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Case Report

Gonadoblastoma in a 15-Year-Old Female With 46, XY Swyer Syndrome

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Abstract

Swyer syndrome or 46, XY pure gonadal dysgenesis is a very uncommon sex development disorder. This sex development disorder gives the appearance of a female phenotype with non-functional streak gonads and normal development of the Müllerian system but with an XY chromosome configuration. Due to the presence of Y chromosomal components, there is a significantly increased risk of gonadoblastoma and dysgerminoma in Swyer syndrome patients. In the present case, a 15-year-old phenotypically female patient diagnosed with Swyer syndrome underwent a prophylactic bilateral gonadectomy. There were no masses identified by the imaging study and the tumor markers were normal prior to the procedure, but the pathology evaluation confirmed the presence of a gonadoblastoma in situ in the right streak gonad upon microscopic analysis because the tumor would otherwise have been malignant. The patient is currently following a regimen of hormone replacement therapy and tumor surveillance.

Introduction

Swyer Syndrome or 46, XY pure gonadal dysgenesis, is characterized by an individual with an XY chromosome pattern but a female phenotype, accompanied by non-functional "streak" gonads consisting of fibroblast tissue (1-4). The underlying pathology is a dysfunctional testicular development in the guise of sex-determining gene mutations, causing a non-expression of anti-Müllerian hormone and testicular testosterone (5). The result is a normally developed internal Müllerian duct derivative consisting of the uterus and the Fallopian tubes. Typically, it is the teenager who becomes aware of their condition due to issues in puberty attainment and primary amenorrhea (2). One of the most important clinical aspects of Swyer syndrome is the significant risk of gonadal malignancies, including particularly gonadoblastoma and dysgerminoma, with a risk of malignancy of 15%-35% (1-3;6;7). The presence of Y chromosomal elements, and specifically the TSPY gene, is considered a strong oncogenic factor (8). It is worth noting that there is a general agreement to proceed with bilateral gonadectomy as soon as the diagnosis is made to circumvent this risk (6). The following is a case report of a teenager with Swyer syndrome. She had undergone prophylactic gonadectomy, in which a microscopic gonadoblastoma was incidentally identified.

Case Report

Presentation

This was a 15 years and 9-month-old genetically male person raised as a female with 46,XY pure gonadal dysgenesis. she had been scheduled for the prophylactic removal of both gonads. she had been raised as a female. She had been on puberty induction therapy with the combination of estrogen progesterone. she had been having withdrawal bleeding, which was normal. She had been having an unremarkable medical history. On physical examination, she had Tanner stage III breast development without any abnormalities of female external genitalia, with no palpable abdominal or pelvic masses. She did not complain of abdominal pain, weight loss, or any systemic symptoms. These features in her presentation were typical for the adolescent-onset characteristics of Swyer syndrome as described in literature. The issue of gonadoblastoma was brought out in surgical counseling. Therefore, removal of the dysgenetic gonads was recommended in consonance with medical treatment protocols.

Investigations

Karyotypic analysis revealed a 46,XY chromosome pattern. Such a fact is consistent with Swyer syndrome. Pelvic ultrasound studies demonstrated an anteverted uterus with regular form and homogenous endometrium. The ovaries were not visualized, consistent with the finding of streak gonads, and there was no adrenal enlargement or pelvic fluid collections, common in patients with Swyer syndrome (4;9). In the preoperative labs, the tumor markers were found to be in the normal range: these included beta hCG and alpha fetoprotein (AFP). Also, despite the tumor markers being in the normal range, gonadoblastoma cannot always be ruled out because sometimes the tumor is non-secretory during the early stages of this type of tumor. During laparoscopy, bilateral streak gonads were noted with elongated fallopian tubes and adhesions in the right lower pelvis with a small uterus and no masses & lesions noted.

Treatment and Further Investigations (Including Microscopic Findings)

The procedure involved a laparoscopic bilateral gonadectomy and adhesiolysis. The surgery proceeded well, and the patient lost very little blood. The gonads were completely removed for histopathological analysis.

Microscopic Pathology:

Right Gonad

On microscopic examination, the right gonad consisted of a mixed germ cell and sex cord stromal tumor typical for gonadoblastoma. There were classic nests and cords of the germ cells intermingled with the sex

cord stromal parts against the background of the basement membrane-like hyaline stroma. The germ cells showed well-rounded nuclei with prominent nucleoli and clear cytoplasm typical for dysgerminoma, whereas the sex cord stromal cells had bland nuclei that formed a peripheral palisading pattern. There were concentric calcifications typical of the tumour. Immunostaining supported the diagnosis: Germ cells were positive for D2-40, CD117, OCT3/4 (focal), and CKAE1/AE3 (focal). Sex-cord cells were focally positive for calretinin and inhibin. This biphasic immunophenotypic profile is characteristic of gonadoblastoma in dysgenetic gonads (10;11).

Left gonad

The sections showed the presence of streak gonadal tissue along with tubal tissue, along with mild vascular congestion. There was no malignancy. A tertiary pathology lab confirmed the presence of gonadoblastoma in situ (pTis). The patient has begun regular tumor marker evaluations and will continue the long-term regimen of hormone replacement therapy to promote puberty development, bone growth, and uterine development and maturation.

Outcome and Follow-Up

The postoperative course was uneventful. The patient had follow-up with no complaints of pain or difficulty or abnormal finding concerning tumor markers or recurrence on imaging. She is presently maintained on estrogen-progesterone therapy and vitamin D supplements with normal pubertal development. These findings are consistent with the literature concerning a greatly diminished risk for cancer with early gonadectomy (2;6;7).

Discussion

This case illustrates several important features of Swyer syndrome: the patient's phenotype is female, the gonads are streaks, the Müllerian ducts are intact, and the chromosomal pattern is 46,XY. The important clinical point about Swyer syndrome is the increased susceptibility to gonadal tumors, particularly gonadoblastoma, which can develop in as many as a third of people with dysgenetic gonads with a Y chromosome. The histopathological findings are characteristic of gonadoblastoma and include nests of germ cells and sex cord stromal cells, a hyaline basement membrane-like substance, and calcifications. Immunohistochemical studies also help establish the diagnosis since the germ cells are immunopositive for OCT3/4, CD117, and D2-40 proteins consistent with the expected profile. That is particularly important in the following case, where the crucial clinical message that has been demonstrated here is that the presence of gonadoblastoma can occur even if the imaging studies and tumor markers are shown to be normal, so just

using the ultrasound findings isn't sufficient. After the diagnosis of Swyer syndrome has been made, prophylactic gonadectomy clearly becomes mandatory. Early removal of the streak gonads not only prevents the risk of malignant transformation but also facilitates the initiation and maintenance of hormone therapy based on the progression of the patient's puberty. The fact that the left gonad appears non-malignant is an aspect that relates to the established pattern of asymmetric growth, in addition to reported unilateral cases of gonadoblastoma. The importance of follow-up with markers, imaging studies, and combined care is not any less critical in management.

Learning Points

Things to remember that are important about Swyer Syndrome:

- Swyer's Syndrome should be included in the differential diagnoses in cases of young girls with delayed puberty and primary amenorrhea, especially when there is hypergonadotropic hypogonadism with a karyotype of 46,XY.
- Dysgenetic gonads with Y chromosome tissue are at high risk of gonadoblastoma and should be excised even if the appearance on imaging studies is normal.
- It may remain microscopic and asymptomatic; hence, careful examination of the histopathologic specimen is necessary.
- The role of hormone replacement therapy in regards to the promotion of pubertal growth, the maintenance of bone density, and the promotion of the reproductive organs' health.

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