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CASE REPORT

Laparoscopic Gonadectomy for Complete Androgen Insensitivity Syndrome

Lakkanna Suggaiah, Usharani Rathnam, Preetham Raj

ABSTRACT

Introduction: Androgen insensitivity syndrome is a form of male pseudohermaphrodite where the phenotype female has male gonads and is genotypically male.

Case report: We report a case of complete androgen insensitivity syndrome in a 22-year-old who underwent laparoscopic gonadectomy.

Discussion: Androgen insensitivity syndrome is the most common cause of male pseudohermaphroditism and third most common cause of primary amenorrhea.

Conclusion: Laparoscopy is an effective method in androgen insensitivity syndrome treatment, mainly due to the increased risk of malignant transformation of the testes. Psychosexual needs should be addressed along with low-dose hormonal therapy to maximize long-term success.

Keywords: Androgen insensitivity syndrome, Laparoscopic gonadectomy.

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INTRODUCTION

Androgen insensitivity syndrome (AIS) was previously called testicular feminization syndrome. Intersex is a rare X-linked recessive condition due to mutation at xq 11-q 12 localization on the androgen receptor gene. AIS is a form of male pseudohermaphrodite where the phenotype female has male gonads and is genotypically male. Importance of this syndrome is development of testicular tumor especially seminoma after puberty. The diagnosis is often based on absence of uterus, cervix, tubes and a vagina of variable length with nondysplastic testis.

CASE REPORT

A 22-year-old woman was admitted to ESIC Medical College PGIMSR, Rajajinagar, Bengaluru, with complaints of primary amenorrhea and infertility, referred to the surgical department by gynecology department for bilateral inguinal swelling. The patient was 175 cm tall, weighing 60 kg. External physical examination revealed well-developed breasts, abundant scalp hair with scanty pubic and axillary hair (Fig. 1). The vulva and perineum appeared normal and the vagina measured 5 cm in length, ending blindly. Family history revealed she was the only child with no similar complaints in the family. Transabdomino-pelvic ultrasound confirmed the absence of uterus and ovary and presence of bilateral masses of 3 × 2.5 cm in size, located near the internal ring of the inguinal canal. Imaging studies noted absence of prostrate and seminal vesicals.

Karyotype report was 46 XY (Fig. 2). Serum FSH was 10.7 μ/ml, serum estradial was 88 pg/ml, all other hormonal parameters and tumor markers were within normal limits. After standard peroperative preparation, operative laparoscopy was performed under GA. Pelvic and abdominal inspection revealed absent uterus and ovaries. Bilateral gonads appearing as testis were attached near the internal ring of both inguinal canals. The pedicles of gonads were coagulated with bipolar cautery and cut with laparoscopic scissors to prevent the spillage of cells and contamination. The gonads were placed in endobags and removed intact after extending the port (Figs 3 and 4). There was no complication during the operation. The patient was discharged on the 3rd postoperative day after surgery.

Gross pathology reports mentioned the tissue marked as the right gonad measuring 3 × 5 × 2 cm with attached

