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CASE REPORT

Acute labio-scrotal pain in a patient with ovotesticular syndrome. Case report

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Abstract Ovotesticular syndrome (OTS) belongs to the group of disorders of sex development (DSD). We present a case of a patient with OTS presenting with acute labioscrotal pain. A surgical exploration was indicated, and hemorrhage was identified. A gonadectomy was performed and the final pathology report revealed an ovotestis with a bleeding follicle, normal ovarian parenchyma and atrophic testicular parenchyma. After reviewing the literature there is scarce information on this complex topic, but conservative management could be an option if the risk of a gonadal malignancy is low.

Introduction

Ovotesticular DSD belongs to the group of disorders of sex development (DSD). The principal characteristic of this syndrome is the presence of testicular as well as ovarian tissue in the same individual [1]. Diagnosis is most frequently made in the early postnatal period, but can be delayed until puberty, or suspected before birth with improved prenatal imaging and knowledge. A multidisciplinary approach is needed and an emergent diagnosis needs to be made. Follow-up is needed. The psychological and social outcome have been extensively studied, and the natural history, fertility, and malignancy potential of the testicular parenchyma. Acute labio-scrotal pain is not a common presentation; we therefore present a rare case of ovotesticular syndrome presenting with acute labio-scrotal pain.

Case report

A 13-year-old female with 46,XX ovotesticular DSD was seen for the first time when scheduled for a hernia repair at the

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age of two years old. At birth she was assigned and then raised as a male by her parents. Prior history was unavailable because she was born in a different hospital. During the herniorrhaphy, the physical examination showed a 2 cm genital tubercle with a proximal meatus. A palpable 2 cc canalicular gonad was palpated on the right side. Intraoperatively a gonad with rudimentary Fallopian tube structures was seen inside the hernia sac on the left side. A biopsy was taken and the pathological evaluation revealed findings compatible with an ovary. A biopsy was also performed on the right, which revealed a normally developed testicle. The karyotype performed then showed a normal 46,XX pattern. No feminizing surgery or gonadectomy was performed at that time because of the parents' wishes and because by law, in Colombia, it is prohibited to perform any feminizing procedure after the age of four, and it is mandatory to wait until the patient decides by him/herself to undergo any such surgical correction after puberty. From that moment on sex was re-assigned to female.

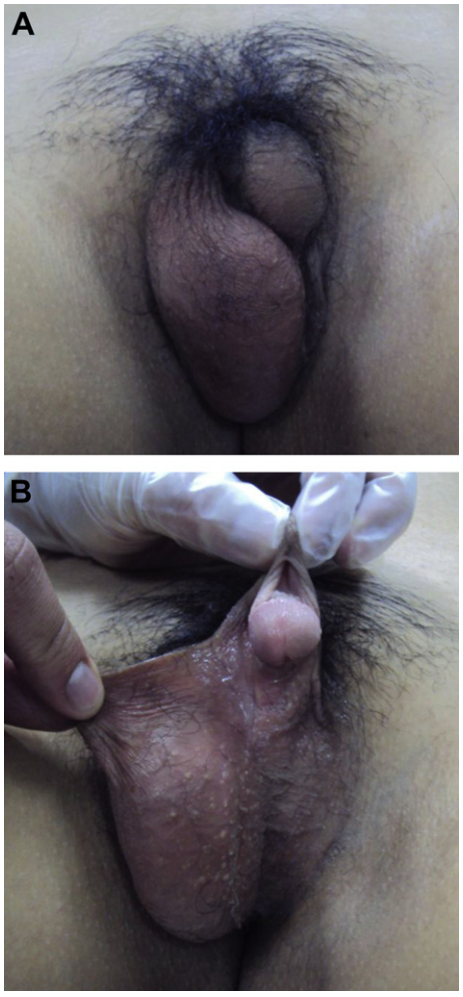

Figure 1 A. Picture taken at the ER. On the right side the labio-scrotal swelling has augmentation of the previous volume with edema, mild erythema and exquisite pain to palpation. The gonad on that side is 5 times larger. No cremasteric reflex is present. B. On the left side the labioscrotal swelling is underdeveloped with no palpable gonads.

