British Journal of Healthcare and Medical Research - Vol. 12, No. 04
Publication Date: August 12, 2025
DOI:10.14738/bjhr.1204.19221.
Islam, S. R., Paul, D., & Sarkar, S. A. (2025). Androgen Insensitivity Syndrome: A Case Report. *British Journal of Healthcare and Medical Research, Vol - 12(04).* 242-245.

Androgen Insensitivity Syndrome: A Case Report

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ABSTRACT

A 22 years old married woman presented with bilateral Inguinal hernia. She gave history of primary amenorrhea and primary infertility. She had well developed breast but scanty pubic hair, no axillary hair and normal looking female external genitalia. Ultra sound revealed absence of uterus, short vagina and bilateral inguinal hernia with inguinal testes. Her Serum testosterone level was elevated. Bilateral orchiectomy and hernia repair was done. Her chromosomal analysis revealed 46 XY. It was confirmed that this was case of Complete variety of Androgen Insensitivity Syndrome (CAIS).

Keywords: Androgen insensitivity syndrome, Inguinal testis, Inguinal hernia, Bilateral orchiectomy.

INTRODUCTION

Androgen insensitivity syndrome (AIS) occurs due to target-organ resistance to androgen. Androgen receptors are insensitive to testosterone. As a result, a genetically male with karyotype 46 XY cannot develop male sexual organ and secondary sex characterstics.[1] Female hormone gets dominance and a female secondary sex characteristics develop. The individual continues to be raised as female since birth. AIS is a rare disease with an estimated prevalence of 1 in 100,000 individuals. AIS was first described by Morris, in 1953, with the clinical description of female patients with testes with female phenotype. For this reason, Morris named the condition as testicular feminization syndrome [2]. Later, this syndrome was found to be resulting from a complete or partial resistance of target organ to androgens in a genetically male individual (46 XY). Then it was named androgen insensitivity syndrome[3].The degree of androgen receptors unresponsiveness ranges from mild, partial to complete. That is why this condition is classified to mild androgen insensitivity syndrome (MAIS), partial androgen insensitivity syndrome (PAIS) and complete androgen insensitivity syndrome (CAIS) [4]. AIS is an X-linked recessive genetic disorder. This genetic disorder occurs due to mutation in the

androgen receptor gene (Xq11-q12) in X-chromosome. [5,6]. The diagnosis of CAIS is made following hormonal evaluation, imaging, and genetic studies [7,8]. Principles of management of cases of CAIS include gender assignment, early orchiectomy and estrogen replacement therapy to maintain secondary sex characteristics, bone and cardiovascular health[9,10]. Early orchiectomy is done to prevent malignancy. The other part of the treatment principles are provision for satisfactory vaginal intercourse either by serial vaginal dilation or by vaginal reconstruction and reproduction. Reproduction is limited to adoption only[9].

CASE REPORT

A 22 years old married woman presented with bilateral Inguinal hernia. She gave history of primary amenorrhea and primary infertility. She had well developed breast but scanty pubic hair and no axillary hair. (Fig-1&4) Her external genitalia was very much normal looking. (Fig-1) Testes like lump were palpable in both inguinal area. (Fig-2) Her Serum testosterone level was elevated. Ultra sound examination revealed absence of uterus, short vagina and bilateral inguinal hernia with inguinal testes on both side. Bilateral orchiectomy and hernia repair was done. (Fig-3)Her karyotype analysis was found to be 46 XY. It was confirmed that this was a case of Complete variety of Androgen Insensitivity Syndrome (CAIS).

