Journal of Pediatric Urology (2013) 9, e31—e34

Journal of Pediatric urology

CASE REPORT

Complete androgen insensitivity syndrome associated with bilateral sertoli cell adenomas and paratesticular leiomyomas: Case report and review of the literature

S. Siminas [a,*], G. Kokai [b], S.E. Kenny [c]

[a] *Alder Hey Children's NHS Foundation Trust, Department of Paediatric Surgery and Urology, UK*
[b] *Alder Hey Children's NHS Foundation Trust, Department of Paediatric Histopathology, UK*
[c] *Alder Hey Children's NHS Foundation Trust, Department of Paediatric Surgery and Urology and University of Liverpool, UK*

Received 29 May 2012; accepted 22 June 2012
Available online 21 July 2012

KEYWORDS
Complete;
Androgen;
Insensitivity;
Syndrome;
Sertoli cell adenoma;
Paratesticular leiomyoma

Abstract *Background:* Complete androgen insensitivity syndrome (CAIS) is a rare and usually unexpected cause of primary amenorrhoea that results from receptor resistance to androgens, producing a female phenotype in genetically male patients.
Case: A 16-year-old girl was diagnosed with CAIS after investigations for primary amenorrhoea. Her left inguinal gonad and the right intra-abdominal gonad were resected and histopathology revealed the presence of dysgenetic multinodular testes with absence of germ cells, significant hyperplasia of Sertoli cells (Sertoli cell adenoma) and presence of paratesticular leiomyomas.
Conclusion: Although the risk of gonadal tumour development is considered to be low, a variety of tumours have been described in association with CAIS, but this is the first report of development of bilateral paratesticular leiomyomas developing concurrently with Sertoli cell adenomas.

Introduction

Androgen insensitivity syndrome (AIS) is an X-linked disorder of sex development (DSD) that arises as a result of receptor resistance to androgens and produces a phenotype with varying degrees of feminization depending on the severity of receptor insensitivity [1] In patients affected by the syndrome, the risk for developing gonadal tumours is variable, depending on the degree of virilisation [1,2]. We present the case of a 16-year-old girl with the complete form of the syndrome (CAIS) who presented with primary amenorrhoea and bilateral Sertoli cell

* Corresponding author. Tel.: +44 1512824597; fax: +44 7786373981.
E-mail addresses: siminas1@gmail.com, sotirios.siminas@alderhey.nhs.uk (S. Siminas).

1477-5131/$36
http://dx.doi.org/10.1016/j.jpurol.2012.06.013

adenomas and paratesticular leiomyomas in her resected gonads.

Case report

A 16-year-old girl was referred by her primary care physician to the gynaecology services of a regional hospital due to primary amenorrhoea. She was an otherwise healthy adolescent with no significant previous medical history. On clinical examination, she had normal stature for her age, breast development appropriate to Tanner stage and scanty pubic hair. External genitalia appeared normal, but a palpable mass could be felt in the left groin. Ultrasonography (US) of the abdomen and inguinal regions demonstrated the absence of ovaries and müllerian structures, the presence of the vagina which measured approximately 5 cm in length, and a solid mass in the left inguinal region measuring approximately 3 cm in diameter, also containing some small peripheral cystic areas and a normal looking intralesional lymph node. The appearance of the lesion was highly suggestive of a dysgenetic gonad. The right gonad could not be visualized. A karyotype revealed the presence of 46XY complement and the diagnosis of CAIS was established and the patient was referred to the Disorders of Sex Development (DSD) Clinic.

Due to the atypical appearance of the left gonad, a left groin exploration was performed in the following days and left gonadectomy was performed. Macroscopically the gonad had the appearance of a testis with the presence of three nodules on the surface and the epididymis was blind-ending with cystic dilatation (Fig. 1). Histopathology of the lesion revealed the presence of immature testicular tissue with multiple nodules that contained only Sertoli cells, absence of germ cells and a large number of seminiferous tubules (Sertoli cell adenoma) (Fig. 2). The intervening interstitium contained large numbers of Leydig cells. The macroscopically nodular areas outside the area of the testis contained smooth muscle fibres with strongly positive labelling for smooth muscle actin (paratesticular leiomyoma) (Fig. 3). The intralesional lymph node exhibited mild lymphoid hyperplasia and otherwise normal architecture. Postoperative course was uneventful.

