Complete Androgen Insensitivity Syndrome: Diagnosis and Psychological Impact in Adolescence, A Case Report

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Abstract

This case report illustrates the case of complete androgen insensitivity syndrome (CAIS) which is a rare form of sexual development disorders (DSD). Complex critical thinking is needed for pathophysiology of primary amenorrhea causes and sex chromosomes differences of sexual development, such as primary ovarian failure, Mullerian Agenesis or disorders with abnormal androgen synthesis or response. This is a phenotypically female who presented with primary amenorrhea at the age of 19 years old. Normal levels of thyroid function test, serum prolactin and follicle stimulating hormone ruled out hypothyroidism, hyperprolactinemia, and primary ovarian failure. Magnetic resonance imaging showed absence of uterus, fallopian tubes, ovaries, but presence of proximal 1/3rd of the vagina. There is a single testis in the left inguinal region with unknown status of spermatogenesis. The chromosomal analysis revealed 46, XY karyotype conveying the patient is genotypically male. The testis-determining factor (TDF) test or sex-determining region Y (SRY) protein for male sex determination was not done. Similar presentation of primary amenorrhea diagnosis of CAIS was made to her 18-year-old sister. Women with CAIS are vulnerable to various psychological conditions caused by the appalling fact of being genotypically male when they have been raised female all their life. The gender confusion, reproductive issues and how others perceive them in the outside world require sensitive support. Hence, accentuate the need to address the emotional, psychological, and psychiatric vulnerabilities in issues pertaining to relationships, infertility and conception.

Keywords: complete androgen insensitivity syndrome, psychological impact, adolescence

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