# Complete 17α-Hydroxylase Deficiency: Clinical Characteristics and Molecular Etiology in a 46, XX Algerian Patient

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**Abstract—** 17α-hydroxylase/17,20–lyase deficiency (17OHD) is a rare form of congenital adrenal hyperplasia (CAH) caused by mutations in the *CYP17A1 gene*. This gene encodes the P450c17 enzyme, which is essential for the production of cortisol and sex steroids in the adrenal cortex, ovaries, and testes.

The aim of this study is to present the clinical and genetic characteristics of an Algerian woman with a classic form of 17OHD.

We report a 28–year-old woman from a first-degree consanguineous marriage, with hypoplasia of the external genitalia, undifferentiated uterine structure and prepubertal ovaries. The hormonal profile reveals high levels of FSH and LH, normal testosterone, low cortisol, and elevated ACTH. Clinical features include iron-deficiency anemia, hypertension, and peripheral adrenal insufficiency, likely related to a 17OHD enzyme block. Ovarian insufficiency is manifested by primary amenorrhea, delayed puberty and low estradiol levels.

The analysis of whole exome sequencing (WES) identified a rare homozygous missense variant, p.Met1Arg, in the CYP17A1 gene. This variant, with a minor allelic frequency of 8.517e-7 in European (non-Finnish) populations, had not been previously reported. The potential pathogenicity of this variant was assessed *in silic*o using bioinformatics tools. Clinically, 17OHD was suspected in the proband, and this genetic finding has confirmed the diagnosis.