

Complete 46, XY Gonadal Dysgenesis (Swyer Syndrome) in an Adolescent with Primary Amenorrhea

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ABSTRACT

Background

Swyer syndrome (46,XY pure gonadal dysgenesis) is a rare disorder of sex development, that presents with delayed puberty and primary amenorrhea in phenotypic females. It is characterized by a 46,XY karyotype and streak gonads due to the absence of functional gonadal tissue.

Case Presentation

We report a case of a 15 years old phenotypic female, born to consanguineous parents, who presented with primary amenorrhea, tall stature, and absent secondary sexual characteristics. Laboratory evaluation revealed hypergonadotropic hypogonadism, and imaging showed a small uterus with streak gonads. Karyotyping confirmed a 46,XY chromosomal constitution.

Management and Outcome: The patient underwent prophylactic laparoscopic gonadectomy to prevent malignancy and was successfully started on hormone replacement therapy to induce puberty.

Conclusion: This case highlights the importance of early diagnosis and multidisciplinary management to ensure optimal outcomes.

Keywords

46,XY gonadal dysgenesis; Fertility; Gonadal dysgenesis; Hormone replacement therapy; Primary amenorrhea; Swyer syndrome

INTRODUCTION

Swyer syndrome (46,XY pure gonadal dysgenesis) is one of the most uncommon disorders of sex development characterized by a phenotypic female with a 46,XY karyotype and streak gonads¹. It typically presents during adolescence with delayed puberty and primary amenorrhea^{1,2}. These individuals lack functional gonads, resulting in estrogen deficiency and lack of secondary sexual characteristics³. Due to the presence of Y-chromosome material, there is a high risk (20–30%) of gonadal tumors such as gonadoblastoma^{4,5}. Early identification and management are crucial to prevent malignancy and support normal pubertal development^{5,6}.

Case report

Our patient was a 15 years old adolescent of female phenotype who was brought to the gynaecology outpatient clinic by her family due to the absence of menarche and failure to develop breast tissue. She was psychologically upset. She is the third issue of a consanguineous marriage, where maternal age was 39. There was no significant medical, surgical, or family history. Height (166 cm), weight (72 Kg) and BMI 26.12 kg/m². Physical examination revealed tall stature with no palpable swelling in labia majora, Tanner stage I breast development, and sparse pubic/axillary hair. No dysmorphic features or systemic abnormalities were noted; vagina was capacious.

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Diagnostic assessment included laboratory investigations which showed elevated FSH (110 mIU/mL) and LH (52 mIU/mL) with low estradiol (<20 pg/mL), testosterone (0.18 ng/mL) and prolactin (7.11 ng/mL) consistent with hypergonadotropic hypogonadism. Anti-Müllerian hormone (AMH) was undetectable. Pelvic ultrasound showed a hypoplastic uterus and absence of ovaries, with bilateral streak gonads noted, findings which were subsequently confirmed by MRI (FIG 1). Karyotype analysis demonstrated 46,XY, confirming the diagnosis of Swyer syndrome (FIG 2).

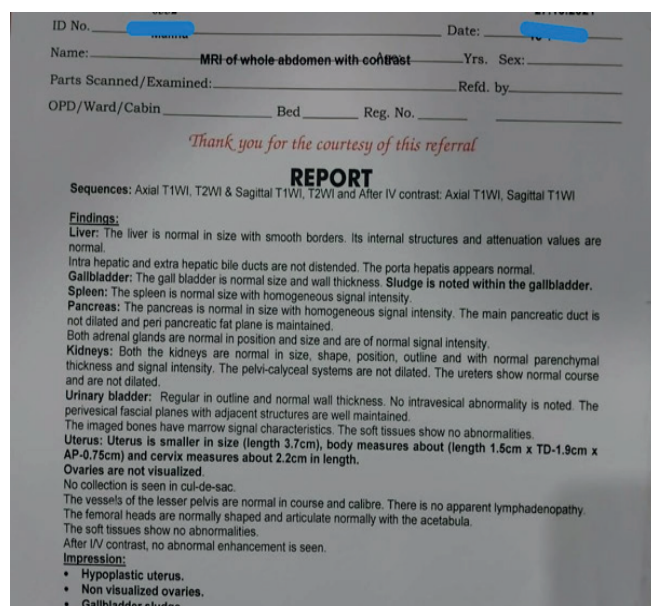


FIGURE 1: MRI report of the patient.

