Case Report

Swyer Syndrome with Heterotopic Adrenal Cortical Tissue in Streak Gonads: A Rare Case and Review of Literature

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Abstract

Swyer syndrome is a 46 XY pure gonadal dysgenesis. Affected individuals are phenotypically female with external female genitalia, hypoplastic Mullerian structures, and the presence of streak gonads. Gonadectomy is indicated due to increased risk of development of malignancy in streak gonads. We hereby report a case of 15-year-old patient with Swyer syndrome who underwent laparoscopic gonadectomy. Histopathological examination showed streak gonads with the presence of accessory adrenal cortical tissue. The patient was put on hormonal replacement therapy postsurgery. Heterotopic adrenal rest may be an incidental finding in normal ovary and testis. Its presence in streak gonads has never been reported, and we propose a theory of increased risk of gonadal and adrenal tumors, possibly because of mutations in disorders of sex differentiation.

Keywords: *Adrenal tissue, streak gonads, Swyer syndrome*

Introduction

Disorders of sexual development are defined as congenital conditions, in which development of chromosomal, gonadal, or anatomical sex is atypical. They are caused by structural or numerical anomalies of the sex chromosomes or mutations in one of the genes involved in the formation of the urogenital ridge and sex determination of the bipotential gonad.

Swyer syndrome is pure XY gonadal dysgenesis. External genitalia is that of female with the patent vagina. Hypoplastic Mullerian structures are present with streak gonads and chromosomal constitution XY. It needs to be differentiated from androgen insensitivity syndrome.

The streak gonads in Swyer syndrome have potential to undergo malignancy, most commonly, gonadoblastoma is found with associated dysgerminoma. Thus, it is necessary to remove gonads as soon as the diagnosis is made. We report a case of Swyer syndrome with adrenal ectopic tissue rest in streak gonads.

Case Report

A patient, aged 15 years old presented to the outpatient department at a tertiary center with primary amenorrhea. The patient had ill-developed breasts and scanty axillary and pubic hair. She had a normal intelligence.

The patient was 150 cm tall and weighed 46 kg. Blood pressure was within normal range. Breasts were tanner Stage II, pubic hair was tanner Stage I. There was no evidence of webbed neck. There was no galactorrhea or thyroid enlargement. On local examination, labia and urethral orifice were normal; vagina was patent and approximately 5–6 cm in length. The patient underwent a battery of investigations which are as follows: serum testosterone 0.20 ng/mL (normal levels for a female), luteinizing hormone 20.64 mIU/mL, and follicle-stimulating hormone 112.04 mIU/ml. Prolactin and thyroid function tests were normal. Blood sugars, complete blood count, liver function test, and kidney function test were normal. Karyotype revealed 46 XY chromosome setup. Ultrasound and magnetic resonance imaging showed hypointense streak like structure, suggesting Mullerian remnant. Bilateral ovaries, cervix, and upper part of the vagina were not visualized, suggesting streak gonads and bilateral kidneys normal.

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The diagnosis of Swyer syndrome was made, and laparoscopic bilateral gonadectomy was done followed by hormone replacement therapy for breast development.

Intraoperatively, small-sized uterus and bilateral tubes were seen. Streak gonads were beneath the fallopian tubes attached to infundibulopelvic ligament laterally. No gonadal tissue identified in inguinal canal bilaterally [Figure 1]. Histopathology revealed fibrofatty tissue with streaks of fibrous tissue resembling ovary with few tubules lined by columnar to cuboidal cells with small nucleoli and Mullerian epithelial cysts. Occasional foci of heterotopic adrenal cortical rest tissue were seen as encapsulated nodule comprised groups and nests of large round cells separated by thin vascular channels. Cells also had abundant eosinophilic granular cytoplasm. All of this suggested the presence of heterotopic adrenal rest in streak gonadal tissue [Figure 2].

Postoperative period was uneventful. At the time of discharge, the patient was started on conjugated equine estrogen.

