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Ovarian dysgerminoma with contralateral gonadoblastoma in a 46XY phenotypic female: Swyer syndrome

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Özet

46XY fenotipli kız çocukta over disgerminomu ile birlikte gonadoblastom olgusu: Swyer sendromu

Onbeş yaşında, bir yıldır primer amenore ve sağ inguinal şişlik yakınması olan olgunun fizik baskısında; kız fenotipi, bir miktar kıllanma ile birlikte klitoris hipertrofisi mevcuttu ancak göğüsleri gelişmemişti. Ultrason ile yapılan incelemede sağ over kaynaklı kitle saptandı. Ameliyat esnasında sağ salpingoooferektominin yanısıra şüpheli görünüme sahip sol taraftan da biyopsi yapıldı. Histolojik inceleme sonucunda sağ overde disgerminom, solda ise gonadoblastom saptanması üzerine ilk ameliyattan bir ay sonra sol gonadektomi ve kliteroplasti yapıldı. Olgumuz, kız fenotipine sahip sağ overde disgerminom, solda ise gonadoblastomu olan ilginç bir Swyer sendromu olma özelliğini taşımaktadır.

Anahtar kelimeler: *Gonadal disgenezi (46XY), gonadoblastom, disgerminom*

Summary

A 15 year-old-girl was admitted to the pediatric surgery department with primary amenorrhea and right inguinal swelling of one year duration. On physical examination, the patient presented with a female phenotype, but no breast development; somewhat hirsute with hypertrophic clitoris. Ultrasound examination disclosed a palpable mass originating from the right ovary. At operation, a right salpingoooferectomy was performed, and a biopsy specimen was obtained from the left ovary which did not appear normal. Histopathologic examination revealed a right ovarian disgerminoma and a left gonadoblastoma. One month after the first operation, a left gonadectomy was performed with accompanying cliteroplasty. An original case of Swyer syndrome is reported in a patient with female phenotype, dysgerminoma of the right and gonadoblastoma of the left ovaries.

Key words: *Gonadol dysgenesis(46XY), gonadoblastoma, dysgerminoma*

Introduction

Swyer's syndrome or 46XY gonadal dysgenesis is a rare condition characterised by female phenotype with primary amenorrhea, failure in breast development, hirsutism and clitoris hypertrophy with bilateral streak gonads, fallopian tubes and hypoplastic uterus [1]. Due to the presence of the H-Y antigen, these patients are at increased risk for malignancy, estimated at a rate of 25-50 percent [2].

We report a rare case with a right ovarian dysgerminoma and a left ovarian gonadoblastoma in a 46XY phenotypic female.

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Case Report

A 15-year-old girl presented with a one year history of right inguinal swelling. She had primary amenorrhea. Physical examination revealed a somewhat hirsute female with no breast development, a suprapubic mass and clitoral hypertrophy. Serum hormone profile was: AFP 3 u/ml (normal:<4.2), HCG 10.4 IU/ml (normal:<3.0), FSH 6.8 IU/l (normal: 3-11). Radioimmunoassay kit (manifactured by CIS) was used to measure FSH; LH 105 IU/l (normal: 1-11); testesteron 9.2 ng/ml (normal:<0.6). Ultrasonography showed a solid mass originating from the right ovary. At operation, a right ovarian solid tumour, 10x10 cm in dimentions and a left dysgenetic gonad of 2x1 cm, and a hypoplastic uterus were found. A right salpingo-ooferectomy and a left

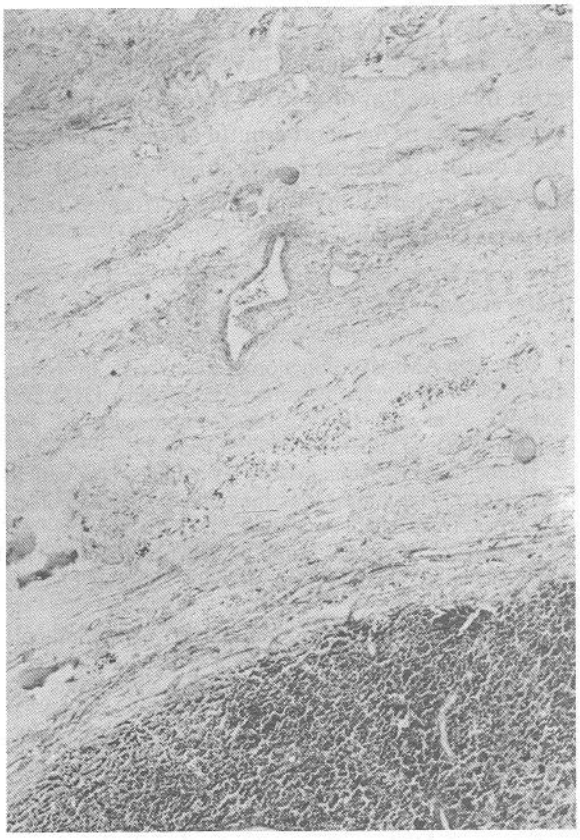

Figure 1. Disgerminoma of the right ovarium with fibrous capsule (HEx40).

gonadal biopsy were performed which proved histopathologically to be a right ovarian dysgerminoma and a left ovarian gonadoblastoma.