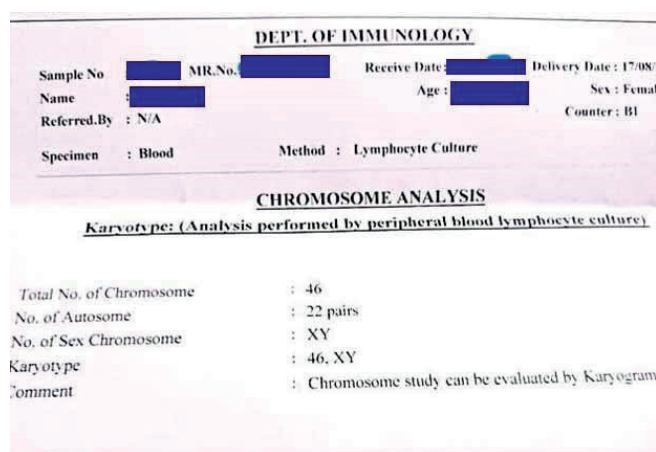


Figure 2: Karyotype analysis showing 46,XY chromosomal constitution

DISCUSSION

This case demonstrates a classic presentation of Swyer syndrome. The evaluation of any adolescent with primary amenorrhea should include hormonal testing and pelvic imaging ^{2,6}. Hypergonadotropic hypogonadism in a phenotypic female requires karyotype analysis to distinguish between ovarian insufficiency and disorders of sex development ^{2,3}.

Differential diagnosis included androgen insensitivity syndrome, which was ruled out by the presence of a uterus and absent breast development. Turner mosaicism was considered unlikely due to the absence of Turner stigmata and the confirmation of a 46,XY karyotype. Enzymatic defects, such as 17 α -hydroxylase deficiency, were excluded based on the patient's normal blood pressure and electrolyte profile^{6,7}.

Prophylactic gonadectomy was done to reduce high tumor risk in histopathology no gonadal (ovarian or testicular) tissue was seen and lifelong HRT was started to develop and maintain secondary sexual characteristics, menstrual cycles, bone health, and cardiovascular protection ^{4,5,8}. Psychological support and fertility counseling was given to patient and family for long term care ^{6,7}. Now patient is receiving regular HRT following withdrawal bleeding and patient is psychologically stable than before. Histopathological report of gonadectomy is added below. After receiving HRT, size of uterus increased. Ultra-sound of lower abdomen after receiving HRT is given below.

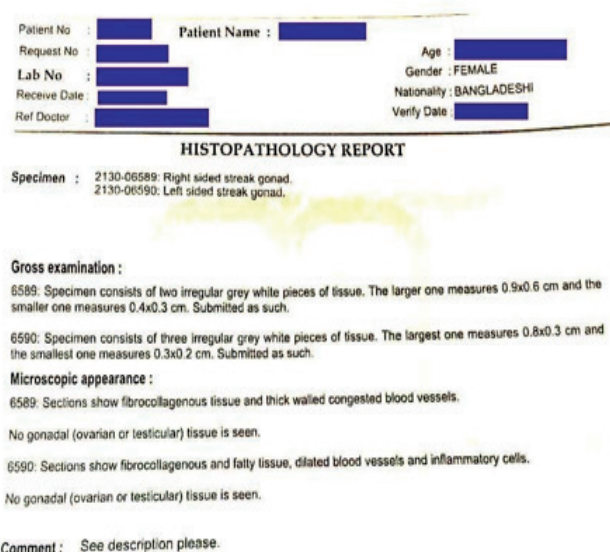


FIGURE 4: USG of lower abdomen.

