Congenital Adrenal Hyperplasia

Precision Panel

Overview

Congenital Adrenal Hyperplasia (CAH) is a group of diseases that are associated with variants or mutations in the genes that are involved in hormone production. Specifically, CAH occurs when there is an excess of hormones in the adrenal gland. The classical form presents with prenatal onset of virilization caused by enzyme deficiency. Additionally, there is an imbalance of body hormones which could have severe effects throughout and individual's lifetime including problems with fertility, obesity and hypertension. The nature of the genetic variants and genes associated with CAH means that testing for the condition is not straight forward. CAH can be tested for in three ways – CYP21A2 gene sequencing, CYP21A2 del/dup by MLPA and a gene panel associated with CAH not including the CYP21A2 gene. This condition is associated with recessive inheritance and is usually diagnosed at birth.

The Igenomix Congenital Adrenal Hyperplasia Precision Panel can be used to make a directed and accurate differential diagnosis of ambiguous genitalia 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 Congenital Adrenal Hyperplasia Precision Panel is indicated for those patients with clinical suspicion of an CAH presenting with the following manifestations:

- Excessive hormone development
- Ambiguous genitalia
- Cortisol deficiency
- Infertility
- Obesity
- Hypertension

Clinical Utility

The clinical utility of this panel is:

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

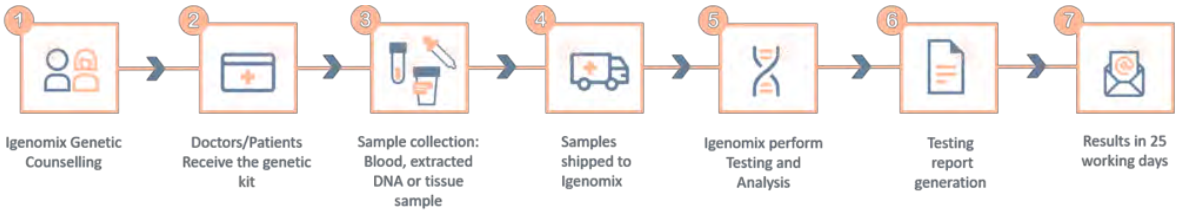
- Early initiation of treatment with a multidisciplinary team for pharmacologic treatment of lacking hormones, corrective surgery, specialist referral and dietary modifications.
- Risk assessment of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ARMC5	ACTH-Independent Macronodular Adrenal Hyperplasia, Cushing Syndrome Due To Macronodular Adrenal Hyperplasia	AD	99.97%	52 of 53
CYP11A1	46,XY Disorder Of Sex Development-Adrenal Insufficiency Due To Cyp11A1 Deficiency, Congenital Adrenal Insufficiency		100%	39 of 39
CYP11B1	Congenital Adrenal Hyperplasia Due To Steroid 11-Beta-Hydroxylase Deficiency,Familial Hyperaldosteronism Type I, Glucocorticoid-Remediable Aldosteronism	AD,AR	100%	144 of 144
CYP11B2	Corticosterone Methyloxidase Type I Deficiency, Corticosterone Methyloxidase Type II Deficiency, Familial Hyperaldosteronism Type I, Familial Hypoaldosteronism	AR	100%	42 of 42
CYP17A1	46,XY Disorder Of Sex Development Due To Isolated 17,20-Lyase Deficiency, Congenital Adrenal Hyperplasia Due To 17-Alpha-Hydroxylase Deficiency	AR	100%	127 of 127
CYP21A2	Congenital Adrenal Hyperplasia Due To 21-Hydroxylase Deficiency	AR	99.98%	278 of 280
HSD3B2	Congenital Adrenal Hyperplasia Due To 3-Beta-Hydroxysteroid Dehydrogenase 2 Deficiency, Congenital Adrenal Hyperplasia Due To 3-Beta-Hydroxysteroid Dehydrogenase Deficiency	AR	100%	70 of 70
PDE11A	Pigmented Nodular Adrenocortical Disease, Primary Pigmented Nodular Adrenocortical Disease	AD	99.98%	17 of 17
PDE8B	Autosomal Dominant Striatal Neurodegeneration, Pigmented Nodular Adrenocortical Disease, Primary Pigmented Nodular Adrenocortical Disease	AD	99.98%	10 of 10
POR	Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis, Congenital Adrenal Hyperplasia Due To Cytochrome P450 Oxidoreductase Deficiency, Disordered Steroidogenesis Due To Cytochrome P450 Oxidoreductase	AD,AR	99.98%	67 of 68
PRKAR1A	Acrodysostosis, Acute Promyelocytic Leukemia, Carney Complex, Familial Atrial Myxoma, Pigmented Nodular Adrenocortical Disease, Primary Pigmented Nodular Adrenocortical Disease	AD	95.93%	165 of 171
STAR	Familial Glucocorticoid Deficiency, Lipoid Congenital Adrenal Hyperplasia	AR	100%	80 of 80

*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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