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Meryam Alahyane, Nawal El Ansari

Corresponding author: Meryam Alahyane, Department of Endocrinology, Diabetology, Metabolic Diseases and Nutrition, Mohammed VI University Hospital, Marrakesh, Morocco. meryam.alahyane1@gmail.com

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Clinical characteristics of 46,XX male with congenital adrenal hyperplasia in Morocco

Meryam Alahyane[1,&], Nawal El Ansari[1]

[1]Department of Endocrinology, Diabetology, Metabolic Diseases and Nutrition, Mohammed VI University Hospital, Marrakesh, Morocco

[&]Corresponding author

Meryam Alahyane, Department of Endocrinology, Diabetology, Metabolic Diseases and Nutrition, Mohammed VI University Hospital, Marrakesh, Morocco

Image in medicine

We report the case of a Moroccan patient with severe virilization and a delayed diagnosis of congenital adrenal hyperplasia due to 21-hydroxylase deficiency, diagnosed at the age of 9. The diagnosis was clinically suspected following an adrenal crisis, precocious puberty, and disorders of sex differentiation, presenting as Prader Stage 5. It was confirmed by a 46,XX karyotype (A) and elevated levels of 17-hydroxyprogesterone (1500 nmol/L), testosterone (5.27 ng/mL), and progesterone (287 ng/mL). Born via home birth without prior medical assessment, the patient was raised as male. Hydrocortisone (10 mg/day) was initiated for glucocorticoid replacement. The pediatric team respected the

male gender assigned at birth and opted for hysterectomy and bilateral salpingo-oophorectomy, without testicular prosthesis placement. The patient was lost to follow-up for 7 years but maintained hydrocortisone therapy. At 21 years of age, he presented to the Adult Endocrine Department. Clinical examination revealed a masculine phenotype (B), including a penis length of 8.5 cm, empty scrotum, Stage 5 pubic (C) and axillary hair (D), height of 1.38 m (E), bilateral adipomastia, moderate obesity and normal blood pressure. Testosterone levels were measured at 4.31 ng/mL. The current management plan focuses on optimizing hydrocortisone supplementation. However, it raises significant ethical and religious dilemmas, particularly regarding testosterone supplementation to improve the patient's sexual life as a phenotypic male despite a genetically female identity.

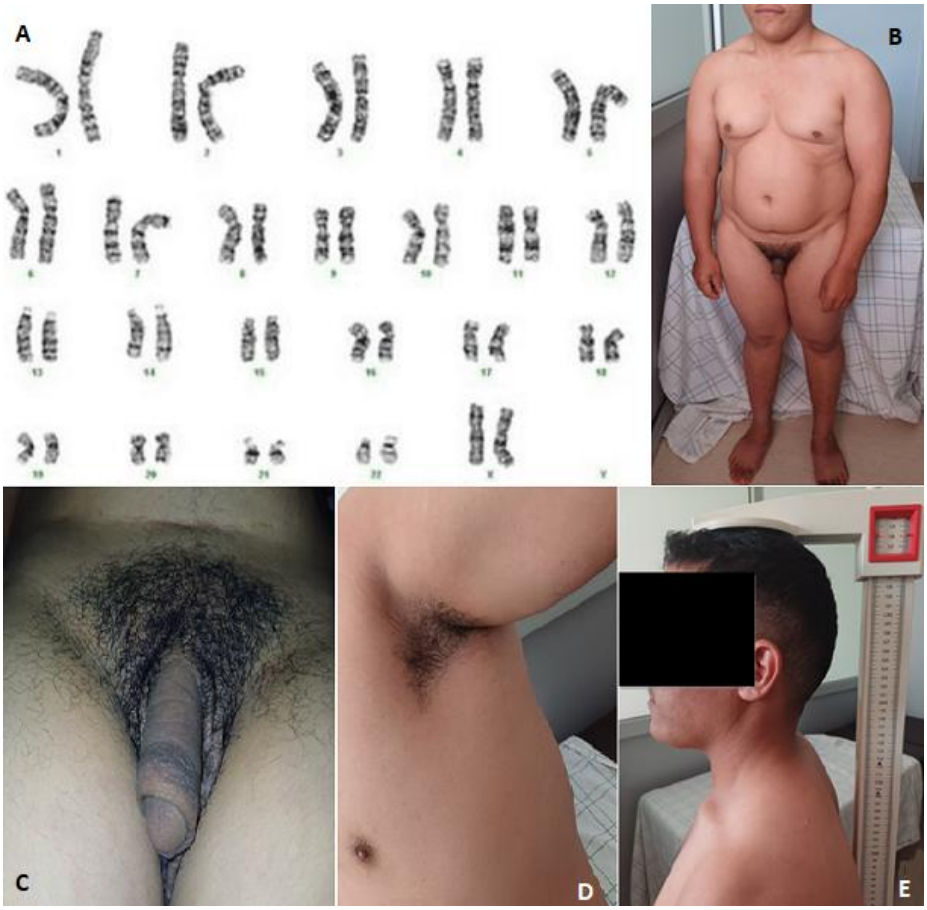

Figure 1: A) 46,XX karyotype confirmation; B) masculine phenotype; C) penis length of 8.5 cm, empty scrotum, pubic hair; D) axillary hair; E) height of 1.38 m