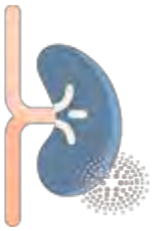
Bartter Syndrome

Precision Panel



Overview

Bartter Syndrome is an autosomal recessive renal tubular disorder caused by a defective salt reabsorption in the thick ascending loop of Henle resulting in hypokalemia, hypochloremia, metabolic alkalosis, high renin and aldosterone with normal blood pressure. In neonatal cases, it can be suspected before birth and diagnosed soon after birth whereas in classic cases the presentation can begin at 2 years of age or younger. The genetic heterogeneity of this disease comes from genetic mutations in either the sodium chloride/potassium chloride cotransporter or the potassium channel transporter in the thick ascending loop of Henle.

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

- Family history of Bartter Syndrome
- Maternal polyhydramnios
- Fetal fluid loss and volume depletion
- Failure to thrive
- Polyuria
- Polydipsia
- Vomiting
- Constipation
- Salt craving

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.

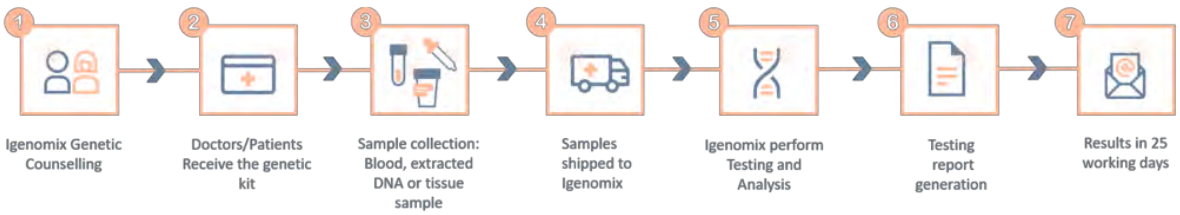
- Early initiation of treatment with a multidisciplinary team in the form of medical care with potassium sparing diuretics, ion supplements and counselling regarding pregnancy-related considerations.
- Risk assessment of asymptomatic family members according to the mode of inheritance.
- Translation of genomic-informed medicine allowing for a better phenotype-genotype relationship.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	%GENE COVERAGE (20X)	HGMD**
BSND	Bartter Syndrome Infantile With Sensorineural Deafness	AR	99.95	21 of 21
CASR	Neonatal Severe Primary Hyperparathyroidism, Autosomal Dominant Hypocalcemia, Hypocalciuric Familial Hypercalcemia, Hereditary Chronic Pancreatitis	AD,AR	100	445 of 446
CLCNKA	Bartter Syndrome		99.93	5 of 5
CLCNKB	Bartter Syndrome, Gitelman Syndrome	AR	99.86	145 of 145
GJB2	Autosomal Dominant Nonsyndromic Sensorineural Deafness With Keratopachydermia And Constrictions Of Fingers And Toes, Keratitis-Ichthyosis-Deafness Syndrome, Hereditarium Mutilans And Palmoplantar Keratoderma, Kid Syndrome	AD,AR,X,XR,MU,D,G	99.89	413 of 419
HBA1	Alpha-Thalassemia, Heinz Body Anemias, Hemoglobin H Disease, Hbh, Alpha-Thalassemia-Intellectual Disability Syndrome Linked To Chromosome 16, Hb Bart's Hydrops Fetalis	AD	98.87	125 of 152
HBA2	Alpha-Thalassemia, Heinz Body Anemias, Hemoglobin H Disease, Alpha-Thalassemia-Intellectual Disability Syndrome, Hb Bart's Hydrops Fetalis	AD	74.46	118 of 231
KCNJ1	Antenatal Bartter Syndrome	AR	100	67 of 67
MAGED2	Antenatal Transient Bartter Syndrome	X,XR,G	96.97	NA of NA
RIPK4	Lethal Type Popliteal Pterygium Syndrome; Bartsocas-Papas Syndrome, Chand Syndrome	AR	99.98	16 of 16
SLC12A1	Bartter Syndrome	AR	99	90 of 95
TAZ	Barth Syndrome, Familial Isolated Dilated Cardiomyopathy	X,XR,G	100	NA of NA

*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

1. Cunha, T., & Heilberg, I. P. (2018). Bartter syndrome: causes, diagnosis, and treatment. *International journal of nephrology and renovascular disease*, *11*, 291–301. https://doi.org/10.2147/IJNRD.S155397
2. Bao, M., Cai, J., Yang, X., & Ma, W. (2019). Genetic screening for Bartter syndrome and Gitelman syndrome pathogenic genes among individuals with hypertension and hypokalemia. *Clinical and experimental hypertension (New York, N.Y. : 1993)*, *41*(4), 381–388. https://doi.org/10.1080/10641963.2018.1489547
3. Mrad, F., Soares, S., de Menezes Silva, L., Dos Anjos Menezes, P. V., & Simões-E-Silva, A. C. (2021). Bartter's syndrome: clinical findings, genetic causes and therapeutic approach. *World journal of pediatrics : WJP*, *17*(1), 31–39. https://doi.org/10.1007/s12519-020-00370-4
4. Cunha, T., & Heilberg, I. (2018). Bartter syndrome: causes, diagnosis, and treatment. *International Journal Of Nephrology And Renovascular Disease*, *Volume 11*, 291-301. doi: 10.2147/ijnrd.s155397
5. Seys, E., Andrini, O., Keck, M., Mansour-Hendili, L., Courand, P., & Simian, C. et al. (2017). Clinical and Genetic Spectrum of Bartter Syndrome Type 3. *Journal Of The American Society Of Nephrology*, *28*(8), 2540-2552. doi: 10.1681/asn.2016101057
6. Daniluk, U., Kaczmarski, M., Wasilewska, J., Matuszewska, E., Semeniuk, J., Sidor, K., & Krasnow, A. (2004). Zespól Barttera [Bartter's syndrome]. *Polski merkuriusz lekarski : organ Polskiego Towarzystwa Lekarskiego*, *16*(95), 484–489.