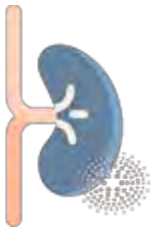
Meckel-Gruber Syndrome

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

TTC21B	Asphyxiating Thoracic Dystrophy, Nephronophthisis, Joubert Syndrome, Jeune Syndrome	AD,AR	100	67 of 67
WDPCP	Bardet-Biedl Syndrome, Congenital Heart Defects, Hamartomas Of Tongue, And Polysyndactyly, Meckel Syndrome	AR	99.3	8 of 8
ZNF423	Nephronophthisis, Joubert Syndrome	AD,AR	100	10 of 10

*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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