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Case Report

Complete Androgen Insensitivity Syndrome Presenting as Primary Amenorrhea in an Adolescent Female: A Case Report

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ABSTRACT

Introduction: Complete Androgen Insensitivity Syndrome (CAIS) is a rare X-linked recessive disorder caused by mutations in the androgen receptor gene, resulting in androgen resistance in individuals with a 46,XY karyotype. It commonly presents with primary amenorrhea, female phenotype, absent uterus, and undescended testes.

Case Presentation: An 18-year-old phenotypic female presented with primary amenorrhea. She exhibited normal breast development with sparse pubic and axillary hair, normal external genitalia, and a short blind-ending vaginal pouch. Hormonal evaluation revealed elevated serum testosterone and luteinizing hormone levels, with normal estradiol. Imaging studies, including pelvic ultrasonography and MRI, demonstrated absent Müllerian structures and bilateral intra-abdominal testes. Karyotyping confirmed a 46,XY chromosomal pattern. After diagnosis, the patient and her family received counseling regarding infertility, risk of gonadal malignancy, and options for elective gonadectomy. A multidisciplinary plan was formulated, including gonadectomy after spontaneous pubertal development, estrogen replacement therapy, and psychological support for long-term follow-up. **Result**: The patient had normal breast development with sparse pubic and axillary hair and a blind-ending vagina. Hormonal evaluation revealed elevated serum testosterone and LH levels with normal estradiol. Imaging demonstrated absent uterus and bilateral intra-abdominal testes. Karyotyping confirmed a 46,XY chromosomal pattern, establishing the diagnosis of CAIS. The patient underwent multidisciplinary counseling regarding infertility, gonadectomy, and hormonal management.

Conclusion: CAIS should be suspected in phenotypic females presenting with primary amenorrhea and absent uterus. Early diagnosis and multidisciplinary management are essential for appropriate hormonal, surgical, and psychological care.

Keywords: *Complete Androgen Insensitivity Syndrome; CAIS; Primary Amenorrhea; 46,XY Disorder of Sex Development; Androgen Receptor Gene; Phenotypic Female; Gonadal Dysgenesis; Undescended Testes; Disorders of Sex Development (DSD).*

INTRODUCTION

Androgen Insensitivity Syndrome (AIS) is a rare X-linked recessive disorder of sex development caused by mutations in the androgen receptor (AR) gene, leading to resistance to androgen action in individuals with a 46,XY karyotype [1]. Despite normal production of testosterone and dihydrotestosterone, defective androgen receptor function results in impaired masculinization and varying phenotypic presentations ranging from complete female phenotype to mild male

infertility [2,3]. AIS is classified into Complete AIS (CAIS), Partial AIS (PAIS), and Mild AIS (MAIS) depending on the degree of androgen resistance [4].Complete Androgen Insensitivity Syndrome is the most severe form and is characterized by phenotypic females with normal breast development, sparse pubic and axillary hair, absent Müllerian structures, and undescended testes [1,5]. The condition is commonly diagnosed during adolescence while evaluating primary amenorrhea [6]. The androgen receptor gene is located on chromosome Xq11–12 and plays a vital role in male sexual differentiation [5]. Failure of androgen action leads to feminization, whereas anti-Müllerian hormone secretion by Sertoli cells causes regression of the uterus and fallopian tubes [7].Patients with CAIS typically present with primary amenorrhea despite normal secondary sexual characteristics [8]. Hormonal evaluation usually reveals elevated testosterone levels within the normal male range and elevated luteinizing hormone levels [3]. Imaging studies demonstrate absent uterus and intra-abdominal testes, while karyotyping confirms a 46,XY chromosomal pattern [4,8]. One of the major concerns in AIS is the risk of gonadal malignancy associated with undescended testes, particularly after puberty [10]. Therefore, gonadectomy is generally recommended after spontaneous pubertal development, followed by hormone replacement therapy and psychological counseling as part of multidisciplinary management [11].

In this report, we present a classic case of Complete Androgen Insensitivity Syndrome in an adolescent phenotypic female presenting with primary amenorrhea and diagnosed through hormonal, radiological, and cytogenetic evaluation.

