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CASE REPORT

From Diagnosis to Gender Identity Resolution in Androgen Insensitivity Syndrome: Clinical and Psychological Insights from a Case Study

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Abstract

Androgen insensitivity syndrome (AIS), formerly known as testicular feminizing syndrome, is a rare genetic condition where individuals with a male XY karyotype are resistant to male hormones (androgens), leading to the development of female physical characteristics despite having a male genetic makeup. The condition results from mutations in the AR gene. Defective receptor assembly leads to varying degrees of androgen insensitivity, ranging from complete AIS (CAIS) to partial (PAIS) or mild forms (MAIS).

We present a case report of patients in their 20s raised as girls, who presented with primary amenorrhea and ambiguous genitalia, accompanied by signs of masculinization. MRI findings revealed absent reproductive structures, such as the uterus and ovaries, but showed the presence of bilateral testicles. After careful discussions among medical professionals and the patient's family, a decision was made to assign the male gender. The patient was subsequently scheduled for surgical intervention, including the first stage of chordee repair and right orchidopexy.

AIS, an X-linked disorder, results from mutations in the AR gene, leading to a spectrum of androgen insensitivity. Managing AIS requires a multidisciplinary approach due to its complexity. In this case, surgical interventions such as chordee repair and genitoplasty successfully addressed medical and cosmetic concerns. Studies on cosmetic outcomes post-genioplasty show significant improvement and satisfaction among patients, emphasizing the importance of early intervention and long-term follow-up. Following multidisciplinary evaluation and surgical intervention (chordee repair and orchidopexy), the patient successfully transitioned to male gender assignment with satisfactory functional and cosmetic results. The case highlights the critical need for tailored care in managing AIS.

Introduction

Androgen insensitivity syndrome (AIS), formerly known as testicular feminizing syndrome, is a rare genetic disorder in which individuals with a 46, XY karyotype cannot respond properly to androgens. Consequently, individuals who are afflicted develop female external characteristics despite possessing male

chromosomes.[1] AIS is an XY disorder of sex development (DSD) that is frequently diagnosed, with an incidence of 1:40,800 to 1:99,000.[2] AIS is caused by a gene mutation that affects the androgen receptor (AR), also known as NR3C4. This receptor allows cells in many tissues to respond to androgens by binding to testosterone and dihydrotestosterone (DHT). When the receptor is assembled incorrectly, it can result in varying degrees of insensitivity of the cells to androgens, ranging from mild to severe.[3] Mutations of variable severity in the AR gene are associated with a wide phenotypic spectrum. This ranges from CAIS to a partial form, partial androgen insensitivity syndrome (PAIS), or a mild form, mild androgen insensitivity syndrome (MAIS). Patients with CAIS typically have external female genitalia, undescended testes, and fully developed breasts with little to no armpit and pubic hair. Patients with PAIS may present with predominantly male or female phenotypes, including hypospadias, clitoromegaly, ambiguous genitalia, and variable degrees of gynecomastia at puberty. Patients with MAIS present with normal external male genitalia and may experience infertility.[4] CAIS is a genetic condition caused by mutations in the androgen receptor (AR) gene on the X chromosome. This mutation prevents the body from responding to androgens, including testosterone, which are crucial for male sexual differentiation. In individuals with CAIS, the body's cells are unable to respond to these hormones. As a result, female external characteristics develop despite having a male genetic makeup and internal testes. Individuals with CAIS are typically raised as females because they have external female genitalia, including a typical-looking vulva and vagina, though often the vagina may be shorter than average and end in a blind pouch.[2] The internal reproductive organs typically include undescended testes, no uterus or fallopian tubes, and no menstruation. Breast development usually occurs at puberty due to the body's conversion of testosterone to estrogen, but they do not have a menstrual cycle or fertility due to the absence of a functional uterus and ovaries. Diagnosis of CAIS often occurs during adolescence when expected puberty-related changes do not happen, such as the onset of menstruation, or in early childhood if hernias or undescended testes are present. It can also be discovered during genetic testing or prenatal testing for other reasons. Learning about CAIS and one's genetic makeup can lead to significant emotional and psychological impacts, particularly because it involves understanding a divergence between chromosomal sex and gender identity.

This case offers valuable insights into the importance of personalized surgical management in CAIS, especially in adult patients, a population less commonly discussed in scientific literature. The patient's presentation with ambiguous genitalia, primary amenorrhea, and psychosocial concerns illustrates the complex and multifaceted nature of managing gender identity in CAIS. The case highlights not only the surgical complexities involved in procedures such as genitoplasty and orchidopexy but also emphasizes the critical role of psychological support and individualized, patient-centered counseling throughout the treatment process.

