

CASE REPORT

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Swyer syndrome: a rare cause of primary amenorrhea

Ujjwala Sandilya¹ and Sangam Jha^{1*}

Abstract

Purpose Swyer syndrome is one of the rare causes for disorders of sexual development.

Case report An 18-year-old girl presented with complaints of inability to attain menarche. Physical examination revealed underdeveloped breast with absent pubic and axillary hair; however, the vagina was well canalized. Her hormonal profile was of hypergonadotropic hypogonadism. USG and MRI demonstrated hypoplastic uterus with very small ovaries. Chromosomal analysis reported 46 XY genetic makeup. Given the higher risk of malignancy in dysgenetic gonads, the patient underwent laparoscopic gonadectomy and had been put on hormone replacement therapy.

Conclusion Swyer syndrome, though a very rare entity, should be kept in mind while evaluating primary amenorrhea. Gonadectomy should be performed early once diagnosis is made to avoid the risk of malignancy.

Keywords Primary amenorrhea, Dysgenetic gonad, Swyer syndrome, Disorder of sexual development

Introduction

Swyer syndrome, also known as complete gonadal dysgenesis, is one of the rare causes of primary amenorrhea, with a reported incidence of 1:80,000 [3]. It is characterized by 46 XY genetic makeup with phenotypic female pattern and normal Mullerian derivative structure [6]. This condition is usually diagnosed at the adolescent age for the absence of menses and delayed puberty. Herein, we report a case of primary amenorrhea in an adolescent phenotypic female with a dysgenetic gonad.

Case report

An 18-year-old girl presented with the complaints of inability to attain menarche. There was no other significant history in her past. Her elder sister has attained menarche at an appropriate age. Her birth and development were normal, and her scholastic performance was

good. She was 155 cm (10th percentile) tall and weighed 47.8 kg (25th percentile) with an arm span of 145.6 cm. On examination, the breast was Tanner stage II, axillary hair and pubic hair were absent, and vagina length was around 8 cm.

Laboratory investigation

A hormonal analysis was performed which showed high FSH of 96.73 mIU/ml (reference range: 3–10 mIU/ml in pre-follicular phase) and LH-26.84 mIU/ml (reference range: 2–8 mIU/ml in pre-follicular phase) with low estradiol < 20 µmol/dl (reference range: 30–400 µmol/dl). Her testosterone was 32 ng/dl (normal range for the female age). The laboratory reports were suggestive of hypergonadotropic hypogonadism, confirming gonadal dysgenesis. Ultrasound revealed infantile uterus of size 1.2 × 3.8 × 2.8 cm with non-visualization of the ovaries. Both the kidneys were normal. MRI findings confirmed the ultrasound findings. Endometrium was visualized as thin T2 hyperintense structure measuring 2.3 mm in thickness. Genetic analysis showed a karyotype of 46 XY.

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Management

On the basis of history, clinico-laboratory findings of sexual infantilism, hypergonadotropic hypogonadism, presence of Mullerian structure and absence of gonads, and chromosomal constitution of 46 XY, diagnosis of Swyer syndrome was made. Careful disclosure of the condition was done. Parents were counseled regarding the risk of malignancy and infertility. Given the higher risk of malignancy in dysgenetic gonads, the patient underwent laparoscopic excision of the gonads. Laparoscopy revealed small uterus, normal fallopian tubes, and fibrous bands in the place of the ovary (Fig. 1A, B). The inguinal canal was empty on both sides. Histopathology returned as fibro-cartilaginous tissue focal ovarian stroma. The patient was started on estrogen first followed by cyclical estrogen and progesterone. The patient started menstruating at 6-month follow-up.

