Swyer Syndrome with Gonadoblastoma: A Case Report

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Abstract

Background: Swyer syndrome is a condition of pure gonadal dysgenesis with 46, XY karyotype. These patients have female external phenotype and usually presented with primary amenorrhea. Detailed history, clinical, radiological and hormonal evaluation, karyotyping and histopathology are needed for diagnosis. Due to the presence of Y chromosomes these patients have streak gonads with increased risk of development of gonadoblastoma. Early diagnosis of Swyer syndrome followed by prophylactic gonadectomy is very important.

Case presentation: A 19-year-old female presented with the complaints of primary amenorrhea. The clinical, radiological, hormonal assessment and karyotyping test made the diagnosis of Swyer syndrome. The histopathological examination revealed left sided streak gonad and right sided gonadoblastoma.

Conclusion: Swyer syndrome is a rare form of disorders of sexual development. As there is high chance of gonadal malignancy, these patients should be counseled for preventive bilateral salpingo-oophorectomy

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Keywords: Swyer syndrome, Gonadal dysgenesis, Gonadoblastoma.

Introduction

Swyer syndrome is a form of disorders of sexual development.[1] It is a condition of pure gonadal dysgenesis with 46, XY karyotype and results from abnormal sexual differentiation during intrauterine life.[1]The incidence rate is about 1:80,000.[1,2] Reporting these cases are very important to establish the value of early evaluation of cases of primary amenorrhea to exclude Swyer syndrome or any other chromosomal abnormalities associated with gonadal malignant transformation.Here we present a case of Swyer syndrome with gonadoblastoma including the clinical, radiological, hormonal, karyotyping and histopathological findings.

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Case Presentation

Clinical history: In May 2023 a 19-year-old female had presented with the chief complaints of primary amenorrhea at Sir Salimullah Medical College Hospital, Dhaka. Patient had no past history of any significant ailment. Nosignificant family history of any inherited disease or congenital anomaly was present. On clinical examination, the height of the patient was 157.48 cm. She was 47.57 kg in weight. Her pulse rate was 68/min, blood pressure was 110/70 mm Hg. She had underdeveloped breasts, very sparse axillary and pubic hair. She had normal female external genitalia with well develop labia majora and clitoris. The per vaginal examination revealed the presence of red dry vaginal introitus with a 3 cm long blind tract. The labia majora is well differentiated and minora is small.

Radiological findings: The pelvic ultrasound revealed a 3x0.4 cm small rudimentary uterus with non-visualized ovaries. The pelvic MRI examination confirmed the presence of a 1x0.3 cm rudimentary uterus with 1 mm cervix and slit-like endometrial cavity. Both ovaries were non-visualized. Karyotyping was done at Department of Pathology, Bangabandhu Sheikh Mujib Medical University. It revealed genotype 46, XY (Figure 1).

Laboratory findings: The laboratory investigations showed low levels of testosterone and high follicular stimulating hormone and luteinizing hormone.

Histopathological Findings: Laparoscopic exploration followed by excision of both sided adnexal tissues was done.

Gross examination: Her left sided adnexal tissue showed 2 cm long fallopian tube with 0.3 cm diameter and a 0.6 X 0.5 cm gonad. The right adnexal tissue showed 2 cm long fallopian tube with 0.3 cm diameter and a 0.5 X 0.5 cm gonad.

Histopathological examination: The both tissues revealed right sided gonadoblastoma[Figure 2(A, B)] and left sided streak gonad (Figure 3).

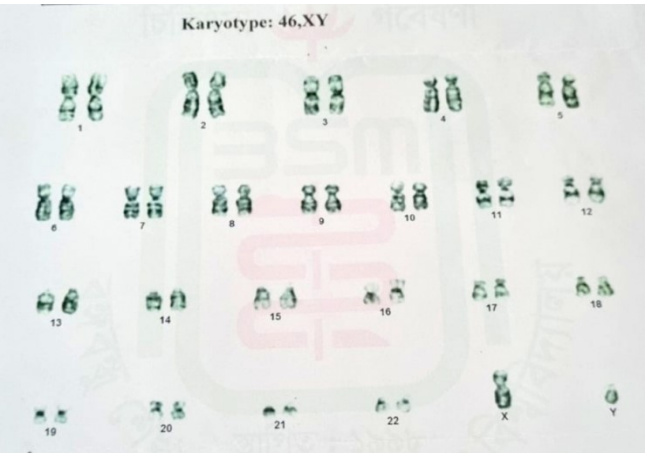

Figure 1: Karyotyping of the patient shows 46, XY.