Additionally, this case contributes to the growing understanding of the broader implications of gender identity and social adaptation in patients with CAIS. It reinforces the evolving perspective on gender and sex development, drawing attention to the necessity of a multidisciplinary approach to ensure both physical and psychological well-being. This sets an important precedent for the future management of adult CAIS cases.

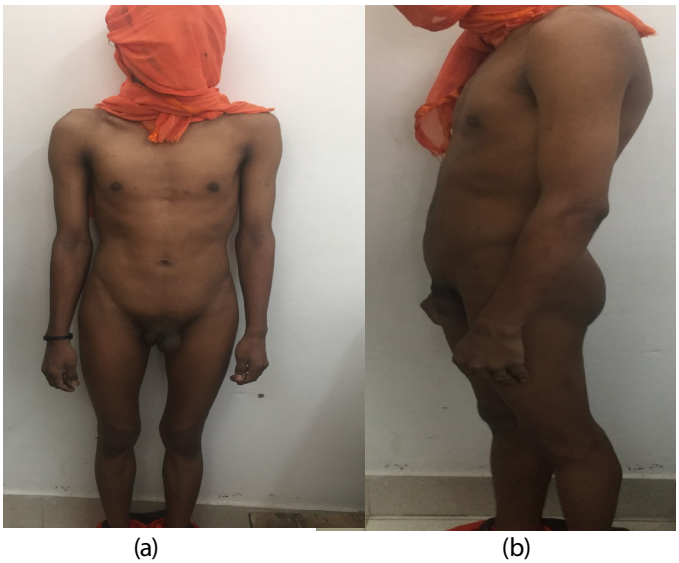


Figure 1: Male-typical body features with underdeveloped breast tissue- (a) Front view; (b) Lateral view (preoperative)

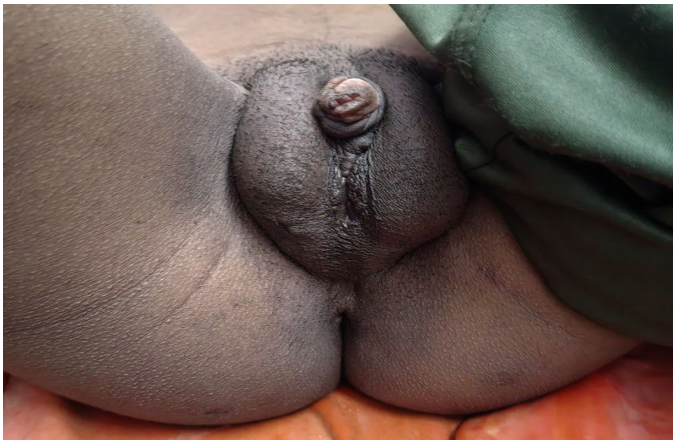


Figure 2: Patient's genital stage 1 repair of chordee correction and right orchidopexy (post-operative)

Case Report

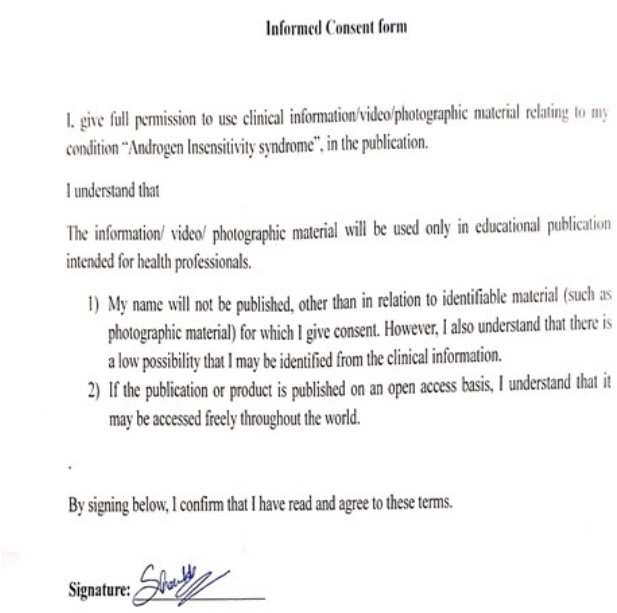
A 24-year-old individual assigned female at birth visited the outpatient department of Niswan wa Qabalat at Ajmal Khan Tibbiya College and Hospital in 2024. She was experiencing primary amenorrhea and lacked secondary sexual characteristics. Upon examination, she measured 161 cm in height, weighed 65 kg, and had a body mass index (BMI) of 25.07 kg/m^2. The preoperative clinical photographs (front and lateral views) demonstrated a male-typical appearance with minimal axillary hair and underdeveloped breast tissue (Figures 1a and b). Breast development was determined to be B1

Table 1: Laboratory test results

Laboratory test	*Patients Result*	*Reference range*
Serum FSH	2.4 IU/L	1.5–12.4 IU/L
Serum LH	9.5 IU/L	1.7–8.6 IU/L
Serum testosterone	320 ng/dl	300–1000 ng/dl
Karyotype	46,XY	46,XX (female)/ 46,XY (male)

Table 2: Imaging findings (MRI)

Imaging modality	*Findings*
MRI	Absence of uterus and ovaries
MRI	Hypoplastic Vagina
MRI	Intra abdominal gonads (likely testes)



Informed Consent form

I, give full permission to use clinical information/video/photographic material relating to my condition "Androgen Insensitivity syndrome", in the publication.