Discussion

Swyer syndrome was first described by Jim Swyer in 1955. It is 46 XY complete gonadal dysgenesis and has been estimated to occur in approximately 1/100,000 people.[1] There is a defect in embryogenesis, that is, indifferent gonads fail to differentiate into testes in a XY, genetically male fetus. It is usually caused by a mutation in the SRY gene due to deletion in the DNA-binding region of the SRY gene, in 10%–20% cases. It is well known that heterozygous small deletion in 9p, containing DMRT 1 can cause complete or partial gonadal dysgenesis.[1]

The gonads of XY pure gonadal dysgenesis are at increased risk of developing gonadoblastoma and germ cell tumor, particularly dysgerminoma; so gonadectomy is advised in cases of XY gonadal dysgenesis.[2]

The occurrence of adrenal rest at paragonadal location is a normal variation. Accessory adrenal tissue (AAT) is a common observation seen in children underwent groin surgeries, with an incidence varying from 1.63% to 3%, but it is a rare finding in adults around 1%.[3,4] During embryological development, in the adrenal gland, medulla invades the cortex, occupying the central position. During this process, accessories form as the cortical tissue splits off and maybe dragged along with the descending gonads toward the inguinal region, the most common site is retroperitoneum and hilum of gonads. Lower occurrence in adults could be explained by either involution of ectopic rests, less complete dissection, under-reporting, or due to difficulty in identification in the adult groin majority of the cases of AAT reported are in males.[4]

In histopathology, AAT shows encapsulated nodule comprised groups and nests of large round cells separated by thin vascular channels. Cells have abundant eosinophilic granular cytoplasm with periphery having smaller darkly staining cells[4] as in our case.

Ovarian adrenal rest tissue (OART) has been reported in the literature rarely as compared to testicular adrenal rest tissue. The first case of masculinovoblastoma (tumor of ovarian adrenal rest) was reported in the literature in 1957.[5] Although OART is large benign tumors, usually the patient undergoes oophorectomy, as there are no data available about the natural course of these tumors. However, in our patient, heterotopic adrenal rest was an incidental finding in streak gonads which have been never reported. Genetic mutation can attribute to the simultaneous presence of disorders of sexual development and presence of adrenal rest tissue in streak gonads.

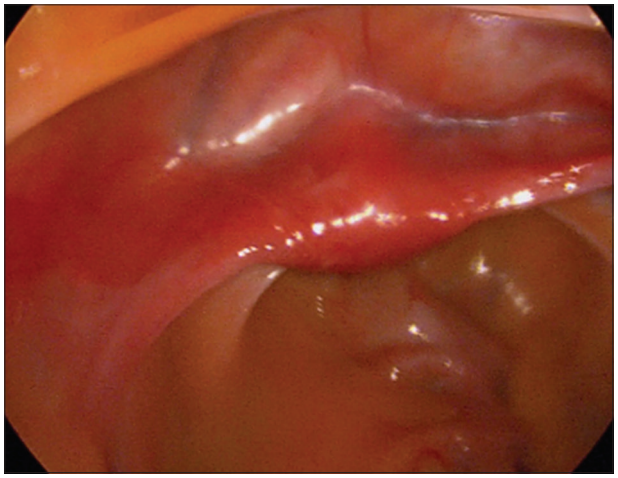

Figure 1: Rudimentary uterus with bilateral tubes; normal gonads not seen

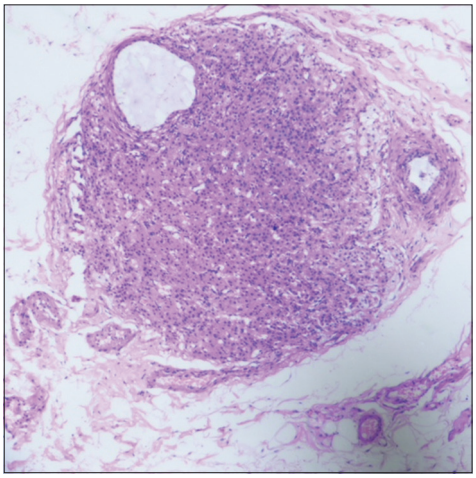

Figure 2: Ectopic adrenal cortical rest in streak gonads, H and E, ×4

Ectopic accessory adrenal tissue rest is sometimes present in gonads of patients with non-classical CAH which may grow into tumor by raised adrenocorticotropic hormone. It may mimic gonadoblastoma in these patients. Hence, nonclassical CAH needs to be differentiated from Swyer syndrome in the presence of AAT.

Conclusion

The presence of AAT in a patient with Swyer syndrome does not necessarily mean a casual occurrence. This case is reported to emphasize the occurrence of ectopic adrenal tissue in dysgenetic gonads, which may propose a theoretical risk of abnormal development of the genitourinary system and the risk of adrenal rest tumors along with gonadoblastomas. In Swyer syndrome, this ectopic tissue may be found in the streak gonads because of some genetic mutations and removal of gonads at early age in such patients should be done to prevent the development of tumors.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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