Fig. 1: Thick long hair

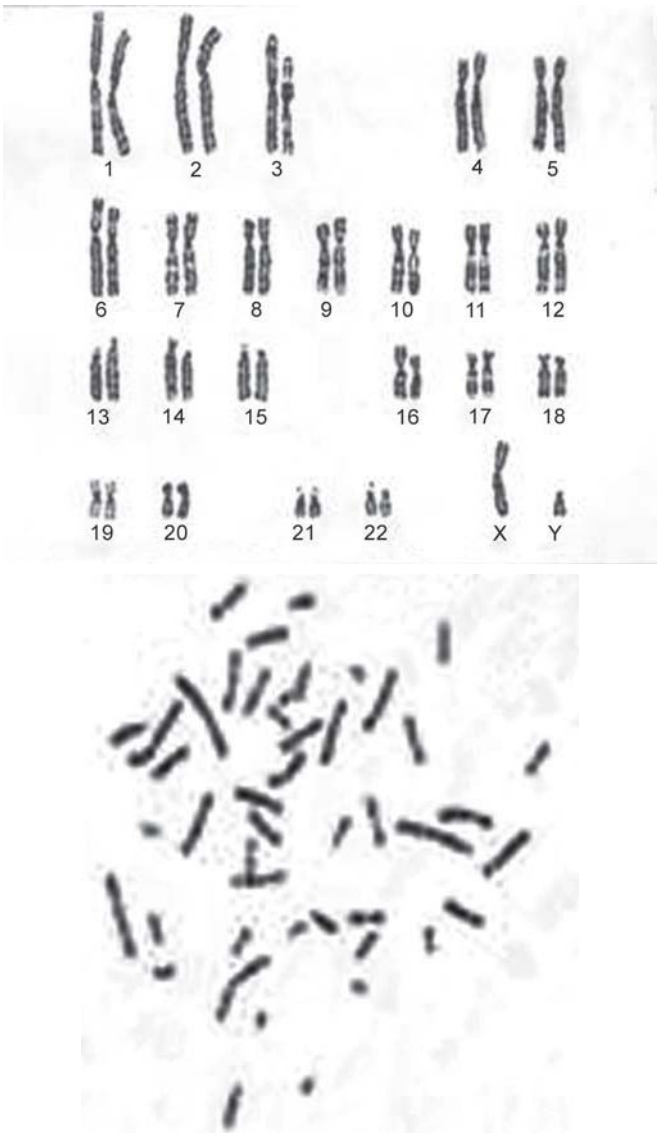

Fig. 2: Karyotype analysis—male karyotype-46XY

duct and the left, 3 × 2 × 2 cm. Bilateral sertoli hyperplasia was noted on histology (Fig. 5).

In view of this, long-term conjugated estrogen 0.625 mg per day was started.

DISCUSSION

AIS is the most common cause of male pseudohermaphroditism and third most cause of primary amenorrhea.[2] It is also known as testicular feminization syndrome. Intersex male pseudohermaphroditism has an incidence of 1 in 20,000 to 64,000 male births.[1,2] John Morris (1953) described the anatomical, histological and clinical features based on 82 cases collected from over nearly 150 years of medical literatures.[3]

The typical mode of presentation is an adolescent female, who has breast development with the pubertal growth but has not attained menarche with absent or scanty pubic and axillary hair. Complete androgen insensitivity syndrome (CAIS)[4] may also be present in early infancy with bilateral inguinal or labial swellings. Bilateral inguinal hernias are rare in girls and it has been estimated that 1 to 2% of such cases have CAIS.[5] In review of literature, the case of CAIS in a 22 years female were the presence of testis, prostatic tissue, seminal vesicals which was confirmed by ultrasound of abdomen, hormonal analysis, operative findings and HPE.[6] Diagnosis of CAIS is usually with the absence of female internal genital organs on physical examination aided by pelvic ultrosonography, karyotyping, molecular genetic testing of the AR gene mutation (chromosomal locus xq 11q 12) and elevated testosterone LH level.[5] In our case,