I understand that

The information/ video/ photographic material will be used only in educational publication intended for health professionals.

1) My name will not be published, other than in relation to identifiable material (such as photographic material) for which I give consent. However, I also understand that there is a low possibility that I may be identified from the clinical information.
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By signing below, I confirm that I have read and agree to these terms.

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Figure 3:

(Tanner stage 1/underdeveloped) on the sexual maturity rating (SMR/Tanner staging), with axillary hair being A1 (sparse) and pubic hair being P1–P2 (scanty/poor development). The patient was later referred to a higher center for further management due to ambiguous genitalia. In this case, a notable factor is the family history of androgen insensitivity syndrome (AIS) in a younger sibling, highlighting the genetic inheritance pattern of this condition. It was her first visit regarding her symptoms.

She was consulted in plastic surgery for gender assignment purposes. Upon examination, it was found that she had ambiguous genitalia, had fused labia majora, a significantly enlarged clitoris (clitoromegaly), and no apparent vaginal opening. She also had a well-formed glans with hooding of the prepuce on the dorsal side of the penis, while the ventral prepuce was absent. The chordee urethral opening was present at the penoscrotal junction, approximately 2 mm in size and oval in shape. Both testes were descended, but the right testis was retractile and located in the scrotum. Additionally, there is penoscrotal hypospadias.

At birth, the patient was assigned female based on ambiguous external genitalia, though no further investigation was pursued. During infancy and childhood, no significant developmental concerns were raised, and the ambiguous genitalia remained

unaddressed. At the onset of puberty, around 12 to 14 years of age, the patient did not experience menstruation, indicating primary amenorrhea. Additionally, no secondary sexual characteristics, such as breast development or body hair growth, were observed. By the age of 24, the patient presented with concerns about primary amenorrhea and the absence of breast development, and was noted to have ambiguous genitalia upon physical examination. These findings prompted further evaluation, leading to a more thorough investigation into the underlying cause of the patient's condition.

The hormonal profile presented in Table 1 indicated that the patient had normal serum follicle-stimulating hormone (FSH) levels at 2.4 IU/L, along with slightly elevated luteinizing hormone (LH) levels of 9.5 IU/L, and a testosterone level of 320 ng/mL. Further evaluation with magnetic resonance imaging (MRI), as shown in Table 2, revealed the absence of the uterus and ovaries, a hypoplastic vagina, and intra-abdominal gonads, likely testes, suggesting androgen insensitivity syndrome (AIS). Karyotyping confirmed a 46, XY pattern. After considering the patient's personal choice and discussions between the family and medical professionals, the patient was assigned a male gender, rather than the gender assigned at birth. The patient was scheduled for a procedure involving stage 1 chordee repair correction and right orchidopexy in April 2024. Figure 2 shows the patient's genital following surgery.

Follow-up and Outcome

The patient expressed high satisfaction with the cosmetic results postoperatively, and no immediate post-operative complications were noted. No hormonal therapy was given. The patient consistently followed post-operative care protocols, with regular check-ups and full compliance observed during recovery.

Discussion

AIS is a rare X-linked recessive disorder that results in individuals with a male genotype (46, XY) being resistant to androgens, the male sex hormones. Consequently, despite their genetic makeup, these individuals develop female or ambiguous physical characteristics. The clinical presentation of AIS is influenced by the extent of androgen receptor functionality. In CAIS, androgen receptors are entirely non-functional, resulting in a complete lack of response to testosterone. These individuals typically present with female external genitalia despite their male genotype, as testosterone does not influence sexual development. In PAIS, there is a partial response to testosterone, leading to ambiguous genitalia, with a phenotype that may range from predominantly male to predominantly female. MAIS, the mildest form, results in predominantly male genitalia with some degree of feminization. Ambiguous genitalia are often recognized at birth, but in some cases, if undiagnosed, children may be raised as males with a small phallus or as females with clitoromegaly and other atypical genital features. Common clinical manifestations of AIS include bilateral (39%) or unilateral (28%) inguinal hernias, a positive family history (21%), a discrepancy between the karyotype and phenotypic sex detected during amniocentesis (6%), and primary amenorrhea (6%).[5] In some cases, affected individuals may develop breast tissue during puberty despite a male genetic makeup. In cases where clitoromegaly or hypospadias is present, affected individuals may be raised as females, as seen in this case.

