

Caught Inbetween

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Introduction

Partial androgen insensitivity syndrome is a rare syndrome that presents with a spectrum of defects in virilization and vary from women with mild degrees of virilization to sometimes fertile but undervirilized men.

Case report

A case of a 20-year-old male who presented with gynecomastia. He noted beginning enlargement of the breasts, no discharges but with tenderness, starting around 11 years old. During puberty, he had minimal hair growth on face, axilla, pubic area and legs, minimal deepening of voice, minimal scrotal enlargement. He identifies himself with the male gender and plays basketball and street games. He also had numerous opposite sex relationship in high school but denies sexual contact. He claims he masturbates since 13 years old. On physical examination, he had minimal facial, axillary and leg hair. Had tanner stage 5 mature developed breasts, pubic hair distribution of a female, covering smaller area than adult. Penis was 2cm in length with presence of prepuce and glans with hypospadias. Scrotum examination revealed less than 2 cm nontender symmetrical testicles. Laboratory workup revealed Karyotype 46 XY male. FSH (2.938 IU/L), LH (5.455 IU/L), DHEAS (3.12 umol/L), B-HCG (<0.1 mIU/mL), Prolactin (7.834 ng/mL), TSH (2.042 uIU/mL), FT4 (16.58 pmol/L), and cortisol (18.7 ug/dL) were all normal. While 17-hydroxprogesterone (3.007 ng/mL), Estradiol (88.490pmol/L), Testosterone (51.958 nmol/L) were elevated.

Conclusion:

The patient underwent intensive psychologic support. Eventually he underwent corrective surgery with mastectomy and hypospadias repair. He was also advised to have high dose androgen therapy.