The righ ovarium showed typical features of dysgerminoma with fibrous stroma, focal invasion of fibrous capsule, lymphocyte infiltration with tumour cells lying in rows (Fig. 1). In the left ovarium, lamellar calcifications were seen to be adjoined by typical, oval or elongated nests of gonodoblastoma under the fibrous capsule (Fig. 2).

Gonadoblastoma cells were arranged around the typical eosinophilic, hyaline-like material yielding a pseudo follicular appearance. At the second operation a left ooferectomy and cliteroplasty was performed. Lymphocyte culture determined the karyotype as being 46XY. However, karyotypes of the family members were compatible with their phenotypes.

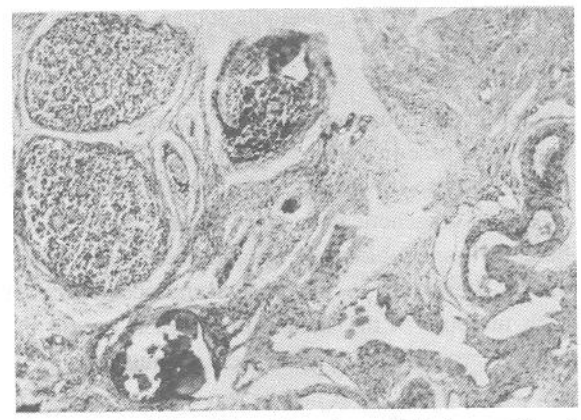

Figure 2. The left ovarium showed gonadoblastoma with adjacent dense lamellar calcification (HEx40).

Discussion

Swyer syndrome was diagnosed in a case presenting with female phenotype and streak gonads. The 46XY genotype designated the case to be a "Pure XY gonadal dysgenesis", probably inherited as an autosomal trait which is distinct from other entities with streak ovaries [1]. In addition, Y-specific probes have detected instances in which there were deleted segments of the short arm of the Y chromosome where the gonads consist of almost totally undifferentiated streaks [1,3].

Cases with Swyer syndrome exhibit sex reversal but uterus and fallopian tubes are present and grossly the gonads appear to be ovaries. Affected patients have normal stature and female phenotype. At puberty breasts fail to develop and menarche fail to occur and patients present with hypergonadotropic primary amenorrhea [1,3]. The gonads almost always consist of undifferentiated streaks. Gonadal streaks in gonodoblastomas have been shown to be capable of producing both androgens and oestrogens that seems to be the most likely source of β-HCG while increased androgens were probably produced by the gonodoblastoma.

In our present case, β-HCG was found 104 mIU/ml. The primitive gonad cannot accomplish any testicular function but is capable of producing some androgens leading to virilization at puberty, e.g., clitoris enlargement, hirsute appearance [9]. Intersex patients with karyotypes including a Y chromosome are known to be at high risk of developing po-

tentially malignant gonadoblastomas or dysgerminomas [4,7].

The streak gonads may undergo neoplastic changes as was the case in our present report. Dysgerminoma may also arise in women with normal ovaries but is typical of disgenetic gonads [2]. Although cases with bilateral gonodoblastoma/dysgerminoma are reported in literature [8], the concurrent occurrence of both tumours in the same patient, each one in the other ovary, as in the present case, is unusual.

Schulze et al. [6], reported a 10-year-old girl with acute abdominal symptoms and at explorative laparotomy, they observed dysgenetic gonads which later proved histopathologically to be gonadoblastoma. The karyogram of that patient performed after the operation revealed normal male 46XY. Their proposal was to perform a karyogram if dysgenetic gonads and rudimentary uterus were encountered during an operation.

It was reported that a family of four siblings in which two phenotypically female XY and one male developed germ cell tumours which demonstrates that brothers of affected sisters might also be at risk [4]. Gonadoblastomas and dysgerminomas should be removed soon after ascertainement regardless of age [5]. Thus, in addition to the initial right salpingo-ooferectomy a left ooferectomy was performed soon after the receipt of the pathology report.

In conclusion, we suggest that all patients with primary amenorrhea should have a complete karyogram for detection of a 46XY chromosome or deleted segments of the short arm. In addition, if the gonads consist of almost totally undifferentiated streaks early gonadectomy should be performed before the arise of gonadal tumours.

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