

Meckel-Gruber Syndrome

Precision Panel



Overview

Meckel-Gruber Syndrome (MKS) is a lethal, autosomal recessive neurodevelopmental condition characterized by a triad of symptoms which are occipital encephalocele, large polycystic kidneys and postaxial polydactyly. It is caused by mutations in genes encoding proteins that allow an appropriate structure and function of the primary cilium. It belongs to a group of diseases known as ciliopathies, and since cilia are present in a variety of organs in the human organism it has several other manifestations. Associated abnormalities include oral clefting, genital anomalies, CNS alterations and liver fibrosis. The leading cause of death is pulmonary hypoplasia.

The Igenomix Meckel-Gruber Syndrome Precision Panel can be used to make a directed and accurate diagnosis ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved.

Indications

The Igenomix Meckel-Gruber Syndrome Precision Panel is indicated for those patients with a clinical suspicion or diagnosis of Meckel-Gruber Syndrome presenting with:

- CNS abnormalities: occipital encephalocele, Dandy-Walker malformation, hydrocephalus, Arnold-Chiari malformation, microcephaly
- Polycystic kidneys
- Polydactyly
- Hepatic fibrosis
- Cardiac malformations: atrial septal defect, coarctation of aorta and pulmonary stenosis
- Cleft lip and palate
- Genital anomalies

Clinical Utility

The clinical utility of this panel is:

