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# Ambiguous Genitalia in a 3-Year-Old: A Case Highlighting 5α-reductase deficiency

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**Abstract**

5α-reductase deficiency is a rare autosomal recessive disorder of sex development resulting from mutations in the SRD5A2 gene, which impairs the conversion of testosterone to dihydrotestosterone (DHT). We report a case of a 3-year-old child, first born of consanguineous parents, who presented with severe pallor, growth failure, and ambiguous genitalia since birth. He was born preterm with low birth weight and required NICU admission for delayed crying. Initially reared as female due to apparently female genitalia, parental concern arose at 6 months when progressive phallic enlargement and bilateral inguinal swellings were noted. Cytogenetic analysis confirmed a normal 46,XY male karyotype. Bilateral orchidopexy was performed at 1.5 years. Despite three intramuscular testosterone injections microphallus persisted. Examination revealed moderate anemia, growth retardation, bifid scrotum with pea-sized testes, phallus length 2.5 cm, and penoscrotal hypospadias (EMS 3). Hematological evaluation suggested iron deficiency anemia, necessitating four blood transfusions. β-hCG stimulation test showed elevated testosterone (250.34 ng/dl) with disproportionately low dihydrotestosterone (2.88 ng/dl) and raised T/DHT ratio(86.92:1), indicating 5α-reductase deficiency as the likely etiology of 46,XY DSD. Molecular genetic testing has been advised. This case highlights the complexity of managing 46,XY DSD with suspected 5α-reductase deficiency, particularly when compounded by systemic comorbidities such as chronic anemia and growth failure. Early diagnosis is crucial for gender assignment decisions, counseling of parents, and planning future surgical and hormonal interventions. *[J Assoc Clin Endocrinol Diabetol Bangladesh, 2025;4(Suppl 1): S63]*

**Keywords:** 46,XY DSD, Ambiguous genitalia, 5α-reductase deficiency, SRD5A3 gene, Testosterone/DHT ratio

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