CASE PRESENTATION

An 18-year-old phenotypic female presented to the Gynaecology Outpatient Department with the chief complaint of primary amenorrhea. She had attained normal pubertal breast development at approximately 13 years of age. There was no history of cyclical abdominal pain, virilization, genital ambiguity, or chronic medical illness. Her developmental milestones were normal, and there was no significant family history of disorders of sex development or infertility.

On general physical examination, the patient was moderately built and well nourished with a height of 160 cm and weight of 52 kg. Secondary sexual characteristics revealed Tanner stage V breast development with sparse pubic and axillary hair corresponding to Tanner stage I–II. External genital examination showed normal female genitalia with normal labia and clitoris. Vaginal examination revealed a short blind-ending vaginal pouch. No inguinal swelling or palpable gonadal mass was detected.

Routine hematological and biochemical investigations were within normal limits. Hormonal evaluation revealed mildly elevated follicle-stimulating hormone (FSH) and elevated luteinizing hormone (LH) levels. Serum estradiol was within the normal female pubertal range, whereas serum testosterone levels were elevated within the normal adult male range. Thyroid profile and serum prolactin levels were normal.

Pelvic ultrasonography demonstrated absent uterus and ovaries with bilateral intra-abdominal gonadal structures suggestive of testes. Magnetic resonance imaging (MRI) of the pelvis confirmed absence of Müllerian structures and revealed bilateral intra-abdominal testes along with a short blind-ending vagina.

Chromosomal analysis showed a 46,XY karyotype. Based on clinical findings, hormonal profile, radiological evaluation, and cytogenetic analysis, a diagnosis of Complete Androgen Insensitivity Syndrome (CAIS) was established.

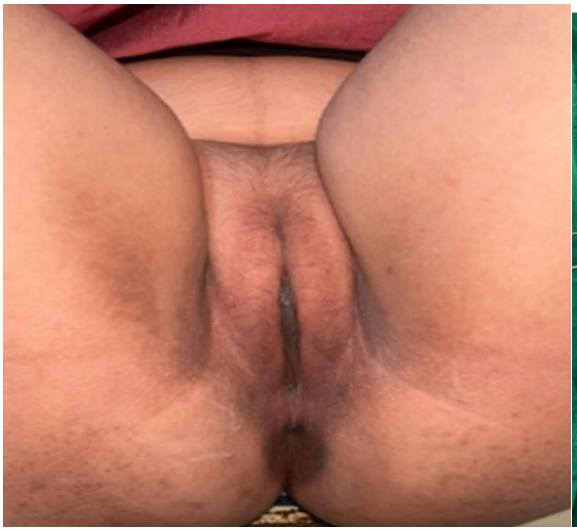

Image 1: Breast development Tanner stage -5 Image 2: Intra op – Gonadectomy gonads)

