

Distinct etiologies of 46,XY differences of sex development within a single family: A case series of 17 β -HSD3 deficiency and complete androgen insensitivity syndrome

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Abstract

Background: 17 β -hydroxysteroid dehydrogenase type 3 (17 β -HSD3) deficiency and complete androgen insensitivity syndrome (CAIS) are rare causes of 46,XY differences of sex development (DSD) that may present with similar undervirilized phenotypes despite distinct underlying mechanisms. We report two related individuals from the same maternal lineage, both raised as females, who were found to have different forms of 46,XY DSD.

Case summary: A 6-year-3-month-old child presented with ambiguous genitalia, bilateral palpable inguino-labioscrotal gonads, and a 46,XY karyotype. Pelvic ultrasonography demonstrated absent Müllerian structures. An hCG stimulation test showed a low testosterone-to-androstenedione ratio, consistent with 17 β -HSD3 deficiency. The second case was the proband's 17-year-old maternal cousin, also with a 46,XY karyotype, who presented with bilateral inguinal swellings, female-appearing external genitalia, and absent Müllerian structures on imaging, a phenotype consistent with CAIS.

Conclusion: These cases highlight that distinct etiologies of 46,XY DSD can occur within the same family and emphasize the importance of individualized biochemical and genetic evaluation of affected relatives rather than assuming a shared diagnosis. A multidisciplinary approach is essential for optimal long-term management and counselling. [*J Assoc Clin Endocrinol Diabetol Bangladesh*, 2026;5(Suppl 1): S79]

Keywords: 46,XY Differences of Sex Development, 17 β -hydroxysteroid dehydrogenase type 3 deficiency, HSD17B3, Complete androgen insensitivity syndrome, Familial DSD, Ambiguous genitalia; hCG stimulation test

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