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Androgen Insensitivity Syndrome Diagnosed in an Elderly Patient During a Strangulated Inguinal Hernia Repair^{☆,☆☆}



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ABSTRACT

INTRODUCTION: A strangulated inguinal hernia is a common indication for emergency surgery. In comparison, complete testicular feminization is a rare genetic disease that can present with an inguinal hernia because of ectopically positioned testicles.

PRESENTATION OF CASE: A 70-year-old female was admitted to the emergency service complaining of a painful swelling in the right inguinal region for 1 day. The physical examination indicated a strangulated inguinal hernia and surgery was performed. On exploring the inguinal region, a strangulated indirect inguinal hernia and hard 2 × 3-cm mass were detected. The histopathological examination of the excised mass showed testicular tissue, and complete testicular feminization (CTF) was diagnosed after further examinations.

DISCUSSION: Androgen insensitivity syndrome (AIS), the most frequent cause of male pseudohermaphroditism. The diagnosis of patients with AIS is usually made at the beginning of the second decade when a healthy person with a female phenotype complains of no menarche. Making a first diagnosis after the 5th decade is extremely rare.

CONCLUSION: While AIS can be diagnosed in early adulthood, cases might not be diagnosed until the patient is of advanced age.

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1. Introduction

Androgen insensitivity syndrome is an X-linked disorder of sex development. Pathophysiological defects in the androgen receptors result in the resistance of the end-organs to androgens.¹ Although affected individuals typically have a 46XY genotype, the external genitalia are characteristically female, with well-developed labia, breasts, and a vaginal orifice. The diagnosis is generally made during the first decades of life on examining patients with bilateral inguinal hernia or amenorrhea. If an early diagnosis is not made, the possibility of malignancy development increases with age. This article presents a case of geriatric complete androgen insensitivity syndrome diagnosed incidentally following strangulated inguinal hernia surgery, in which malignant development was not seen in the excised testicle tissue.

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2. Case report

A 70-year-old-female was admitted to the emergency service complaining of swelling in the right inguinal region. This swelling had been present intermittently for 1 year, but the patient had been able to reduce it; this time, however, it had not been reduced since the previous day. The physical examination suggested a right strangulated inguinal hernia and emergency surgery was performed.

On exploring the inguinal region, the cord structure was similar to the male anatomy and a strangulated indirect inguinal hernia was diagnosed. When a herniotomy was performed, dark, non-ischemic omentum was observed in the hernia sac. The omentum was reduced into the abdomen and high ligation was performed. Given the male anatomical appearance, a detailed exploration showed a solid 2 × 3-cm mass (Fig. 1) adhering to the medial wall of the inguinal canal distally; this mass was excised. The patient's postoperative recovery was uneventful and she was discharged on the second day. Histopathologically, the excised mass was testicle tissue (Fig. 2).

On further questioning, the patient had been married for 35 years, had never menstruated, never became pregnant, and had adopted one child. The physical examination showed normal breast development, but reduced auxiliary and pubic hair (Fig. 3). The gynecologic examination, pelvic ultrasonography, and magnetic

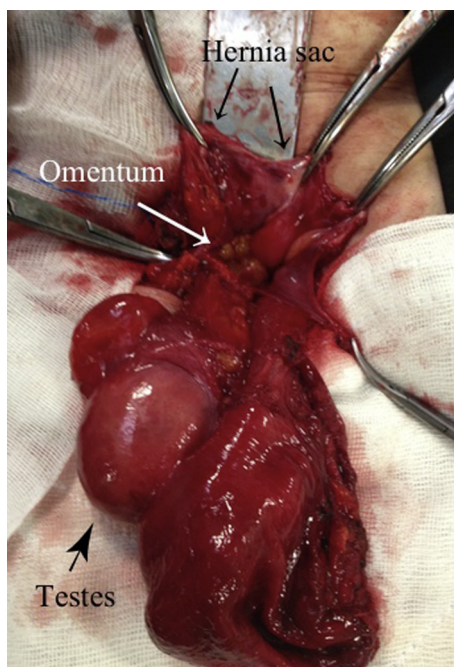


Fig. 1. Operative findings.

