

Joubert and Meckel-Gruber Syndromes via *RPGRIP1L* Gene Sequencing (Test #276)

Brief Description of Clinical Features: Joubert syndrome (JS) (OMIM 213300) is marked by ataxia, hypotonia, abnormal ocular movements, apraxia, neonatal respiratory anomalies, mental retardation, agenesis/hypoplasia of the cerebellar vermis and a brain malformation known as the "molar tooth sign" (MTS) on cranial MRI. JS patients have substantial phenotypic variation. MTS is considered to be the most characteristic diagnostic feature. Some JS patients develop retinal dystrophy and/or progressive renal failure. For more information, see Parisi and Glass (Gene Reviews, www.genetests.org, 2007).

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.

Genetics: Joubert and Meckel-Gruber Syndromes both exhibit autosomal recessive inheritance.

Description of This Particular Test: This test involves bidirectional sequencing using genomic DNA of all 26 coding exons (exons 2-27) of the *RPGRIP1L* gene. The full coding region of each exon plus ~50 bp of flanking non-coding DNA on either side are sequenced. We will also perform sequencing of any single or pair of exons for family members of patients with known mutations and to confirm previous results (\$190-340).

Reference Sequences: Genomic: NC_000016.8 mRNA: NM_015272.2 Protein: NP_056087.2

Indications for Test:

Sensitivity of Test:

Turn Around Time: Maximum of 40 days.

Specimen Requirements: See page 4 of the Requisition Form.

Price: Sequencing of *RPGRIP1L* Gene **\$ 1190**

CPT Codes:

Sample Ascertainment	83890	\$ 30	DNA Isolation	83891	\$ 40
Amplification x26	83898	\$ 380	Sequencing x26	83904	\$ 580
Separation	83894	\$ 70	Interpretation/Report	83912	\$ 90

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

Contact: Dr. Keith Nykamp, keith.nykamp@preventiongenetics.com, www.preventiongenetics.com