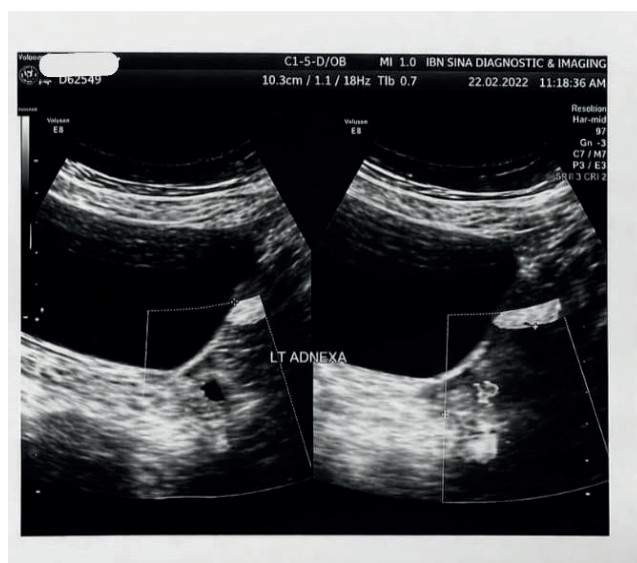
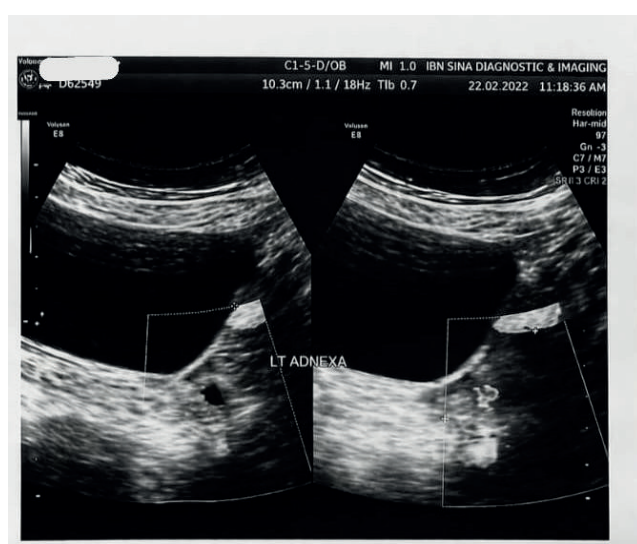
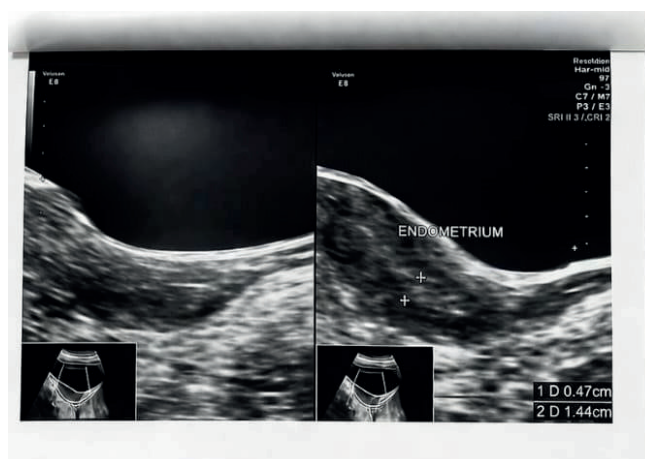


Figure 5: USG of lower abdomen

CONCLUSION

Swyer syndrome is a rare but important cause of primary amenorrhea. Early diagnosis through adequate hormonal, imaging, and genetic evaluation enables timely gonadectomy and appropriate hormone replacement therapy. With proper management, individuals with Swyer syndrome can achieve normal secondary sexual development and have reproductive potential through assisted reproductive techniques.

Patient Consent

Written informed consent was obtained from the patient and guardian for publication of this case report and accompanying details. All identifying information has been removed.

Ethical Clearance

As this is a single case report, formal ethical approval was not required according to institutional policy.

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