Hormone levels demonstrated persistently increased testosterone levels, indicating the presence of testicular tissue, most likely intra-abdominally. Meetings were held with the patient and family, as well as clinical psychologists, in order to explain the diagnosis and involve the patient in the decision-making regarding the fate of her right gonad. She coped very well with the situation and eventually opted for a laparoscopic right gonadectomy after approximately one year which was carried out uneventfully as a day-case procedure. Metachronous excision of gonads occurred to allow adequate time for full explanation of the condition to the patient prior to contralateral gonadectomy. The excised intra-abdominal gonad exhibited almost identical findings to the inguinal one, specifically Sertoli cell adenoma and separate extra-testicular areas of leiomyoma.

The patient was started on hormone replacement treatment and is being followed-up regularly in the Disorders of Sex Development (DSD) clinic.

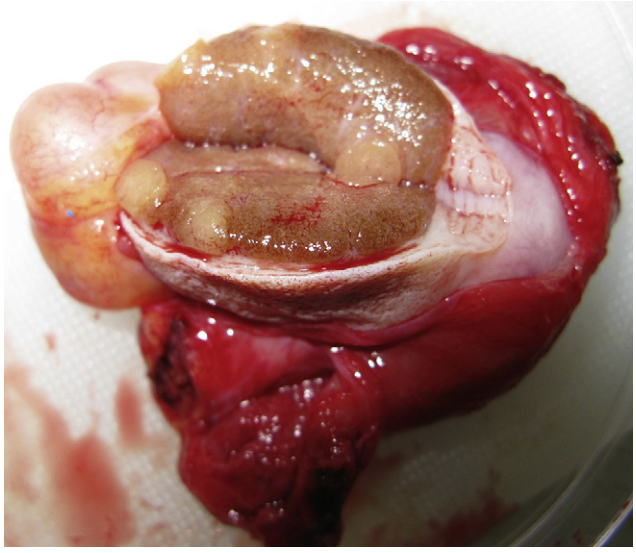

Figure 1 A longitudinally opened left testicular remnant containing several well demarcated bulging pale yellowish nodules (Sertoli cell adenomas) within the testicular tissue. Note the lobulated larger pale tumour nodule on the left side of the sample (leiomyoma).

Discussion

CAIS is considered to be a rare anomaly (approximately 1/20,000 to 1/100,000 genetic males), although the prevalence data are based on small series and can be inaccurate [3,4]. The androgen receptor (AR) gene is located in the long arm of the X chromosome (Xq11-12), and so far, over 200 mutations of the gene have been described, including complete or partial deletions, point mutations, and insertions, all leading to loss of function [1] Receptor loss of function leads to a variety of undervirilisation phenotypes depending on the degree of receptor function loss. An essential common feature in AIS is the regression of the müllerian duct derivatives,

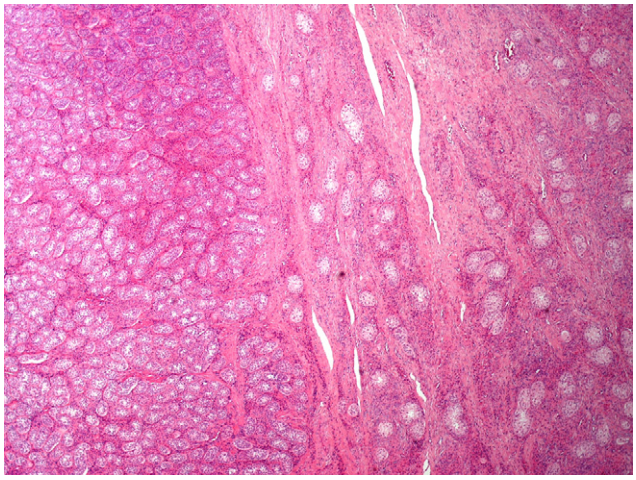

Figure 2 Routine H&E staining of the testis from the boundary zone between the tumour nodule Sertoli adenoma (left) and the background testicular tissue (right).