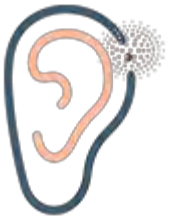
Branchiootorenal Syndrome

Precision Panel



Overview

Branchiootorenal Syndrome (BOR) is a rare autosomal dominant disorder that disrupts the development of tissues in the neck and causes malformations of the ears and kidneys. It is characterized by branchial arch anomalies, hearing loss and renal anomalies ranging from hypoplasia to bilateral renal agenesis. Some individuals progress to end-stage renal disease (ESRD) later in life. It is a clinically heterogeneous disorder with variability in the presence, severity and type of branchial arch, otologic, audiologic and renal abnormality.

The Igenomix Branchiootorenal Syndrome Precision Panel can be used to make an accurate and directed 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 Branchiootorenal Syndrome Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Deafness
- Preauricular pits
- Auricular malformations
- Renal anomalies
- Branchial cleft cyst and fistula
- Long, narrow face
- Cleft palate

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 surgical care of anatomic defects, surveillance for hearing impairment and renal function and prevention of complications such as infections.
- Risk assessment and genetic counselling of asymptomatic family members due to the autosomal dominant mode of inheritance.

- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
EYA1	Branchiootic Syndrome, Branchiootorenal Syndrome, Otofaciocervical Syndrome	AD	100	197 of 199
SIX1	Branchiootic Syndrome, Branchiootorenal Syndrome, Autosomal Dominant Deafness	AD	73	20 of 20
SIX5	Branchiootorenal Syndrome	AD	93.16	11 of 11
TFAP2A	Branchiooculofacial Syndrome	AD	98.61	37 of 37

*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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