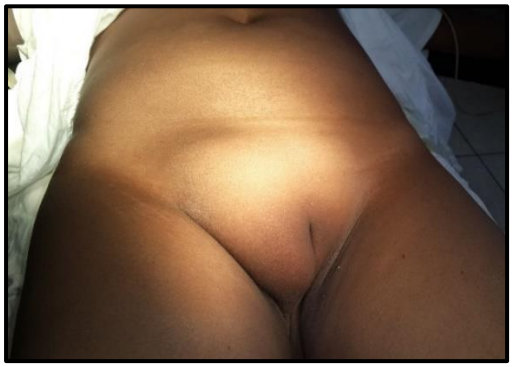

Fig 1: Bilateral Inguinal hernia with bilateral inguinal lump

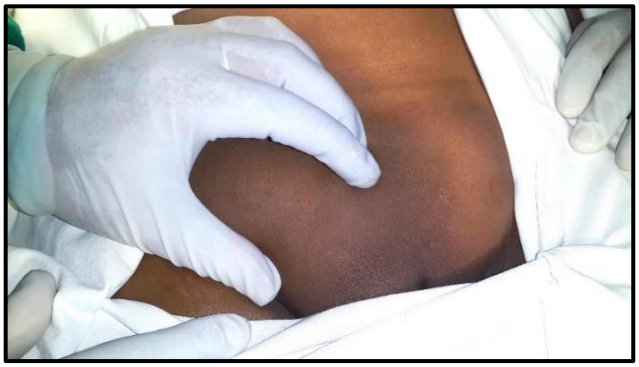

Fig 2: Inguinal testes were easily palpable

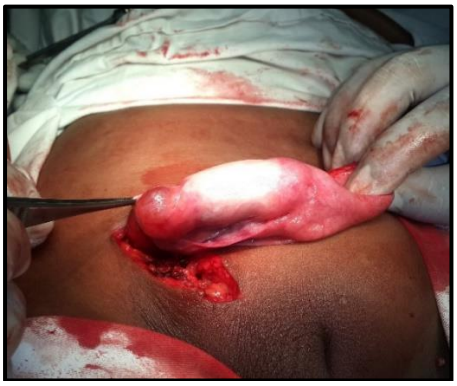

Fig 3: Peroperative view of inguinal testis

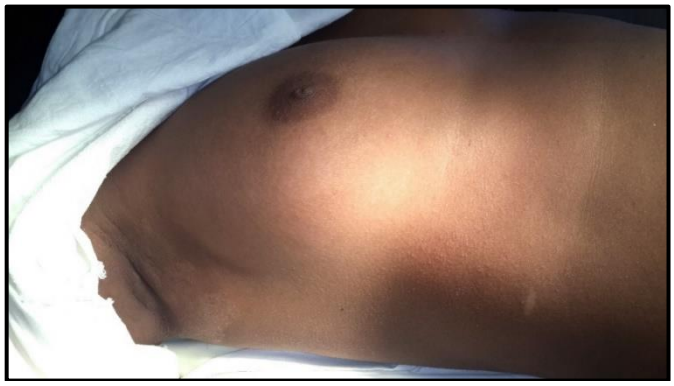

Fig 4: Well developed breast and no axillary hair

DISCUSSION

This patient presented during adult life with normal female external genitalia with bilateral palpable gonads in both inguinal region with inguinal hernia. Abdominal and inguinal ultrasound revealed absence of uterus, short vagina and presence of bilateral testes. It is due to

URL: http://dx.doi.org/10.14738/bjhr.1204.19221.

the explanation that, in complete variety of AIS, Leydig cell secretion of testosterone from testes is normal, which is normally converted into dihydrotestosterone (DHT) through 5- alpha reductase. But the effect of DHT is virtually nil due to the presence of non-functioning androgen receptor (AR). Moreover, anti-mullerian hormone (AMH) secretion from Sertoli cells in the gonads is normally maintained, thus preventing the Müllerian system from developing into a uterus and other internal structures.[11] Therefore, a blind-ending vagina develops. Uterus and other female internal genital organs do not develop. While the gonads (testes) descend independently of androgen.[12] The serum concentration of luteinizing hormone, follicle-stimulating hormone, and testosterone remain elevated or normal.

In adolescent women with AIS, breast and female adiposity develop because estrogens are converted from androgens by the normal functioning of aromatase enzyme. However, pubic and axillary hair is absent or sparse.[13] Individuals with CAIS are mostly raised as females. Orchiectomy after puberty is recommended due to high risk of developing germ cell tumors after adolescence. Risk of developing testicular cancer is considered to be as low as 0.8% to 2.0% before adolescence. Spontaneous puberty occurs when gonads are present in patients with CAIS.[14] After puberty, they require subsequent sustained hormone replacement therapy. After adolescence, the individual raised as female, may need self-dilatation therapy and vaginoplasty procedures to address hypoplastic vagina.[15] Our patient was raised as female and was even married. She was having normal sexual intercourse in her married life before presenting in surgery outpatient with bilateral inguinal hernia. As a result, she did not require any procedure for vaginal dilatation. She was advised to have estrogen replacement therapy only. On follow up it was found that her married life continued as usual even after disclosure of details of the diagnosis to the couple. The couple accepted her permanent infertility.

CONCLUSION

Androgen Insensitivity Syndrome requires expert and sympathetic handling. A team approach involving endocrinologists, clinical geneticists, urologists, gynecologists and psychologists is required. It is very important to do proper counselling and address issues of coping with social stigma. Attention should be paid to timing of orchiectomy and repair inguinal hernia if any. It is also important to ensure proper sexual function and quality of life.

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