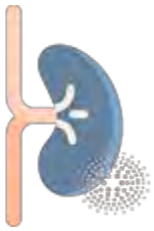
Liddle Syndrome

Precision Panel



Overview

Liddle syndrome is a genetic disorder characterized by low-renin hypertension that appears early in life. It is caused by mutations affecting the epithelial sodium channel (ENaC) located in the collecting duct of the nephron. The most common presentation of this disease is early onset hypertension, hypokalemia, metabolic alkalosis with suppressed plasma renin activity and low plasma aldosterone. Despite this typical phenotype, the disease can be clinically heterogeneous, even with mild phenotypes. It is transmitted in an autosomal dominant pattern.

The Igenomix Liddle Syndrome Precision Panel can be used to make a directed differential diagnosis of resistant hypertension 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 Liddle Syndrome Precision Panel is indicated for those patients with a clinical suspicion or diagnosis of Liddle Syndrome presenting with:

- Family history of early onset hypertension or hypokalemia
- Hypertension at early age
- Hypokalemia
- Metabolic alkalosis

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 and surveillance for complications of hypertension.
- 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**
SCNN1A	Bronchiectasis With Or Without Elevated Sweat Chloride, Liddle Syndrome, Pseudohypoaldosteronism, Brugada Syndrome	AD,AR	99.95	46 of 46
SCNN1B	Bronchiectasis, Liddle Syndrome, Pseudohypoaldosteronism	AD,AR	100	56 of 56
SCNN1G	Bronchiectasis With Or Without Elevated Sweat Chloride, Liddle Syndrome, Pseudohypoaldosteronism	AD,AR	100	28 of 28

*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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