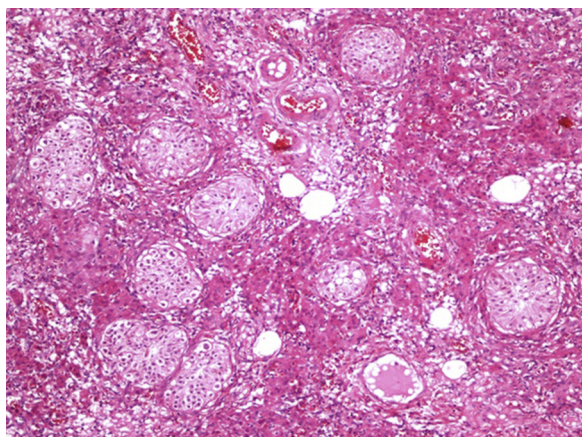


Fig. 2. Microscopic appearance of the excised mass (testicular tissue) showing tubular structures composed of vascular stroma and Leydig cells (H&E 10×).



Fig. 3. The breasts and external genitalia appeared normal, but the axillary and pubic hair was sparse.



Fig. 4. MRI shows a second testis in the left inguinal canal (arrowheads), a blind vagina (arrows), and no uterus or ovarian tissues in (a) transverse and (b) coronal cross-sections.

resonance imaging (MRI) showed a blind vagina, no uterus or ovarian tissue, and a second testicle in the left inguinal canal (Fig. 4). Laboratory examinations showed dehydroepiandrosterone (DHEA)-S 52.6 $\mu\text{g/dl}$, testosterone 1.33 ng/dl , follicle-stimulating hormone (FSH) 55.48 mIU/ml , luteinizing hormone (LH) 30.57 mIU/dl , and prolactin 7.13 ng/ml . The patient's karyotype was 46 XY. Consequently, a diagnosis of complete androgen insensitivity syndrome (testicular feminization) was made. Excision of the left testicle was recommended, but the patient refused.

3. Discussion

Androgen insensitivity syndrome (AIS), the most frequent cause of male pseudohermaphroditism, is rare with an estimated incidence between 1/20,000 and 1/60,000.² The syndrome is graded as complete, partial, or mild according to the level of androgen resistance. The complete form is also called testicular feminization. In the partial forms, the external genitalia may show hypospadias and micropenis, cryptorchidism with bifid scrotum, or a female phenotype with cliteromegaly.³

In the complete form, the Müllerian structures regress because of the Müllerian inhibiting substance excreted by the gonads (testes). Therefore, the fallopian tubes, ovaries, and proximal vagina cannot develop. Despite the excreted testosterone, the existence of end-organ resistance prevents male-oriented development, such as pubic and auxiliary hair growth. As the increased testosterone is converted into estradiol, there is female-oriented breast development.⁴ In testicular feminization, the gonads are testicles, which can be located in the abdomen, inguinal canal, duct, or labia major. In our case, external findings of complete androgen insensitivity syndrome were present.

The diagnosis of patients with androgen insensitivity syndrome is usually made at the beginning of the second decade when a healthy person with a female phenotype complains of no menarche. It can also present with the formation of bilateral inguinal hernia in a female neonate or child or in a woman in her 4th or 5th decade who has never menstruated, or with the development of a neoplastic inguinal or pelvic mass in the abnormally located testicular tissue.^{2,5,6} Making a first diagnosis after the 5th decade is extremely rare. Our patient was a 70-year-old who developed a unilateral inguinal hernia and testicular tissue was found during the surgery.

Although there is a possibility of malignant development in androgen insensitivity syndrome, the true risk is not known since there have been very few cases. The risk of prepubertal malignant transformation is low, but it increases with age, and reaches a maximum of 33% after 50 years of age.⁷ The malignancy in AIS can arise from testicular germ cells, testicular stromal cells, or other mesenchymal tissues.^{8,9} Given the possibility of malignant transformation, a gonadectomy must be performed in patients with AIS,

after the spontaneous termination of puberty.¹⁰ This is because puberty and sexual maturation in these patients commence at the same age, and the sexual maturation stages are the same as in normal female children; additionally, the risk of prepubertal malignant transformation is low. In addition, these patients must be given genetic and psychological support because the psychological consequences can be enormous at any age. Although our patient was 70 years old and the possibility of malignancy was explained, she was not able to accept her situation and refused surgery to excise the other testicle.

In conclusion, while AIS can be diagnosed in early adulthood, cases might not be diagnosed until the patient is of advanced age. Nevertheless, these elderly patients also need psychological support, in addition to surgical treatment.

Conflict of interest

The authors declare that they have no conflict of interest.

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None.

Ethical approval

We have obtained written consent from the patient for publication of this case report and accompanying images.

Author contribution

Yusuf Arslan and Orhan Veli Ozkan contributed to study design. Yusuf Arslan, Omer Yalkin and Yasemin Gunduz contributed to data

collection. Yusuf Arslan, Fatih Altintoprak and Zeynep Kahyaoglu contributed to data analysis. Yusuf Arslan and Fatih Altintoprak contributed to writing.

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