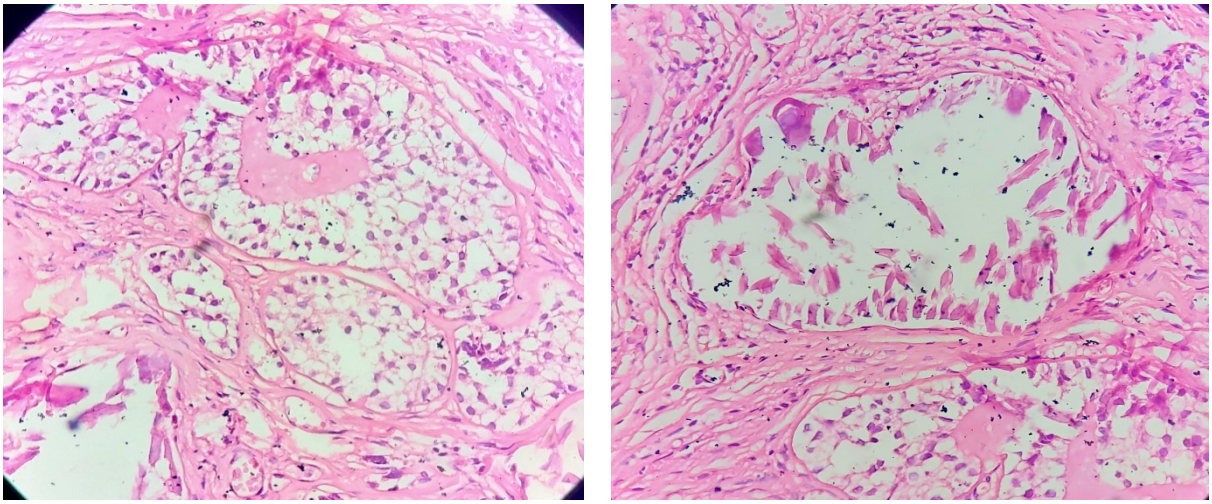

Figure 2 A & B (H&E x400 magnification): Histopathology images of right gonad: Gonadoblastoma (Nests of primitive germ cells and sex cord stromal cells surrounded by ovarian stroma and foci of calcification).

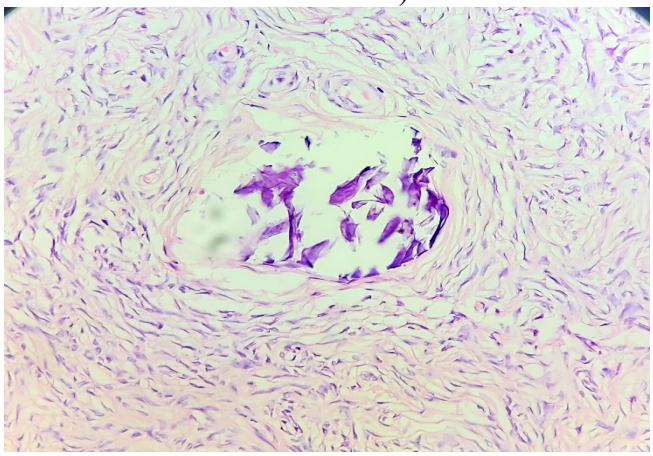

Figure 3 (H&E x400 magnification): Histopathology image of left gonad: Streak gonad (Fibrosis with characteristic calcification).

Discussion

Swyer syndrome is a unique condition of pure gonadal dysgenesis with female phenotype and 46, XY karyotype.Presence of SRY gene in Y chromosome and "Testis Determining Factor (TDF)" are necessary for early stages of intrauterine testicular development. Mutation of the genes present in the Y chromosome, particularly mutation of the SRY gene, is a major cause of Swyer syndrome.[3]Due to the presence of these mutations, testicular development does not occur which leads to absence of testosterone and anti-Mullerian hormone. As a result, the Mullerian duct is persisted and the patient may have rudimentary uterus, fallopian tube and vagina.[4]These patients are phenotypically female with female external genitalia withgonadal dysgenesis (small or rudimentary uterus, normal to small sized fallopian tube and upper part of vagina).[3] As these patients have streak gonads, they are presented with primary amenorrhea. Presence of Y chromosome leads to increased risk of development of gonadoblastoma. Congenital Androgen Insensitivity Syndrome is one of the main differential diagnoses of Swyer syndrome. In Congenital Androgen

Insensitivity Syndrome, the patients have 46, XY karyotype with high testosterone, normal but undescended testis, absent structure and normal female phenotype. On the other hand, Testosterone level is low in Swyer syndrome.[1,3] True hermaphrodite is another differential diagnosis of Swyer syndrome. They can present with female phenotype but may have ovotestes.[1,5] In Swyer syndrome, the presence of Y chromosomes may increase the risk of germ cell neoplasm at an early age. Gonadoblastoma and dysgerminoma are more frequent. Thus, prophylactic bilateral gonadectomy is the definitive treatment to reduce risk of gonadal malignancy.[5]Gonadoblastoma is a mixed germ cell tumor of undifferentiated cells. It is a precursor of malignant germ cell tumors. It is associated with 50-60% cases of dysgerminoma and 10% cases of other malignant germ cell tumors like yolk sac tumor, embryonal carcinoma, choriocarcinoma etc.[6] It can be presented with no visible tumorous lesion grossly. The biopsy samples of our patients also did not show any visible tumorous lesion.[7,8] Although the patients with Turner syndrome are usually presented with short stature and primary amenorrhea, our patient did not have any clinical stigmata of Turner syndrome.[1,3.4]

Conclusion

In this case report, Swyer syndrome the rare form of pure gonadal dysgenesis with its complication (gonadoblastoma) is evaluated and highlighting the role of proper clinical evaluation, karyotyping, imaging (USG and MRI) and hormonal assessment (serum testosterone) for accurate diagnosis. Importance of early prophylactic bilateral gonadectomy to prevent gonadal malignancy is also stated in this case report.

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