In addition to the medical management of AIS, surgical and non-surgical aesthetic procedures play a significant role in the treatment and satisfaction of patients. Genioplasty, a common surgical intervention for individuals with atypical genitalia, has been shown to improve cosmetic outcomes significantly.[6] Studies indicate that both parents and surgeons report high satisfaction with post-operative appearance, but this does not always correlate with functional outcomes or long-term patient satisfaction. The complexities of living with AIS extend beyond physical health. Individuals often face significant emotional, social, and economic challenges. The uncertainty surrounding their gender identity, societal pressure, and the inability to bear children can lead to feelings of isolation and confusion.[7] Managing these challenges requires a multidisciplinary approach, including psychiatrists, gynecologists, and other healthcare professionals. These profes-

sionals play a vital role in providing counseling about the condition, sexual identity, the need for surgical interventions, hormone replacement therapy, and post-operative care.

By emphasizing the obstacles posed by late diagnosis and the intricacies of gender assignment, this case significantly adds to our understanding of AIS management in adults. Unlike complete AIS (CAIS), which is usually assigned female at birth, this case of partial AIS (PAIS) with profound virilization required an approach focused on the individual, resulting in male gender reassignment at age 24. The decision illustrates the importance of individualized care that integrates anatomy, hormones, and psychology. Moreover, the case shows the possibility of surgical procedures in adulthood, such as chordee repair and orchidopexy, which are less addressed compared to the childhood feminizing genitoplasties that receive the majority of attention. In addition, the case highlights the necessity of complex care for all ages that integrates lifelong endocrinology and urology, psychological support, and multidisciplinary genetic counseling for family-oriented cases. In summary, this report continues to underline the differences in AIS presentation while advocating for autonomous management approaches in adult patients.

The literature contains reports of analogous instances of AIS characterized by primary amenorrhoea and the absence of Müllerian structures. Fulare *et al.*[8] wrote about a 17-year-old girl with AIS who had primary amenorrhoea and swelling in both groins. Imaging showed that she didn't have a uterus or ovaries, and that her testes were in the groin area. In that report, the patient underwent bilateral orchidectomy, followed by counseling and hormonal replacement therapy, while maintaining a female gender identity. This shows that AIS management is not the same for everyone and needs to be tailored to each person, especially when it comes to gender assignment and surgical planning, which should be based on the patient's mental health, gender identity, and informed choices. In this case, the patient was evaluated and counseled by a team of experts, and they were okay with being assigned a male gender. They then had surgery to fix the problem, and the outcome was satisfactory and their mental health improved.

The societal concept of gender, which is often viewed as a binary construct, does not always align with the biological diversity observed in individuals with conditions like AIS. External genitalia, hormone profiles, reproductive anatomy, and chromosomal patterns cannot fully define a person's gender identity. It is essential to recognize and respect the complexity and fluidity of gender, especially in the context of AIS, where traditional markers of sex and gender are disrupted.

Conclusion

AIS is a complex condition that impacts physical, emotional, and psychological well-being. Early diagnosis, prompt medical and surgical interventions, and ongoing psychological support are essential for individuals with AIS to lead healthy and fulfilling lives. "Providing multidisciplinary care that prioritizes patient-centered decision-making is crucial for addressing the specific needs of individuals dealing with the realization of having male genetics while identifying as female. This comprehensive approach ensures that their medical, psychological, and social needs are fully met. Such individuals may experience feelings of confusion, distress, and a sense of loss or betrayal about their body and identity. Societal expectations and norms concerning gender and sex can further exacerbate these challenges. Individuals may experience a range of emotions, including shock, anger, sadness, and confusion. These reactions can be exacerbated by feelings of isolation, stigma, or shame, particularly when there is a lack of understanding or support from family, friends, and medical professionals. Therefore, the patient was scheduled with a psychiatrist for counseling. Psychiatric counseling provides essential support, helping individuals navigate their gender identity journey with greater clarity, resilience, and confidence.

During follow-up appointments, the patient conveyed contentment with the gender assignment and surgical results, noting an enhanced quality of life. The post-operative period was uneventful, with no medical issues or difficulties associated to the treatment.

Patient perspective

The treatment process was a life-changing experience for me. At first, I was anxious about the surgeries due to the complexity of my condition. However, the medical team provided constant support, ensuring that I understood every step and allowing me to participate in decision-making actively. I am very pleased with the surgical results, as they have helped me feel more aligned with my true gender and at ease in my body.

Even though I didn't undergo hormonal therapy, I appreciated the personalized care plan, which included thorough follow-up evaluations. The regular check-ups gave me confidence that I was healing properly. Overall, the experience has been positive both physically and emotionally, and I now feel more assured about the future.

Note

Consent was obtained from the patient.

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