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THE USEFULNESS OF LAPAROSCOPIC REMOVAL OF DYSGENETIC GONADS: A CASE REPORT

GÔNADAS DISGENÉTICAS; GONADOBLASTOMA, LAPAROSCOPIA, GONADECTOMIA

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ABSTRACT

Females with XY karyotype and streak gonads are classified as presenting XY gonadal dysgenesis. These patients are at higher risk of developing malignancy in their gonads; in such cases prophylactic surgical removal is indicated. This article reports a case in which a patient with dysgenesis presented 45,X/46,XY karyotype. Laparoscopic prophylactic gonadal resection was performed. Laparoscopy plays an important role in exploring pelvic structures and allows the diagnosis, prognosis and treatment of lesions. In addition, it provides an effective method for gonadectomy. The prophylactic removal of dysgenetic gonads using this surgical technique has been preferred in selected patients lately.

Key-words: Dysgenetic gonads; gonadoblastoma; laparoscopy; gonadectomy.

Mulheres com cariótipo XY e gônadas em fita são consideradas portadoras de disgenesia gonadal. Essas pacientes apresentam um risco mais alto de desenvolver doenças nas gônadas. Em tais casos, a remoção cirúrgica profilática é indicada. Este artigo relata o caso de uma paciente com disgenesia, apresentando cariótipo 45,X/46,XY. Foi realizada ressecção profilática das gônadas através de laparoscopia. A laparoscopia tem um papel importante na exploração de estruturas pélvicas e permite o diagnóstico, o prognóstico e o tratamento de lesões. Além disso, oferece um método efetivo para a gonadectomia. A remoção profilática das gônadas disgenéticas utilizando essa técnica cirúrgica tem sido preferida em pacientes selecionados.

Unitermos: Gônadas disgenéticas; gonadoblastoma, laparoscopia, gonadectomia.

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INTRODUCTION

Turner's syndrome occurs in 1:5000 births, presenting a 45,X chromosome constitution in 50% of the cases. The other cases are caused by mosaicism, i.e., the presence of two cell lines, especially 45,X/46,XX and structural changes in X chromosome. Approximately 5% of the patients present Y chromosome in a cell line (1). Patients who are phenotypically female, with XY karyotype and streak gonads, are classified as having XY gonadal dysgenesis. An XY karyotype with Turner stigmata, but with

ambiguous external genitalia, is a chromosomal form of what is called mixed gonadal dysgenesis. In such cases, a 46,XY/45X karyotype is common, and the importance of this type of dysgenesis is the possibility that the gonads may develop malignancy and require prophylactic surgical removal (2-4).

The development of laparoscopy began in 1940 with the studies of Raoul Palmer in Paris. Since then, it has been used as a method to explore the pelvis; more recently, its surgical therapeutic use has been extended to the diagnosis, prognosis and treatment of lesions. All these steps can be performed in a single surgical procedure (5). The preservation of organic integrity and function, along with the identification of unproved pathologies of the genital tract, has revolutionized surgery in this area. The use of laparoscopy in gynecological emergencies (extrauterine pregnancy and infections), infertility problems (adhesions, tubal lesions and endometriosis), tumors, cysts, uterine prolapse and urinary incontinence has grown considerably (6).

The laparoscopic surgical resection of the gonads was recently described by several authors as presenting a reduction of costs, post-surgical complications and hospitalization in comparison to laparotomy (2,7-12). Thus, our objective is to report a case of dysgenetic gonad, and discuss the importance and usefulness of laparoscopy to gonadal resection.

CASE REPORT

An 18-year-old, black female patient was seen at the Department of Gynecology and Obstetrics of Hospital de Clínicas de Porto Alegre to investigate primary amenorrhea in October 2000. She was the seventh daughter of an unrelated couple, out of eight siblings, and no similar cases have been reported in the family.

At physical examination, some positive data were found. She weighed 41 kg, her height was 147.5 cm, and her arm span was 149.5 cm. She had a longish face, high palate, short neck, asymmetrical breasts - Tanner M1 on the left and M2 on the right -, bilateral shortening of the 4th metacarpal, hyperconvex nails and partial syndactyly between the second and third toes.