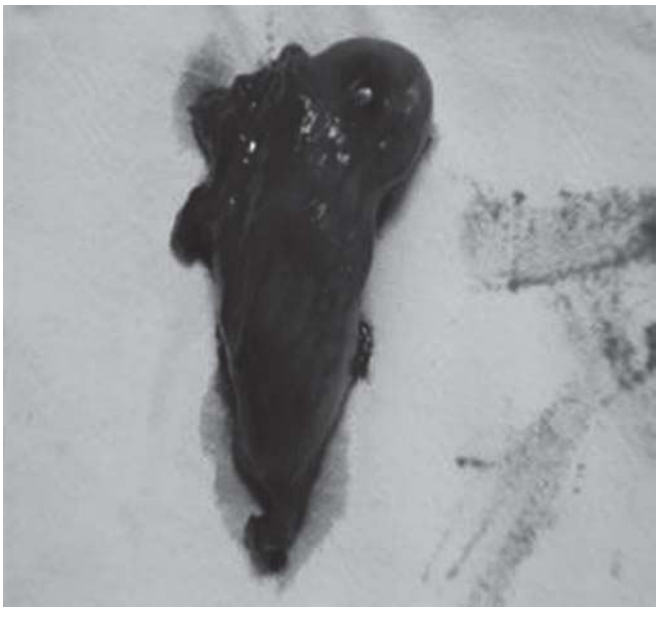

Fig. 3: Left gonad

Fig. 4: Right gonad

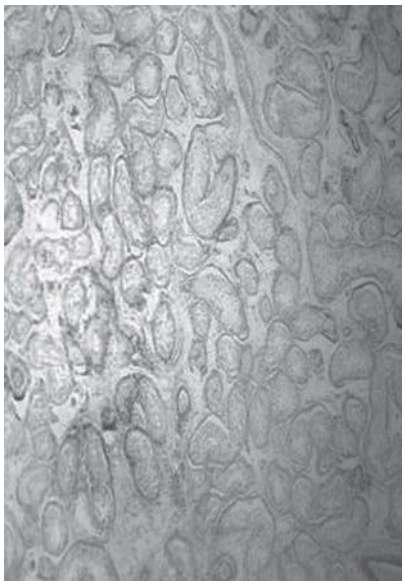

Fig. 5: HPE-sertoli cell hyperplasia

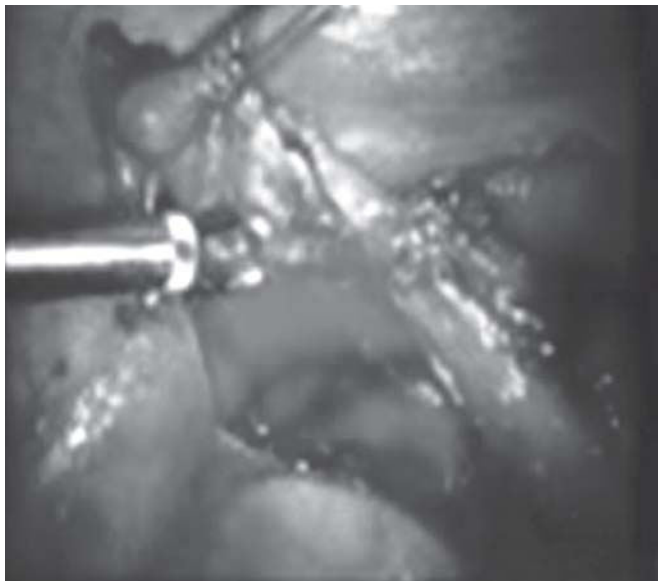

Fig. 6: Laparoscopic gonadectomy

the diagnosis of CAIS was based on gynecological examination, laparoscopy and the karyotyping.

Partial androgen insensitivity syndrome (PAIS) is another category of intersex and is characterized by perineoscrotal hypospadiasis, micropenis and bifid scrotum. The testis may also be undescended. The most severe form of PAIS presents as isolated clitromegaly.[7]

Mild androgen insensitivity syndrome (MAIS) as a category of AIS was realized following investigation for male factor infertility which suggested defect in the androgen action leading to oligospermia with normal level of testosterone and increased life span.[7]

Gonadal tissue can be located in the inguinal canal or any where in the abdomen-sites that are invisible during laparoscopy. MRI has proven of value, for localization of nonpalpable undescended testis.[8] There is increased risk of dysgenetic gonads developing malignancy, which can be as high as 30%. In contrast to the other forms of gonadal dysgenesis, the incidence of tumors in AIS cases is rare before puberty and significantly higher after the age of 35 years.[6] Prophylactic gonadectomy is necessary in the postpuberty period to allow the development of the secondary sexual characters during puberty.[2,9] Laparoscopic removal of gonads has many advantages compared to laparotomy, foremost being minimal blood loss, rapid recovery, shorter hospital stay and minimum psychological trauma. Laparoscopy has better visualization of the entire abdomen and pelvis compared to laparotomy[10] (Fig. 6). Patients should be treated with long-term hormone replacement therapy (HRT) after gonadectomy.[1,2,11] Androgen supplementation is not useful because of insensitive androgen receptor.[12]

CONCLUSION

AIS should be suspected in cases with primary amenorrhea. Laparoscopic gonadectomy can be performed safely via a small caliber laparoscopy after puberty with long-term low dose hormone therapy because of the increased risk of malignant transformation of the testicles. Attention to psychological consideration in such patient is important to maximize long-term success.

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