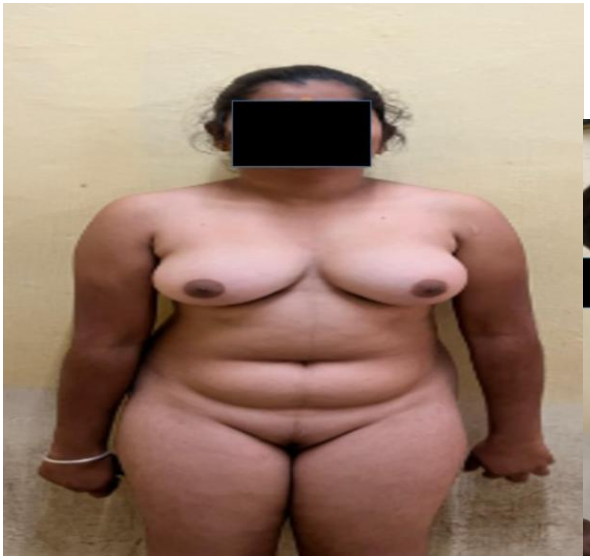

Image 3: Pubic hair tanner stage 2

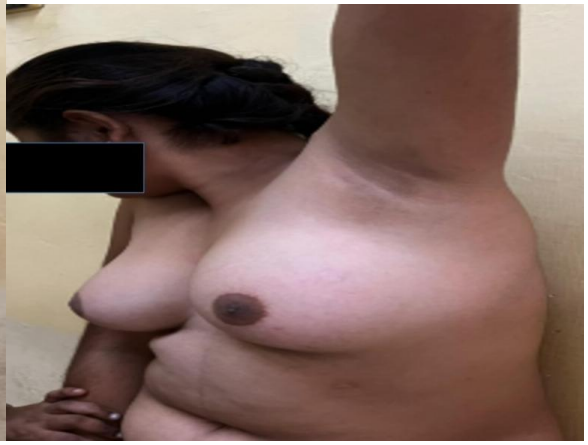

Image 4: Axillary hair tanner stage 2

The patient and her family were counseled regarding the nature of the disease, female gender identity, infertility, risk of gonadal malignancy, and the need for long-term follow-up. After multidisciplinary consultation, the patient was referred for elective gonadectomy following completion of spontaneous pubertal development. Psychological counseling was initiated, and estrogen replacement therapy was planned postoperatively. The patient was advised regular follow-up for endocrine, surgical, and psychological assessment.

DISCUSSION

In the present case, an 18-year-old phenotypic female presented with primary amenorrhea, Tanner stage V breast development, sparse pubic and axillary hair, normal female external genitalia, and blind-ending vagina, which are classical features of CAIS. Chen et al. (2023) reported that CAIS patients commonly present with primary amenorrhea, female phenotype, sparse pubic hair, absent uterus, and undescended testes [12]. Liu et al. (2020) observed that 27 out of 39 AIS patients (69.2%) had CAIS, with sparse or absent pubic hair present in 84.6% cases [13]. He et al. (2017) reported primary amenorrhea, female phenotype, and absent uterus in all 6 CAIS patients studied [14]. Fulare et al. (2020) similarly described a 17-year-old phenotypic female presenting with primary amenorrhea and normal secondary sexual characteristics [15].Our patient demonstrated elevated serum testosterone within the adult male range, elevated LH, mildly raised FSH, and normal estradiol levels. Liu et al. reported mean testosterone levels of 18.3 ± 6.7 nmol/L with elevated LH levels in 79.5% of CAIS patients [13]. He et al. reported testosterone levels ranging from 16.2–27.8 nmol/L in all six CAIS patients [14]. Pelvic ultrasonography and MRI pelvis in our patient revealed absent uterus and bilateral intra-abdominal testes, while karyotyping showed a 46,XY chromosomal pattern. Liu et al. reported absent uterus in 100% of CAIS patients with undescended testes in most cases [13]. He et al. also observed absent Müllerian structures and bilateral intra-abdominal testes in all six patients [14]. Fulare et al. similarly reported absent uterus with bilateral gonads in a phenotypic female having 46,XY karyotype [15]. Jääskeläinen reported that AIS results from mutations of the androgen receptor gene located on chromosome Xq11–12, causing impaired androgen signaling [16]. Chen et al. explained that preserved anti-Müllerian hormone secretion leads to regression of Müllerian structures despite female phenotype [12]. In our case, the patient underwent counseling regarding infertility, gonadal malignancy risk, and gonadectomy after puberty, followed by planned estrogen replacement therapy and psychological counseling. Chen et al. recommended delayed gonadectomy after spontaneous puberty to permit natural feminization [12]. Jääskeläinen emphasized multidisciplinary management including hormonal therapy and psychological support [16].

CONCLUSION

Complete Androgen Insensitivity Syndrome should be suspected in phenotypic females presenting with primary amenorrhea, normal breast development, and absent uterus. The present case demonstrated classical clinical, hormonal, radiological, and cytogenetic features of CAIS. Early diagnosis and multidisciplinary management are essential for appropriate counselling, timely gonadectomy, and hormonal therapy. Long-term psychological and endocrine follow-up is important for optimal patient outcomes.

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