Joubert Syndrome and Related Disorders

Precision Panel

Overview

Joubert Syndrome (JS) and Related Disorders (JSRD) are a group of ciliopathies characterized by mid-hindbrain malformation, developmental delay, hypotonia, oculomotor apraxia, and breathing abnormalities. Cilia play a crucial role in appropriate axonal growth and connectivity which are essential for functional wiring of the brain. The classic midbrain-hindbrain malformation is a hallmark image finding known as molar tooth sign. Joubert Syndrome and Related Disorders are a group of clinically and genetically heterogeneous disorders involving ciliopathy-related genes. Therefore, clinical manifestations have multiorgan involvement, mainly retinal dystrophy, hepatic fibrosis and polydactyly, among others. With the exception of rare X-linked recessive cases, Joubert Syndrome and Related Disorders follow an autosomal recessive inheritance pattern.

The Igenomix Joubert Syndrome and Related Disorders Precision Panel can serve as a screening and diagnostic tool 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 Joubert Syndrome and Related Disorders Precision Panel is indicated for those patients with clinical and/or imaging findings suggestive of Joubert Syndrome and Related Disorders presenting with the following manifestations:

- Hypotonia
- Ataxia
- Developmental delay
- Abnormal eye and tongue movements
- Respiratory control disturbances
- Polydactyly
- Cleft lip or palate
- Seizures

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 involving a multidisciplinary team focusing on respiratory and feeding problems in neonates and infants. Cognitive and behavioral assessments with adequate neuropsychological rehabilitation.
- Risk assessment of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.
- Identification of molecular defect in couples at risk allowing prenatal genetic tasting where neuroimaging may be uninformative early in the pregnancy.

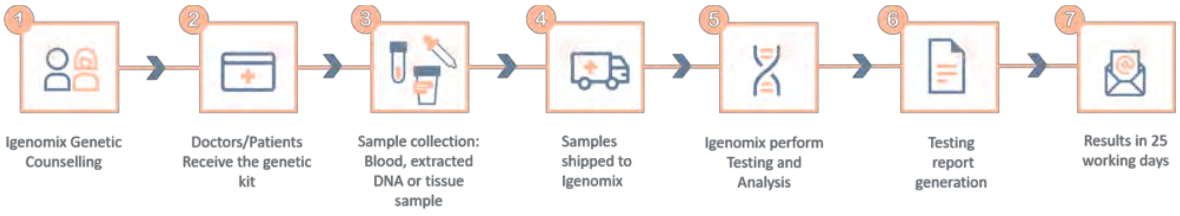
Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20x)	HGMD**
AHI1	Joubert Syndrome With Ocular Defect, Retinitis Pigmentosa	AR	96.79	85 of 97
ARL13B	Joubert Syndrome	AR	99.77	10 of 10
ARL3	Joubert Syndrome, Retinitis Pigmentosa	AD,AR	99.99	4 of 4
ARMC9	Joubert Syndrome	AR	99.95	10 of 10
B9D1	Joubert Syndrome, Meckel Syndrome Type 9	AR	90.23	11 of 11
B9D2	Meckel Syndrome Type 10	AR	84.81	4 of 5
C2CD3	Orofaciodigital Syndrome	AR	97.25	18 of 18
CC2D2A	Coach Syndrome, Meckel Syndrome Type 6, Joubert 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
CEP164	Nephronophthisis, Senior-Loken Syndrome	AR	99.98	10 of 10
CEP290	Bardet-Biedl Syndrome, Joubert Syndrome, Leber Congenital Amaurosis, Meckel Syndrome Type 4, Senior-Loken Syndrome	AR	96.47	293 of 327
CEP41	Joubert Syndrome	AR	100	17 of 17
CFAP410	Retinal Dystrophy With or Without Macular Staphyloma, Axial Spondylometaphyseal Dysplasia, Amyotrophic Lateral Sclerosis, Cone-Rod Dystrophy	AR	-	-
CPLANE1	Joubert Syndrome, Varadi-Papp Syndrome, Monomelic Amyotrophy, Orofaciodigital Syndrome Type 6	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
HYLS1	Hydrolethalus Syndrome, Joubert Syndrome	AR	100	2 of 2
INPP5E	Joubert Syndrome, Mental Retardation	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
KIAA0753	Orofaciodigital Syndrome	AR	97.73	7 of 7
KIF7	Acrocallosal Syndrome, Hydrolethalus Syndrome, Macrocephaly, Multiple Epiphyseal Dysplasia, Orofaciodigital Syndrome	AR	94.91	47 of 50
MKS1	Bardet-Biedl Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.98	49 of 49
NPHP1	Nephronophthisis, Senior-Loken Syndrome, Bardet-Biedl Syndrome, Joubert Syndrome With Renal Defect	AR	100	58 of 59
NPHP3	Meckel Syndrome, Nephronophthisis, Renal-Hepatic-Pancreatic Dysplasia, 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 Type 6	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	Basal Cell Nevus Syndrome, 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 Type 6	AR	98.74	8 of 8
TMEM231	Joubert Syndrome; Meckel Syndrome; Orofaciodigital Syndrome Type 3	AR	98.63	20 of 21
TMEM237	Joubert Syndrome	AR	100	11 of 11
TMEM67	Bardet-Biedl Syndrome 14; Coach Syndrome; Joubert Syndrome 6; Meckel Syndrome, Type 3; Nephronophthisis 11; Rhyns Syndrome	AR	96.93	177 of 179
TTC21B	Asphyxiating Thoracic Dystrophy 4; Nephronophthisis 12; Joubert Syndrome 11; Jeune Syndrome	AD,AR	100	67 of 67
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



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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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