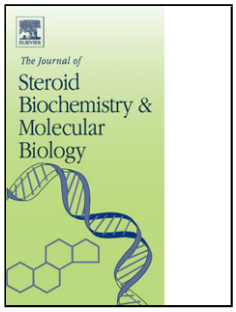

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A high rate of novel *CYP11B1* mutations in Saudi Arabia

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Highlights

- In Saudi Arabia, 11 β-hydroxylase deficiency (11-β OHD) has an unique genotype.
- Novel *CYP11B1* mutations are common in Saudi patients with 11-β OHD deficiency.
- The novel p.448R>P mutation is the most common mutation in this inbred population
- The genotype/phenotype correlation is modest

Abstract

Despite ethnic variation, 11 β-hydroxylase deficiency (11-β OHD) has generally been considered the second most common subtype of congenital adrenal hyperplasia (CAH). We report a high rate of novel mutations in this gene from Saudi Arabia. We studied 16 patients with 11-β OHD from 8 unrelated