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# A report of congenital adrenal hyperplasia due to 17α-hydroxylase deficiency in two 46,XX sisters

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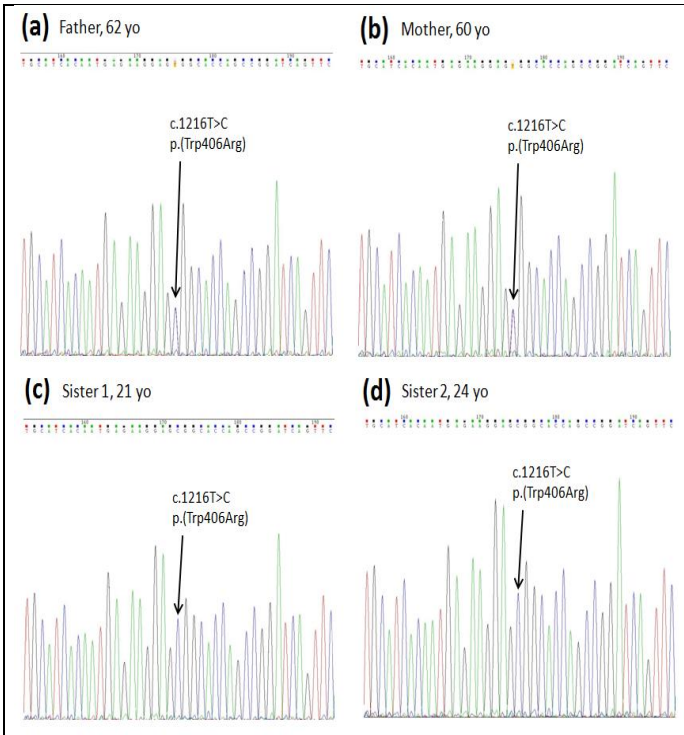


Figure 2. Parents exhibit the same mutation as asymptomatic heterozygotes panels (a) and (b), thereby confirming the homozygosity found in the sisters' panels (c) and (d).

## Abstract

**Context:** Congenital adrenal hyperplasia (CAH) is a group of rare orphan disorders, caused by mutations in seven different enzymes that impair cortisol biosynthesis. The 17α-hydroxylase deficiency (17OHD) is one of the less common forms of CAH, corresponding to approximately 1% of the cases, with an estimated annual incidence of 1 in 50 000 newborns. **Cases description:** Two phenotypically female Ecuadorian sisters, both with primary amenorrhea, absence of secondary sexual characteristics, and osteoporosis. High blood pressure was present in the older sister. Hypergonadotropic hypogonadism profile was observed: decreased cortisol and dehydroepiandrosterone sulphate (DHEAS), increased adrenocorticotropic hormone (ACTH) and normal levels of 17-hydroxyprogesterone, extremely high deoxycorticosterone (DOC) levels, and a tomography showed bilateral adrenal hyperplasia in both sisters. Consanguinity was evident in their ancestors. Furthermore, in the exon 7, the variant c.1216T>C, p.Trp406Arg was detected in homozygosis in the *CYP17A1* gene of both sisters.

**Conclusion:** We report a homozygous missense mutation in the *CYP17A1* gene causing 17OHD in two sisters from Loja, Ecuador. According to the authors, this is the first time such deficiency and mutation are described in two members of the same family in Ecuador.

**Keywords:** Congenital adrenal hyperplasia, 17 alpha hydroxylase deficiency, sisters, consanguineous family.

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