

46, XX disorder of sex development due to simple virilizing congenital adrenal hyperplasia: A case report

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Abstract

Background: Congenital adrenal hyperplasia is an important cause of 46,XX disorder of sex development and may present with virilized external genitalia at birth. Early recognition is essential to guide sex assignment, endocrine evaluation, counselling, and timely surgical planning.

Case summary: A 4-year-old child presented with ambiguous genitalia noted since birth. The child was born at term by caesarean section to non-consanguineous parents, with no neonatal illness or history suggestive of a salt-wasting crisis. At birth, the parents observed an enlarged phallus, a single perineal opening, and no visible testes. The child was initially assigned male and reared as a boy until 8 months, then reared as a girl after further consultations. Examination showed clitoromegaly resembling a phallus measuring 2.5 × 1.5 cm, partial labial fusion, a single urogenital opening, and no palpable gonads, consistent with Prader stage III. Height was between the 3rd and 10th percentiles and weight was below the 3rd percentile. Blood pressure was normal. Karyotype was 46,XX. Serum electrolytes, abdominal ultrasonography, hormone profile, adrenocorticotrophic hormone, and cortisol testing were normal, with no evidence of salt wasting or cortisol deficiency. Based on virilized 46,XX genitalia, absence of hypertension, negative antenatal androgen exposure, and exclusion of 46,XY under-virilization disorders, the final diagnosis was 46,XX disorder of sex development due to simple virilizing congenital adrenal hyperplasia, most consistent with partial 21 β -hydroxylase deficiency.

Conclusion: Ambiguous genitalia in a newborn requires prompt multidisciplinary evaluation before irreversible sex-assignment decisions. This case highlights that simple virilizing 21 β -hydroxylase deficiency may present without salt loss or cortisol deficiency, and that 46,XX karyotyping with careful clinical assessment is pivotal for diagnosis and counselling. [*J Assoc Clin Endocrinol Diabetol Bangladesh*, 2026;5(Suppl 1): S64]

Keywords: Congenital adrenal hyperplasia, 46,XX disorder of sex development, Ambiguous genitalia, 21 β -hydroxylase deficiency, Prader stage III, No palpable gonads

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