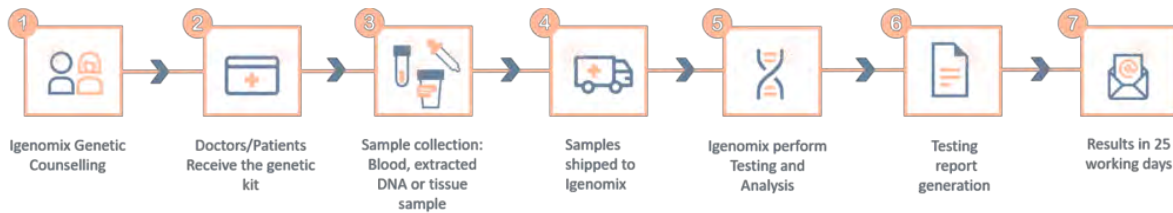
- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team in the form of surgical repair of anatomic abnormalities.
- Risk assessment of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
<i>AHI1</i>	Joubert Syndrome	AR	96.79	85 of 97
<i>ARL13B</i>	Joubert Syndrome	AR	99.77	10 of 10
<i>ARL3</i>	Joubert Syndrome	AD,AR	99.99	4 of 4
<i>ARMC9</i>	Joubert Syndrome	AR	99.95	10 of 10
<i>B9D1</i>	Joubert Syndrome, Meckel Syndrome	AR	90.23	11 of 11
<i>B9D2</i>	Meckel Syndrome	AR	84.81	4 of 5
<i>CC2D2A</i>	Coach Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.43	98 of 100
<i>CEP104</i>	Joubert Syndrome	AR	99.89	9 of 9
<i>CEP120</i>	Joubert Syndrome, Short-Rib Thoracic Dysplasia With Or Without Polydactyly, Jeune Syndrome	AR	99.8	9 of 9
<i>CEP290</i>	Bardet-Biedl Syndrome, Joubert Syndrome, Meckel Syndrome, Senior-Loken Syndrome	AR	96.47	293 of 327
<i>CEP41</i>	Joubert Syndrome	AR	100	17 of 17
<i>CEP55</i>	Multinucleated Neurons, Anhydramnios, Renal Dysplasia, Cerebellar Hypoplasia, And Hydranencephaly, Meckel Syndrome	AR	99.22	3 of 3
<i>CPLANE1</i>	Joubert Syndrome, Varadi-Papp Syndrome, Monomelic Amyotrophy, Orofaciodigital Syndrome	AR	na	na
<i>CSPP1</i>	Joubert Syndrome, Meckel Syndrome	AR	98.32	29 of 30
<i>FAM149B1</i>	Joubert Syndrome, Orofaciodigital Syndrome	AR	99.94	2 of 2
<i>IFT172</i>	Retinitis Pigmentosa, Short-Rib Thoracic Dysplasia With Or Without Polydactyly, Bardet-Biedl Syndrome, Jeune Syndrome	AR	100	37 of 37
<i>INPP5E</i>	Joubert Syndrome, Mental Retardation, Truncal Obesity, Retinal Dystrophy, And Micropenis, Joubert Syndrome	AR	99.89	56 of 56
<i>KATNIP</i>	Joubert Syndrome	AR	99.97	7 of 7
<i>KIAA0586</i>	Joubert Syndrome, Short-Rib Thoracic Dysplasia With Polydactyly	AR	99.84	31 of 32
<i>KIF14</i>	Meckel Syndrome, Autosomal Recessive Primary Microcephaly	AR	99.84	18 of 18
<i>KIF7</i>	Acrocallosal Syndrome, Hydroletharus Syndrome, Macrocephaly With Multiple Epiphyseal Dysplasia And Distinctive Facies, Orofaciodigital Syndrome	AR	94.91	47 of 50
<i>MKS1</i>	Bardet-Biedl Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.98	49 of 49
<i>NPHP1</i>	Joubert Syndrome, Nephronophthisis, Senior-Loken Syndrome, Bardet-Biedl Syndrome	AR	100	58 of 59
<i>NPHP3</i>	Meckel Syndrome, Nephronophthisis, Renal-Hepatic-Pancreatic Dysplasia , NPHP3-Related Meckel-like Syndrome, Senior-Loken Syndrome	AR	99.99	84 of 84
<i>OFD1</i>	Joubert Syndrome, Orofaciodigital Syndrome, Retinitis Pigmentosa, Simpson-Golabi-Behmel Syndrome, Primary Ciliary Dyskinesia	X,XR,XD,G	98.09	NA of NA
<i>PDE6D</i>	Joubert Syndrome, Orofaciodigital Syndrome	AR	100	2 of 2
<i>PIBF1</i>	Joubert Syndrome	AR	99.83	7 of 7
<i>RPGRIP1L</i>	Coach Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.96	52 of 52
<i>SUFU</i>	Joubert Syndrome, Medulloblastoma, Acrocallosal Syndrome, Gorlin Syndrome, Microform Holoprosencephaly	AD,AR	99.99	43 of 43
<i>TCTN1</i>	Joubert Syndrome	AR	94.98	10 of 10
<i>TCTN2</i>	Joubert Syndrome, Meckel Syndrome	AR	100	14 of 14
<i>TCTN3</i>	Joubert Syndrome, Orofaciodigital Syndrome	AR	99.99	13 of 13
<i>TMEM107</i>	Meckel Syndrome, Orofaciodigital Syndrome	AR	100	3 of 3
<i>TMEM138</i>	Joubert Syndrome	AR	99.94	9 of 9
<i>TMEM216</i>	Joubert Syndrome, Meckel Syndrome, Orofaciodigital Syndrome	AR	98.74	8 of 8
<i>TMEM231</i>	Joubert Syndrome, Meckel Syndrome, Orofaciodigital Syndrome	AR	98.63	20 of 21
<i>TMEM237</i>	Joubert Syndrome	AR	100	11 of 11
<i>TMEM67</i>	Bardet-Biedl Syndrome, Coach Syndrome, Joubert Syndrome, Meckel Syndrome, Nephronophthisis, Rhys Syndrome	AR	96.93	177 of 179
<i>TTC21B</i>	Asphyxiating Thoracic Dystrophy, Nephronophthisis, Joubert Syndrome, Jeune Syndrome	AD,AR	100	67 of 67
<i>WDPCP</i>	Bardet-Biedl Syndrome, Congenital Heart Defects, Hamartomas Of Tongue, And Polysyndactyly, Meckel Syndrome	AR	99.3	8 of 8

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant mutations according to HGMD

Methodology



Contact us

Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

References

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