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**2 + 0 CYP21A2 deletion carrier: A limitation of the genetic testing and counseling:
A case report**

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Abstract

Case Report

Methods

Functional Analysis of The M...



Congenital adrenal hyperplasia (CAH) is a group of disorders caused by inborn errors of metabolism of steroid hormones production. The most common form, 21-hydroxylase deficiency (21OH-def), is inherited as autosomal recessive disease. Heterozygote carriers are usually asymptomatic with normal basal hormonal blood level including 17-hydroxyprogesterone (17OH-P), the keystone for diagnosing the disease (1-3). The disease severity varies from asymptomatic or mild disease with some hyperandroge...

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Laboratories with multigene panels comprising very similar lists of genes may manage variants of uncertain significance differently, potentially causing a substantial clinical burden in interpretation (e.g., **testing** additional family members to clarify the result), and **genetic counseling** (e.g., clarifying the difference between a pathogenic ...

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