

Giant Inguinal Hernia Repair Leading To the Diagnosis of Complete Androgen Insensitivity Syndrome in an Elderly Lady

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Abstract

Complete androgen insensitivity syndrome (CAIS) is an X linked disorder characterized by lack of mullerian derivatives, absent uterus, normal breast development and sparse or absent axillary and pubic hairs. Mutation in the androgen receptor is the cause for this rare disorder. Classically diagnosis is made when evaluating for primary amenorrhea in a young girl. These patients are at higher risk for development of gonadal malignancy the risk of which increases with increasing age. Bilateral gonadectomy after puberty is recommended. We are presenting a case of CAIS who was diagnosed at the age of 60 years during surgery for giant inguinal hernia.

Introduction

Complete androgen insensitivity syndrome (CAIS) is a rare disorder characterized by a 46 XY karyotype, negative sex chromatin, bilateral undescended testes, female external genitalia and lack of mullerian derivatives [1]. These patients are phenotypically females but without uterus and a blind shortened vagina. The typical presentation of CAIS is either as an infant with inguinal swelling or a pubertal girl with primary amenorrhea. Presentation in older age group is very unusual particularly in Hindu society where a female has to be a part of a number of rituals. We are presenting a case of CAIS that was detected during repair of a giant inguinal hernia in an elderly lady.

Case Report

An otherwise healthy 60 year old lady presented with history of left groin swelling of 5 year duration (Figure 1A). On examination, she was found to have a 25 × 20 cm left inguinal swelling extending upto the knee joint with clinical signs diagnostic of inguinal hernia (Figure 1B). There was an ulcer of size 5 × 5 cm on the most dependant part of the swelling. In view of large size of the hernia, computed tomography of the abdomen (CECT) was performed. It revealed characteristic features of inguinal hernia along with a cystic lesion in the hernia sac. She was planned for inguinal hernia repair. Intra operatively, there was sliding hernia with sigmoid colon as its content. The distal cystic lesion that was seen on CECT abdomen was found to be hydrocele! (Figure 1C) On detailed examination, she was found to have enlarged clitoris and blind ending vagina (Figure 1D). Review of CECT abdomen revealed absence of uterus. These findings confirmed this as a case of CAIS. Mesh hernioplasty was performed along with repair of right inguinal hernia and right orchidectomy. Patient tolerated the prolonged surgery well and her post operative recovery was uneventful. Patient later revealed that she never had menstruation till the age of 60 years. Histopathological examination of bilateral testis revealed no evidence of any malignancy.

Discussion

Androgen insensitivity syndrome is an X linked recessive disorder resulting from mutation in the androgen receptor. Depending on the phenotype, 3 different types are described: complete androgen insensitivity syndrome (CAIS), ambiguous genitalia partial androgen insensitivity (PAIS) and minimal androgen insensitivity form (MAIS) [2]. The fundamental difference between these subtypes is that in CAIS the external genitalia is that of a normal female, in MAIS the external genitalia is that of a normal male, and in PAIS the external genitalia is

partially, but not fully masculinized. Both individuals with PAIS and CAIS have 46 XY karyotypes [3-5].

CAIS first described by Morris in 1953, is the most common form of male pseudohermaphroditism [6]. It is synonymous with testicular feminization syndrome. The estimated prevalence of this disorder was 1:20,000 to 1:64,000 live male births [7]. Complete androgen insensitivity syndrome (CAIS) is a sex-linked recessive disorder caused by a mutation in the androgen receptor gene [8]. Location of the mutation is at Xq11-q12 in 95% cases. This leads to incomplete development of Wolffian duct and hence incomplete differentiation of male external genitalia. In addition, Mullerian ducts regress because of the presence of anti-Mullerian hormone produced by the sertoli cells [9]. As a result, these patients exhibit female phenotype. However, serum androgen level is comparable with that of a normal male [10].

CAIS is typically diagnosed at puberty in phenotypically female patients with primary amenorrhea due to the absence of female internal genitalia. These patients are phenotypically females. They have normal breast development and growth spurt at the appropriate age. This is related to aromatization of androgens which are responsible for development of secondary sexual characteristics. However these patients have absent or sparse pubic and axillary hairs [11]. Ultrasound examination of the pelvis usually shows absence of mullerian derivatives and vaginal examination reveals a blind-ending vagina [10]. The present patient had all these typical characteristics except for the fact that she presented at the age of 60 years. Although CT abdomen was done, absence of uterus was never paid attention in view of her age and giant inguinal hernia. Post operatively when CT abdomen was reviewed, it was clear that she had all the typical features of CAIS.

Testosterone via androgen receptors plays an important role in the descent of testis. In CAIS because of the defect in androgen

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Figure 1A: Patient profile.
Figure 1B: Giant inguinal hernia with pressure ulcer on the dependent portion.
Figure 1C: Hydrocele found in the hernia sac on left side.
Figure 1D: Clittoromegaly detected after hernia repair.

receptor, descent of testis is hampered and as a result an undescended testis is quite common in this patient population. As a result they are vulnerable to indirect inguinal hernia. About 50% of patients with CAIS have an inguinal hernia. Conversely, 1-2% of apparently female infants with inguinal hernia are found to have CAIS [12]. Risk of gonadal malignancies is also high in these patients with CAIS because of undescended testes. Incidence of germ cell tumors is 3.6% and 33% at 25 years and 50 years of age respectively [13]. Common testicular tumors seen in this syndrome are germ cell tumors (most commonly seminoma), sex cord tumors, sertoli cell tumors, leydig cell tumors and hamartomas [14]. The risk of malignancy is inversely proportional to the age.

Bilateral orchidectomy is recommended in these patients soon after the completion of puberty to decrease the risk of malignancy. This timing is appropriate as the risk of malignant transformation before puberty has been estimated to be 0.8-2% [15,16]. In addition, estrogen produced from testes is essential for the development of a normal female phenotype as well as secondary sexual characteristics. After orchidectomy, hormone replacement therapy is mandatory to improve bone health and general wellbeing [17].

The present patient revealed that she thought she was “Different”, but in view of quite orthodox family she could never discuss her problem with anybody. Only when she developed pressure ulcer over the hernia and there was foul smelling discharge, she sought consultation and even

then never revealed her menstrual status. This stresses the importance of need for public awareness in India regarding intersex which is still considered a taboo.

Conclusion

The purpose of this report is:

- To notify that CAIS can present even in elderly.
- To stress the need to improve awareness as well as understanding among the general public and practitioners regarding the intersex disorders.

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