Androgen Insensitivity Syndrome: A Rare Case Report

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Case Complete androgen insensitivity syndrome (AIS) or testicular feminization syndrome is defined as female phenotype in a 46 XY male with normal testes and normal testosterone production and metabolism. AIS is an X-linked recessive disorder with an incidence of 1:20,000-64,000 male births.¹ Point mutations in the androgen receptor gene,
which results in a defective receptor protein which is unable to bind hormone or bind to DNA are the most common cause of AIS.

A 26-year-old unmarried female presented with primary amenorrhea. Examination revealed secondary sexual characteristics with breast development Tanner Stage IV and normal external genitalia with sparse axillary and pubic hair along with a blind-ending vaginal pouch, on per rectal examination uterus could not be palpated. Investigations revealed serum testosterone: 152.21 ng/ml, follicle-stimulating hormone: 20.29 mIU/l, luteinizing hormone: 31.05 mIU/l. Thyroid function tests were normal, ultrasound examination performed and showed absent uterus and ovaries, bilateral hypoechoic oval structures in both inguinal canals suggestive of testes (right measuring - 3.7 cm × 1.4 cm, left measuring - 3.6 cm × 1.5 cm), buccal smear: Negative for barr body, chromosomal analysis: Karyotype 46 XY. Patient was counseled regarding need for gonadectomy, and the option of vaginoplasty, bilateral high inguinal orchidectomy was performed and testes sent for histopathological examination. Post-operative period is uneventful.

**POINTS TO PONDER**

1. The diagnosis of AIS is based on clinical examination with lab investigations, confirmed by histopathology report showing the presence of testicular tissue
2. Gonadectomy is advised to prevent the risk of gonadal tumor, patient can also opt for vaginoplasty if sexual function is desired.

**REFERENCES**


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