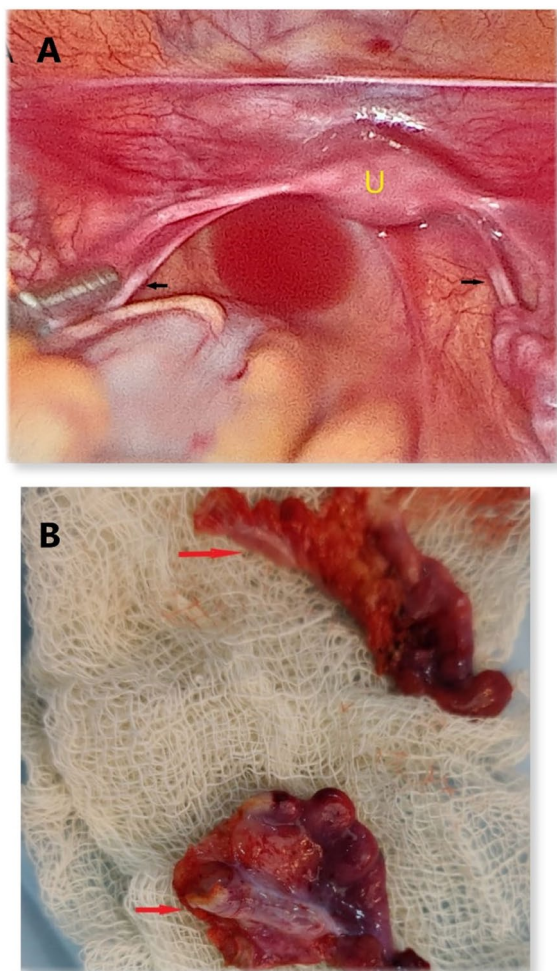


Fig. 1 a Laparoscopic view of the uterus (U) and streak gonads (black arrow). b Specimen of the removed streak gonads (red arrow)

Discussion

Swyer syndrome was first described by an endocrinologist Gerald Swyer in two women with primary amenorrhea, female external genitalia, normal Mullerian derivative structures, and 46 XY karyotype in 1955 [5]. Genetic sex depends on a series of complex events determined by genes and hormones that first lead to the gonadal definition and then to the differentiation of internal genital organs and at the end differentiation of external genital organs. The most important gene to determine the gonadal sex of the fetus is TDF (testis determining factor) in the SRY region (sex determining region) on the Y chromosome, and deletion of this region or mutations of various genes located in this region is thought to be the cause of 46 XY dysgenetic gonad [4]. As there is absence of testosterone and anti-Mullerian hormone, Mullerian ducts develop into the uterus, fallopian tube, and vagina. So, the patient with Swyer syndrome presents with hypergonadotropic hypogonadism with minimal secondary sexual characteristics. This is the disorder of sexual development (DSD) and needs to be differentiated from its close associates like androgen insensitivity syndrome (46 XY with absent Mullerian structures) and rare cases of Turner's syndrome (46 XY and streak gonads) when mosaicism with a normal or structurally abnormal Y chromosome is identified. Usually, Turner's syndrome is associated with the absence of complete or parts of a second sex chromosome. In 5–12% of patients, mosaicism with a Y chromosome is identified. Patients with 46 XY pure gonadal dysgenesis are at a higher risk of developing gonadoblastoma and dysgerminoma at younger age [5, 1]. So, prophylactic gonadectomy should be performed at the time of diagnosis [2], as done in the present case.

Conclusion

DSD (disorders of sexual development) is an extremely rare entity but should be kept in mind while evaluating a case of primary amenorrhea. Early diagnosis is of utmost importance for the initiation of hormonal replacement to promote pubertal growth and early gonadectomy.

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Authors' contributions

Conceptualization, data analysis, manuscript drafting, and revision had been done by SJ. Data collection and analysis were done by SJ and US. Both the authors have read and approved the final version of the manuscript.

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Availability of data and materials

Data will be available from the author on reasonable request.

Declarations

Ethics approval and consent to participate

Ethical approval has been waived off for the case report from the institute All India Institute of Medical Sciences.

Consent for publication

Informed consent for the publication has been obtained from the patient.

Competing interests

The authors declare that they have no competing interests.

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