

Meckel-Gruber Syndrome Gene Sequencing Panel (Test #295)

Brief Description of Clinical Features: Meckel-Gruber syndrome (MKS) (OMIM 249000) is characterized by occipital encephalocele, polycystic kidneys, hepatic developmental defects and postaxial polydactyly (Alexiev et al. Arch Pathol Lab Med 130:1236-1238, 2006). MKS is a common cause of prenatal echogenic kidneys (Chaumoitre et al. Ultrasound Obstet Gynecol 28:911-917, 2006). Nearly all MKS infants are stillborn or die shortly after birth.

MKS clinical features overlap with a group of diseases known as ciliopathies, which include MKS, Joubert syndrome (OMIM 213300), Bardet-Biedl syndrome (OMIM# 209900), Nephronophthisis (OMIM 256100), Senior-Loken syndrome (OMIM# 609294) and Leber congenital amaurosis. For more information, see Hildebrandt et al. J Am Soc Nephrol 20:23-35, 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 will be tested in the order specified in the Table below, unless different order has been requested by the client. Testing is accomplished by amplifying and sequencing the coding exons and ~50 bp of adjacent non-coding sequence.

Reference Sequences:

Gene	Disease	Genomic: NC_	mRNA: NM_	Protein: NP_
<i>MKS1</i>	MKS	000017.10	017777.2	060247.2
<i>TMEM67 (MKS3)</i>	MKS and JS	000008.9	153704.4	714915.3
<i>CC2D2A</i>	MKS and JS	000004.10	001080522.2	001073991.2
<i>RPGRIP1L</i>	MKS and JS	000016.8	015272.2	056087.2
<i>CEP290</i>	MKS and JS	000012.11	025114.3	079390.3

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

Sensitivity of Test: Sensitivity for MKS testing is at least 50% overall.

Turn Around Time: Maximum of 80 days.

Specimen Requirements: See page 4 of the Requisition Form.

CPT Codes and Prices:

Codes	<i>MKS1</i>	<i>TMEM67</i>	<i>CC2D2A</i>	<i>RPGRIP1L</i>	<i>CEP290</i>	Panel
83890	\$ 30	\$ 30	\$ 30	\$ 30	\$ 30	\$ 30
83891	\$ 40	\$ 40	\$ 40	\$ 40	\$ 40	\$ 40
83898	\$250	\$380	\$530	\$380	\$ 755	\$2145
83904	\$380	\$670	\$790	\$580	\$1245	\$3220
83894	\$ 50	\$ 60	\$ 80	\$ 70	\$ 40	\$300
83912	\$90	\$ 110	\$ 120	\$ 90	\$ 80	\$300
Totals:	\$840	\$1290	\$1590	\$1190	\$2190	\$6035*

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

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

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