 209900), Nephronophthisis, Senior-Loken syndrome and Leber congenital amaurosis (LCA). For more information, see Hildebrandt et al. 2009.

**Genetics:** The Ciliopathies exhibit autosomal recessive inheritance. All Ciliopathies have high levels of locus heterogeneity. Ciliopathies are caused by mutations in genes encoding proteins involved in cilia/centrosome structure, maintenance or function (Hildebrandt et al. 2009). These disorders may represent a phenotypic continuum of a single clinical entity.

**Description of This Particular Test:** The following genes are tested in the order specified by the client. Testing is accomplished through bidirectional sequencing of all coding exons along with ~50 bases of non coding flanking DNA on each side. See also the individual Test Descriptions for each gene.

### Reference Sequences:

Gene	Disease	Genomic: NC_	mRNA: NM_	Protein: NP_	CCDS:
<i>NPHP1</i>	NPH and JS	000002.10	000272.2	000263.2	2086.1
<i>INVS/NPHP2</i>	NPH	000009.10	014425.2	055240.2	6746.1
<i>NPHP3</i>	NPH	000003.10	153240.3	694972.3	3078.1
<i>NPHP4</i>	NPH	000001.9	015102.2	055917.1	
<i>IQCBI/NPHP5</i>	NPH and SLNS	000003.11	001023570.2	001018864.2	
<i>CEP290/NPHP6</i>	NPH, JS and MKS	000012.11	025114.3	079390.3	
<i>GLIS2/NPHP7</i>	NPH	000016.8	032575.2	115964.2	10511.1
<i>RPGRIPI1/NPHP8</i>	NPH, JS and MKS	000016.8	015272.2	056087.2	
<i>NEK8/NPHP9</i>	NPH	00017.10	178170.2	835464.1	32597.1
<i>SDCCAG8</i>	NPH, SLSN7 and BBS	000001.10	006642.3	006633.1	31075.1

**Indication for Testing:** Candidates for this test are patients with symptoms consistent with NPH.

**Sensitivity of Test:** Sensitivity for NPH testing is approximately 30% overall, indicating that the NPH causative gene in approximately 70% of the clinically diagnosed cases is still unknown (Hildebrandt et al. 2009).

**Turnaround Time:** Maximum of 80 days.

**Specimen Requirements:** See page 4 of the Requisition Form.

### CPT Codes and Price

Codes	<i>NPHP1</i> Sequencing	<i>INVS</i>	<i>NPHP3</i>	<i>NPHP4</i>	<i>IQCBI</i>	<i>CEP290</i>	<i>GLIS2</i>	<i>RPGRIPI1</i>	<i>NEK8</i>	<i>SDCCAG8</i>	Panel
83890	\$ 30	\$ 30	\$ 30	\$ 30	\$ 30	\$ 30	\$ 30	\$ 30	\$ 30	\$ 30	\$ 30
83891	\$ 40	\$ 40	\$ 40	\$ 40	\$ 40	\$ 40	\$ 40	\$ 40	\$ 40	\$ 40	\$ 40
83898	\$330	\$280	\$420	\$440	\$200	\$ 755	\$140	\$380	\$230	\$ 320	\$3420
83904	\$510	\$420	\$630	\$640	\$310	\$1245	\$200	\$580	\$350	\$ 470	\$5140
83894	\$ 70	\$ 70	\$ 80	\$80	\$60	\$ 40	\$ 50	\$ 70	\$ 70	\$ 60	\$570
83912	\$ 110	\$100	\$120	\$120	\$100	\$ 80	\$ 80	\$ 90	\$120	\$ 120	\$350
Totals:	\$1090	\$940	\$1320	\$1350	\$740	\$2190	\$540	\$1190	\$840	\$1040	\$9550*

\*When five or more of the genes are tested, 15% discount will apply to the total cost.

**Accreditation:** CLIA ID:52D1027685 (expires 1/18/13) CAP ID:7185561, AU ID:1407125 (expires 12/20/12)

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