ACTIA

MEDGENOME

Congenital Adrenal Hyperplasia (CAH)

aka 21-hydroxylase deficiency

Congenital Adrenal Hyperplasia

- An inherited disorder that affects the production of hormones from the adrenal glands and affects health, growth & development of the child.

- In this disorder adrenal glands sends too many hormones for body growth which leads to an early growth spurt, but the final adult height is usually shorter than others in their family.

Prevalence

Classic form :

- 1 in 15,000 newborns.

Non-classic form:

- 1 in 1,000 individuals. Responsible for about 95 percent of all cases of congenital adrenal hyperplasia.

Now your patients can

SAVE MORE!

21-OH deletion/duplication MLPA + Sanger sequencing at an attractive price*

*Contact MedGenome's representative to know more, and avail this offer.

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Congenital Adrenal Hyperplasia

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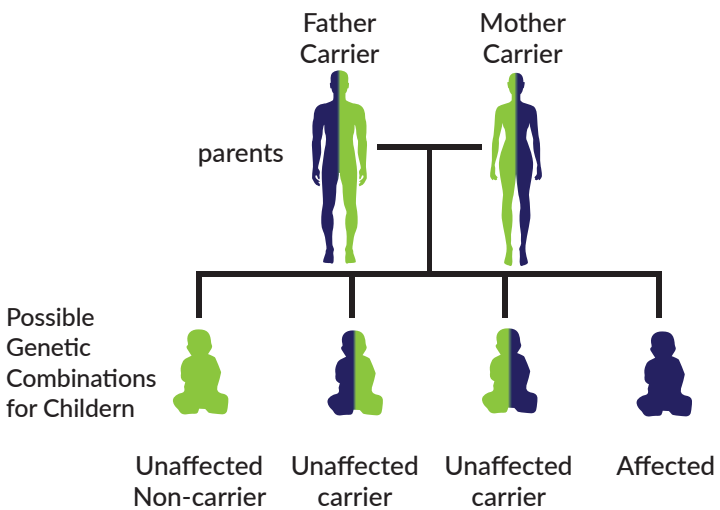
Inheritance of Congenital Adrenal Hyperplasia:



Pathophysiology:

CAH results when our body is unable to produce an essential enzyme in our adrenal glands, known as 21-hydroxylase, due to mutation or deletions in the CYP21A2 gene, which codes for this enzyme.

Inheritance Pattern:

This is an Autosomal recessive condition; meaning that two defective copies of the gene are required to cause the disorder

The symptoms are common and gender-specific.

Common: Poor feeding, weight loss, dehydration & vomiting.

Boys: Small testes.

Girls: Ambiguous genitalia, hirsutism, male pattern baldness, irregular menstruation and decreased fertility.

Diagnosis & Genetic Testing:

MedGenome offers	Test Sample requirements	Methodology	TAT
Congenital adrenal hyperplasia CYP21A2 (21-0H) deletion/duplication analysis	Peripheral blood OR Purified genomic DNA	MLPA	21 days
Congenital adrenal hyperplasia CYP21A2 (21-0H) gene analysis		Sanger	