Panel para hiperplasia suprarrenal congénita

Este panel está diseñado para pacientes con sospecha de CAH. CAH comprende un grupo trastornos hereditarios caracterizados por el funcionamiento inadecuado de las glándulas suprarrenales, resultando en una producción anormal de hormonas esteroideas como el cortisol o la aldosterona. Nuestro panel incluye el análisis del gen CYP21A2, que codifica para la enzima 21-hidroxilasa. Más del 90% de los casos de CAH son causados por la deficiencia de esta enzima.

Nº de genes:	12
Entrega:	25 días
Cobertura:	≥99,5% ≥20x Cobertura media con profundidad ≥150 x
Detalles:	El análisis de CNV incluyó el análisis de CYPA21A2

Resumen de genes y enfermedades asociadas (OMIM) incluidos en este panel:

Genes	OMIM (gen)	Enfermedades asociadas (OMIM)	Herencia
ARMC5	615549	ACTH-independent macronodular adrenal hyperplasia 2	AD, SM
CYP11A1	118485	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	
CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency;Aldosteronism, glucocorticoid-remediable	AR, AD
CYP11B2	124080	Hypoaldosteronism, congenital, due to CMO II deficiency;Hypoaldosteronism, congenital, due to CMO I deficiency	AR
CYP17A1	609300	17,20-lyase deficiency, isolated;17-alpha-hydroxylase/17,20-lyase deficiency	AR

CYP21A2	613815	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency;Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency	AR
HSD3B2	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR
PDE11A	604961	Pigmented nodular adrenocortical disease, primary, 2	AD
PDE8B	603390	Striatal degeneration, AD;Pigmented nodular adrenocortical disease, primary, 3	AD
POR	124015	Disordered steroidogenesis due to cytochrome P450 oxidoreductase;Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	AR
PRKAR1A	188830	Myxoma, intracardiac;Pigmented nodular adrenocortical disease, primary, 1;Carney complex, type 1;Acrodysostosis 1, with or without hormone resistance	AD
STAR	600617	Lipoid adrenal hyperplasia	AR
CYP21A2	613815	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency;Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency	AR

CYP21A2	613815	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency;Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency	AR