Bardet-Biedl Syndrome
Precision Panel

Overview

Bardet-Biedl Syndrome (BBS) is an inherited disease belonging to the group of disorders called ciliopathies, where there is a defect in primary cilia which plays a key role in sensory perception and various signalling pathways. It is a pleiotropic genetic disorder where patients typically present with truncal obesity, intellectual impairment as well as kidney, eye and genitalia anomalies. Most of these symptoms may not be present at birth but appear and progressively worsen during the first and second decades of life. This disorder is clinically and genetically heterogenous with an array of clinical manifestations. It shows an autosomal recessive inheritance and is highly prevalent in consanguineous populations.

The Igenomix Bardet-Biedl Syndrome Precision Panel can serve as a directed diagnostic tool in making a differential diagnosis of ciliopathies 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.

Indications

The Igenomix Bardet-Biedl Syndrome Precision Panel is indicated in those cases where there is a clinical suspicion or diagnosis of BBS and/or the following manifestations:

- Truncal obesity
- Intellectual impairment
- Polydactyly
- Diabetes mellitus type 2, non-insulin dependent
- Night blindness
- Tunnel vision
- Loss of smell
- Small testicular size
- Hydronephrosis (large sized kidneys)

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular diagnosis for an accurate clinical diagnosis and improve prognosis.

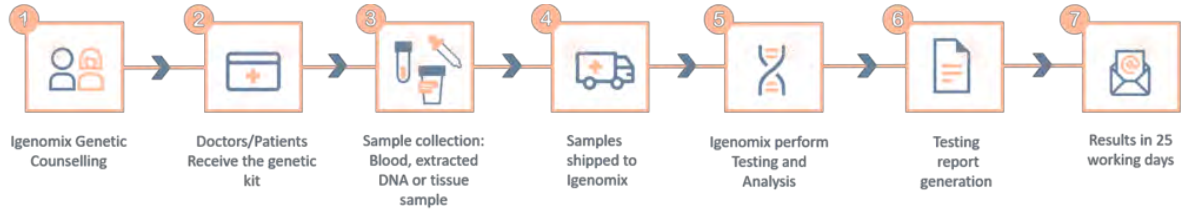
- Early initiation of treatment with a multidisciplinary team in the form of orthopaedic surgical care, appropriate weight reducing strategies, regular surveillance for renal function and early ophthalmology referral.
- Risk assessment and genetic counselling of asymptomatic family members to identify the individuals at risk.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ARL6	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AD,AR,X,XR,G	100	17 of 21
BBIP1	Bardet-Biedl Syndrome	AR	99.88	1 of 1
BBS1	Bardet-Biedl Syndrome	AR	100	102 of 105
BBS10	Bardet-Biedl Syndrome	AR	100	114 of 114
BBS12	Bardet-Biedl Syndrome	AR	99.78	61 of 61
BBS2	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AR	100	99 of 100
BBS4	Bardet-Biedl Syndrome	AR	100	45 of 48
BBS5	Bardet-Biedl Syndrome	AR	99.8	30 of 31
BBS7	Bardet-Biedl Syndrome	AR	100	48 of 48
BBS9	Bardet-Biedl Syndrome	AR	99.56	50 of 51
C8ORF37	Bardet-Biedl Syndrome, Cone-Rod Dystrophy, Retinitis Pigmentosa	AD,AR,X,XR,G	na	na
CCDC28B	Bardet-Biedl Syndrome	AR	99.83	1 of 1
CEP19	Morbid Obesity And Spermatogenic Failure	AR	99.88	2 of 2
CEP290	Bardet-Biedl Syndrome, Joubert Syndrome, Leber Congenital Amaurosis, Meckel Syndrome Type 4, Senior-Loken Syndrome	AR	96.47	293 of 327
CPE	Obesity, Type 1 Diabetes Mellitus	-	96.28	0 of 1
IFT172	Retinitis Pigmentosa, Short-Rib Thoracic Dysplasia With Or Without Polydactyly, Bardet-Biedl Syndrome, Jeune Syndrome	AR	100	37 of 37
IFT27	Bardet-Biedl Syndrome	AR	100	5 of 5
IFT74	Bardet-Biedl Syndrome	AR	99.95	6 of 6
LZTFL1	Bardet-Biedl Syndrome	AR	99.83	4 of 4
MKKS	Bardet-Biedl Syndrome, Mckusick-Kaufman Syndrome	AR	89.96	71 of 71
MKS1	Bardet-Biedl Syndrome, Joubert Syndrome, Meckel Syndrome Type 1	AR	99.98	49 of 49
NPHP1	Joubert Syndrome, Nephronophthisis, Senior-Loken Syndrome, Bardet-Biedl Syndrome	AR	100	58 of 59
SCAPER	Intellectual Developmental Disorder And Retinitis Pigmentosa, Retinitis Pigmentosa	AR	99.92	17 of 18
SDCCAG8	Bardet-Biedl Syndrome, Senior-Loken Syndrome	AR	96.29	18 of 19
TMEM67	Bardet-Biedl Syndrome, Coach Syndrome, Joubert Syndrome, Meckel Syndrome Type 3, Nephronophthisis, Rhyns Syndrome	AR	96.93	177 of 179
TRIM32	Bardet-Biedl Syndrome, Limb-Girdle Muscular Dystrophy Type 2h	AR	100	17 of 17
TTC8	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AR	99.33	28 of 28
WDPCP	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
** HGMD: 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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