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Familial Swyer syndrome in two sisters with undeveloped uterus

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Introduction

Swyer syndrome is characterised by 46,XY karyotype and a female phenotype with normally developed Müllerian ducts, streak gonads, minimal breast enlargement, elevated gonadotropins, low levels of oestrogens, normal levels of androgens and primary amenorrhoea. Here we present two sisters with complete gonadal dysgenesis, one of which remained amenorrheic, despite the oestrogen treatment.

Case report

A 20-year-old female was admitted to our clinic with the complaining of absence of menstrual bleeding and growth retardation of secondary sex characteristics. She first presented with primary amenorrhoea at age 16 years to a different clinic. Her serum FSH level was 193 mIU/ml, LH level was 54 mIU/ml; her $E_2 < 20$ pg/ml. She was given cyclic hormone therapy and responded well to the treatment with menstrual withdrawal bleeding. On physical examination, she had Tanner stage 2 breast development, a normal vagina with Tanner stage 4 pubic hair development. Genetic analysis revealed her karyotype to be 46,XY and after microdeletion studies of AZF (SY81, SY86, SY84, SY182, SY121, SYPR3, SY124, SY127, SY128, SY130, SY133, SY134, SY145, SY152, SY242, SY208, SY254, SY255, SY157) and SRY (SY14) gene, no mutation was identified. Pre-operative pelvic ultrasound revealed a hypoplastic uterus, which was $3.1 \times 5.2 \times 2.50$ cm in diameter, and pelvic magnetic resonance (MR) imaging demonstrated that uterus size was smaller than normal.

We performed laparoscopic bilateral gonadectomy. During the operation, we observed bilateral, elongated, 'streak' gonads and a

small uterus. Pathology revealed atrophic ovarian tissue with diffuse cortical fibrosis (Figure 1).

Her sister had been diagnosed as having pure gonadal dysgenesis at the age of 15. At the time of presentation to our clinic, she was a 26-year-old woman with primary amenorrhoea. Her laboratory studies showed FSH level of 87 mIU/l, LH of 17.7 mIU/ml and E_2 level of 20 pg/ml. The development of secondary sexual characteristics was halted in breasts with Tanner 2 and pubic hair growth with Tanner 3 staging. She was given cyclic hormonal therapy but no menstrual withdrawal bleeding was seen. On her genetic analysis, her karyotype was found as 46,XY but no mutation was observed in the SRY gene. Laparoscopic bilateral gonadectomy was performed 9 years ago. A hypoplastic uterus was reported to be seen during the operation. Her pathology was reported as atrophic ovarian tissue with diffuse fibrosis (streak gonad).

Discussion

Pure gonadal dysgenesis is thought to be the result of a deletion in the short arm of Y chromosome involving SRY gene or a mutation in other genes that leads to inhibition of SRY function (Behzadian et al. 1991). XY gonadal dysgenesis can be inherited as autosomal or X-linked disorder. Familial cases of Swyer syndrome have also been described (Sarafoglou and Ostrer 2000; Bagci et al. 2011).

An interesting finding in our cases was that one sister responded to treatment with hormone therapy with menstrual bleeds but that the other did not. The hypoplastic size of uterus in Swyer syndrome can be explained by inherent factors, by deficiencies in the management of women with delayed puberty in terms of timing and method of induction of puberty (Michala et al. 2008) or by the insufficient function of prenatal anti-Müllerian hormone due to testicular failure (Kahyaoglu et al. 2006).

Hormonal replacement therapy has to be given to promote the development of secondary sexual characteristics and, probably, stimulate the growth of the uterus for an eventual pregnancy (Hétu et al. 2010). Pregnancies in women diagnosed with Swyer syndrome have been reported in the literature (Tulic et al. 2011; Creatsas et al. 2011) and this has shown that normal pregnancies can be achieved with the help of artificial reproductive techniques, despite the undeveloped structure of uterus.

Adoption and surrogacy is another option in having children with these patients. Furthermore in future, possible uterine transplantation may become an option in these women for whom adoption or gestational surrogacy is restricted for personal, societal or religious reasons.

Declaration of interest: The authors report no conflicts of interest. The authors alone are responsible for the content and writing of the paper.

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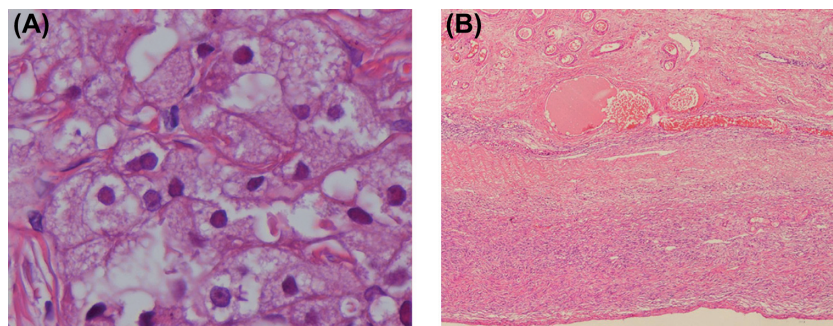


Figure 1. (A) The clumps of Leydig cells (H&E, $\times 600$). (B) A narrow zone of ovarian stroma (H&E, $\times 40$).

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