The karyotype was studied using a lymphocyte culture, which showed two 45,X/46,XY cell lines, with a proportion of 26% and 74%, respectively.

The patient was submitted to prophylactic gonadectomy by means of a laparoscopy in July 2001. After general anesthesia, a puncture was performed to introduce CO_2 and to form a pneumoperitoneum. Then, the trocar was introduced in order to insert a 10 mm endoscope, and two auxiliary punctures were performed to place other 5 mm trocars through which instruments were introduced for surgical manipulation. The pelvis was visualized and bilateral streak gonads were identified. After exposure of the right gonad, using a grasping forceps for traction, it was pulled with identification of the mesosalpinx. Cautery was performed around the gonad, which was excised using scissors and removed through the trocar. The same procedure was performed on the left gonad. The patient was discharged from hospital on the same day.

The anatomopathological examination of the gonads showed bilateral gonadoblastoma.

The patient was then hospitalized to define the stage of the gonadoblastoma. Abdominal CT scan was performed, showing multiple renal cysts. The chest CT scan was normal, as well as the b-chorionic gonadotropin, a-fetoprotein, lactate dehydrogenase and liver function tests. Since no signs of disease were found during the staging tests, and the patient presented full clinical remission, she was considered cured. Later, she underwent hormone replacement therapy and follow-up as an outpatient.

DISCUSSION

The tumors most frequently found in dysgenetic gonads are dysgerminoma and gonadoblastoma, and the former can occur in women with normal ovaries. The gonadoblastoma was initially described by Scully (13). It is a rare benign tumor that has the potential of malignant transformation. It was defined as a gonadal tumor that secretes steroid hormone, and is made up of germ cells and elements derived from the sex cords. These elements are similar to granulosa or Sertoli cells and, sometimes, to stromal elements which look like lutein or Leydig cells. Frequently, it is bilateral. However, when the gonadoblastoma is unilateral, it is the right gonad that is most commonly involved. It develops in 22% of the streak gonads, in 18% of the cases of cryptorchidism, and in 60% of the cases in which the original gonad structure is obliterated by the tumor (13,14). The pure gonadoblastomas are usually small to medium (< 1-8 cm); however, the growth of other germ cell elements often results in very large masses. Macroscopic examination shows a solid tumor with a smooth or lobulated surface, and the presence of calcified granules often occurs (15). The appearance

of the ovary of this patient was normal, although the anatomopathological examination showed a bilateral gonadoblastoma. Often, gonadoblastomas are associated with dysgerminomas (16) and other malignant elements of germ cells (2,17). Clinical prognosis is related to the presence or absence of these elements. Pure gonadoblastomas, in contrast to dysgerminomas, do not metastasize (15).

The chances of developing malignancy are approximately 25% in dysgenetic gonads. It occurs in relatively young individuals, adolescents and young adults, with primary amenorrhea and a flawed development of secondary sexual characteristics (2,18,19). Our patient was 18 years-old and presented primary amenorrhea and scarce axillary and pubic hair.

The highest rate of gonadoblastoma occurs in patients with Turner's syndrome, who have a mosaic chromosomal constitution and the presence of normal or altered Y chromosome (20). The patient presented a 45,X/46,XY karyotype, and the 46,XY cell line constituted 74%. Knowing that the risk of a neoplasm in streak gonads with an XY karyotype is high, their prophylactic removal is recommended.

Laparoscopy has been employed as a surgical procedure because it presents several advantages when compared to laparotomy. It is a simpler and safer procedure, causing less discomfort to the patient, reducing the length of stay in hospital and allowing a quicker resumption of daily activities. In addition, when diagnosis is uncertain, laparoscopy makes it possible, allowing also for a proper treatment (2,4,18,21). Some of the risks of laparoscopic gonadectomy are the need of general anesthesia, intestinal injury, laceration of vessels, pneumothorax, diminished venous return, gas embolism and cardiac arrhythmias.

Therefore, we should perform gonadectomy in patients with XY karyotype and abdominal gonads because of the increased risk for tumor development. In addition, laparoscopy is the preferred surgical approach, reducing post-operative risks and costs.

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