GONADOBLASTOMA: A CLINICOPATHOLOGIC STUDY

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Gonadoblastoma is a rare tumor found almost exclusively in patients with an underlying gonadal disorder, and accounts for two-thirds of gonadal tumors in women with abnormal gonadal development. Three cases of gonadoblastoma are reported here. One had Swyer syndrome (pure gonadal dysgenesis) with a 46-XY karyotype, the second patient had a mixed 46-XY and 46-XO karyotype, and the third patient had male pseudohermaphroditism and a 46-XY karyotype. Patients with pure gonadoblastoma have an excellent prognosis, when bilateral excision is performed. Gonadoblastoma may be overgrown by dysgerminoma, however, there is a good prognosis. Gonadoblastoma has never been detected with metastatic lesions. The prognosis for gonadoblastoma, including cases of Swyer syndrome, is good, provided early excision is performed bilaterally.

Keywords • dysgerminoma • gonadal dysgenesis • gonadoblastoma • Swyer syndrome

Introduction

Gonadoblastoma is an uncommon tumor. The neoplastic nature of gonadoblastoma has been questioned because some lesions are small and may undergo complete regression by hyalinization and calcification. In 1953, gonadoblastoma was described in detail by Scully as a gonadal tumor composed of germ cells and sex-cord derivatives resembling immature granulosa and Sertoli cells. Gonadoblastoma occurs almost entirely in patients with pure or mixed gonadal dysgenesis or in male pseudohermaphroditism. The association with dysgerminoma is seen in 50% of cases and with other, more malignant germ cell neoplasms, in an additional 10% of cases as reported in Iran by Kariminejad et al in 1972.

Case Report

Case 1

A 24-year-old patient with complete female phenotype presented with primary amenorrhea and infertility of 5 years’ duration.

Laparoscopic findings showed a normal uterus and fallopian tubes with small sized fibrotic ovaries. Karyotyping revealed a 46-XY pattern, which is compatible with the diagnosis of Swyer syndrome. The patient underwent bilateral...
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Figure 2. Benign mucinous cystadenoma with a typical endocervical – type epithelium was associated with gonadoblastoma (Hematoxylin and eosin X 100).

gonadectomy. Histopathology of the right gonad revealed discrete cells of sex-cord type along with laminated spheres of calcification. These are the characteristic features of gonadoblastoma (Figure 1). Within the left gonad, cortical gonadal tissue was seen.

Case 2
A 19-year-old patient with female phenotype presented with primary amenorrhea. Ultrasonography indicated uterine agenesis, and showed a 12 mm simple cyst on the right gonad. The left gonad was very small (approximately 8 mm in maximal diameter) on ultrasonography. Karyotyping showed 85% 46-XY and 15% 46-XO patterns. Sex chromatin was negative. Bilateral gonadectomy was performed. Gonadoblastoma in both gonads was documented. A simple mucinous cystadenoma was also detected in the right gonad (Figure 2).

Case 3
A 19-year-old patient with female phenotype presented with primary amenorrhea. A very small infantile uterus (3 x 2 cm) was detected on ultrasonography. Chromosomal analysis revealed a 46-XY pattern. After bilateral gonadectomy, both gonads showed gonadoblastoma overgrown by dysgerminoma (Figure 3).

Discussion
The most distinctive member of the group of tumors composed of a combination of germ cells and sex-cord stromal cells is gonadoblastoma.\(^1,4,5\) This tumor occurs in sexually abnormal individuals, most commonly affected by gonadal dysgenesis and carrying the Y chromosome\(^3, 6 – 9\) (i.e., \(X\)Y gonadal dysgenesis and \(XO\)-\(XY\) mosaicism).\(^10 – 13\) However, gonadoblastoma has also been documented in both phenotypically and chromosomally normal females, even those with successful pregnancies.\(^14\)

All patients in this study showed abnormal karyotyping. Gonadoblastomas are bilateral in over one-third of cases.\(^15\) In this study, two-thirds of the cases were bilateral. The nature of the gonad in which a gonadoblastoma originates is frequently not determined because there is often a mixed pattern of growth by the gonadoblastoma or a neoplastic germ cell element, most frequently a dysgerminoma.\(^5,16\) One case in this study had bilateral gonadoblastomas and was overgrown by dysgerminoma. Patients with a pure gonadoblastoma have an excellent prognosis provided both gonads are excised. The prognosis of patients with gonadoblastoma associated with dysgerminoma is also good.\(^5,16,17\) Gonadoblastoma has never been detected with metastatic lesions and has never been encountered outside the gonads.\(^5\) A unique combination of an ovarian gonadoblastoma, dysgerminoma, and mucinous cystadenoma in a patient with Turner’s syndrome was reported by van del Bijl et al.\(^2\) In our study, the association of a gonadoblastoma with a simple mucinous cystadenoma (tumor of epithelial origin) was observed with interest.

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