THE JOURNAL OF OBSTETRICS AND GYNECOLOGY OF INDIA

The Journal of Obstetrics and Gynecology of India (May–June 2012) 62(3):332–333
DOI 10.1007/s13224-011-0112-x

CASE REPORT

The XY Female (Androgen Insensitivity Syndrome)—Runs in the Family

Dey Ramprasad · Biswas Subhash Chandra · Chattopadhvav Nibedita · Gupta Dipankar · RoyBiswas Rami · Mukhopadhyay Arunima

Received: 22 February 2008 / Accepted: 29 April 2010 / Published online: 7 March 2012

Introduction

Androgen insensitivity syndrome (AIS) was first described in details by Morris [1], who provided the descriptive terms—testicular feminization syndrome for this disorder, which is inherited as X-linked recessive disorder. The underlying pathology is the inability of the end organs to respond to androgens, either due to lack of androgen cytosol receptor or defect in the receptor. Genotypically they are male (XY) but phenotypically and psychologically female [2], usually present with primary amenorrhoea or infertility with well developed breast, but absent axillary and pubic hair, normal external genitalia and short and blind vagina. The upper two-thirds of vagina, uterus and tubes are absent. Gonads (testes) are normally developed but abnormally positioned; either placed in the labia, or inguinal canal or intra-abdominal. The hormone profile in these individual is typical—high LH, normal to slightly elevated testosterone levels, high estradiol (for men), and normal to elevated FSH.

Case Reports

Miss N.J. 20 years of age attended on 23/08/05 with history of primary amenorrhoea and left sided inguinal swelling since birth. She has two younger sisters, one of 18 years of age and other 13 years of age, both of them also having primary amenorrhoea.

Examination revealed average built (height 145 cm, weight 54 kg), absent axillary and pubic hair and breast (Tanner stage III). Abdomen was soft with no organomegaly. There was a non-tender oblong shaped mass of 3 cm × 2 cm size in the left inguinal region, which became prominent on coughing and reduced in size with change of posture and pressure. She had normal looking external genitalia but vagina ended in a blind pouch of about 3–4 cm in length.

Routine investigations like hemogram, blood sugar, urea, and serum creatinine were within normal limits. Hormone estimation showed F.S.H.—3.8 IU/l, L.H.—15.9 IU/l,

Dey R., RMO cum Clinical Tutor · Biswas S. C., Associate Professor · Chattopadhvav N., Assistant Professor
Department of Gynaecology & Obstetrics, IPGMER & SSKM Hospital, Kolkata 700 020, India

Gupta D., Assistant Professor
Department of Paediatrics, IPGMER & SSKM Hospital, Kolkata 700 020, India

RoyBiswas R., Assistant Professor
Department of Pathology, CNMC&H, Kolkata, India

Mukhopadhyay A., General Surgeon
ESI Hospital, Kalyani, WB, India

Dey R. (✉), RMO cum Clinical Tutor
836 Block-P, New Alipore, Kolkata 700 053, India
e-mail: ram_aranima@yahoo.co.in

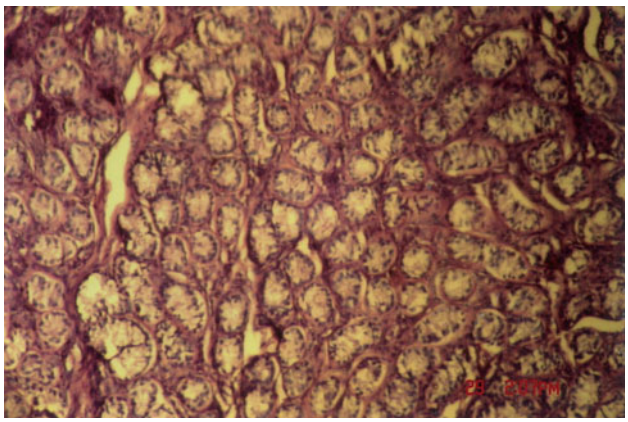

Fig. 1 Histopathological examination of excised gonads showing closely spaced seminiferous tubules with marked loss of germ cells and presence of Sertoli cells and clusters of Leydig cells in the stroma

prolactin—6 ng/ml, T3—150 ng/dl, T4—8.4 μg/dl, T.S.H.–2.9 μU/ml.

Ultrasonography of lower abdomen revealed no uterus, presence of right gonad of 4 cm × 2 cm in size but left gonad not visualized. FNAC from left inguinal swelling showed the cells resembling of germ cells and the features suggestive of testicular tissue. Buccal smear for Barr body was negative. Diagnostic Laparoscopy was done on 17 September 2005, showed absence of uterus and adnexa but presence of both the gonads and each were about 4 cm × 2 cm in size. Chromosome analysis showed 46 XY karyotype. She underwent laparotomy followed by bilateral gonadectomy with left sided herniotomy on 23 September 2005.

Histopathological examination of both the removed gonads showed similar features and consist of closely spaced seminiferous tubules in which there was marked loss of germ cells and presence of Sertoli cells and clusters of Leydig cells in the stroma (Fig. 1). She was discharged from hospital with a final diagnosis of complete AIS and was asked to bring her sister Miss N. N. who was 18 years old with primary amenorrhoea.

On examination she also had similar phenotype and secondary sexual development. Abdomen was normal with no organomegaly and normal hernial orifices. She had infantile external genitalia with a blind vagina of 2–3 cm in length. USG showed no uterus but both the gonads were seen and each were about 4 cm × 2 cm in size. Karyotype was 46 XY. Bilateral gonadectomy were performed and histopathological report showed immature testicular tissue with Leydig cell hyperplasia.

Their youngest sister Miss N.K. 13 years of age till not attended menarche, clinically diagnosed as a case of AIS, declined to come forward to be investigated for primary amenorrhoea because of financial constraints and as there was no apparent health or reproductive benefit.

Discussion

Androgen insensitivity accounts for about 10% of all cases of primary amenorrhoea, the third most common cause after gonadal dysgenesis and congenital absence of vagina [3]. Because of the importance of prophylactic gonadectomy, detection of this syndrome demands careful investigation for other affected family members and pedigree analysis [4]. Apparent sisters of affected individual have a one in three chance of being XY and female offspring of a normal sister of an affected individual have one in six chance of being XY [3]. Many of the affected individual have atypical karyotyping [5]. About a third of the patients have negative family histories and presumably represent new mutations. In two-thirds of all cases, these mutations are inherited from the mother, while the rest occur as a result of a spontaneous mutation in the egg/zygote (AIS support group UK 1999) [6].

References

1. Morris JM. The syndrome of testicular feminization in male pseudohermaphrodites. Am J Obstet Gynecol. 1953;65:1192–211.
2. Ahlquist JA. Gender identity in testicular feminization. BMJ. 1994;308:1041.
3. Speroff L, Fritz MA. Clinical gynecologic endocrinology and infertility. 7th ed. Philadelphia: Lippincott Williams & Wilkins; 2005. p. 339–40.
4. Jophy R, Padmashri V, Jairaj P. Testicular feminization syndrome. J Obstet Gynecol Ind. 2002;52:153.
5. Goswami S, Gutgutia R, Mitra A, et al. Testicular feminization with atypical karyotyping. J Obstet Gynecol Ind. 2002;52:112.
6. Menakaya UA, Aligbe J, Iribhogbe P, et al. Complete androgen insensitivity with persistent mullerian derivatives. J Obstet Gynaecol. 2005;25:403–5.