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 amenorrhoea 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 amenorrhoea. 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 up to 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 cisterna 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 testes 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 amenorrhoea 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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