Patient continued her normal social life without any complications or controls until she developed Hashimoto's thyroiditis at the age of 12 years old. The patient failed to follow up prior to this due to the parents' wishes. She was treated for her condition successfully. A psychological evaluation showed no alterations with a normal social life, good family support, and even normal interactions and attraction to boys. Furthermore, she had menarche at the age of 13 with regular subsequent cycles. Menstruation was seen coming out of the opening at the base of the genital tubercle. Her physical examination at that time showed: Tanner stage 2—3 breast development with axillary hair present, and pubic hair in a male distribution. No areas of hirsutism were seen. A 5 cm genital tubercle with a proximal single opening at its base was noted. The glans size was 1 cm in diameter. The right gonad was still palpable, approximately 2 cc in volume. On the left, no palpable

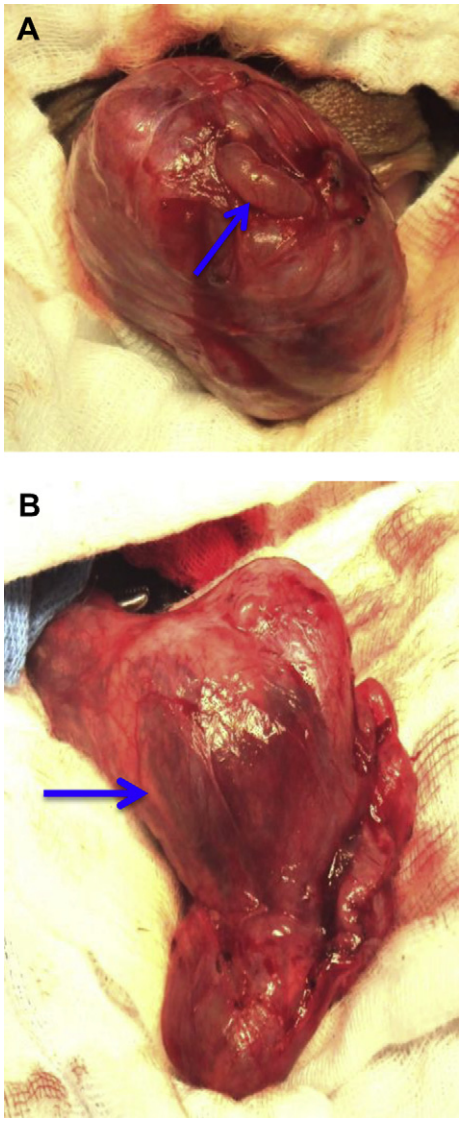

Figure 2 A. Right Gonad without torsion. Arrow shows a rudimentary Falopian tube. B. Arrow shows a blueish discoloration that after specimen exploration presented a focal area of hemorrhage.

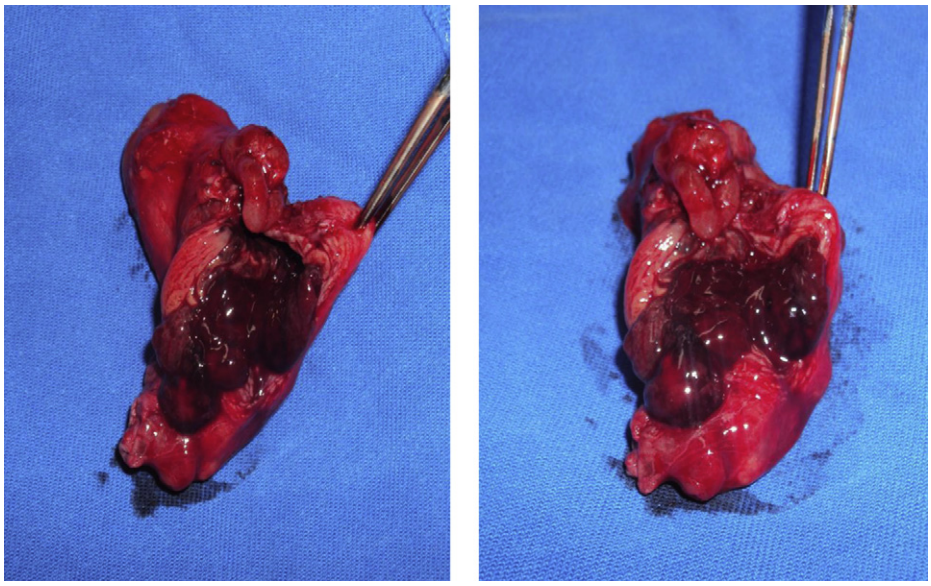

Figure 3 Specimen's exploration presented a focal area of hemorrhage.

gonad was felt. An abdominal ultrasound showed a normally developed uterus. Hormonal studies showed an estradiol level of 55 pg/mL, FSH 5.9 mIU/mL, LH 4.3 mIU/mL, and testosterone level 0.72 ng/mL.

A few weeks later she presented to our reference center emergency room after four days of labio-scrotal pain that increased in severity and intensity. She referred no other symptoms at that time. Physical examination revealed an increase in right labio-scrotal volume with mild erythema and exquisite tenderness to palpation (Fig. 1A and B). A gonadal ultrasound was performed, with findings compatible with gonadal torsion.

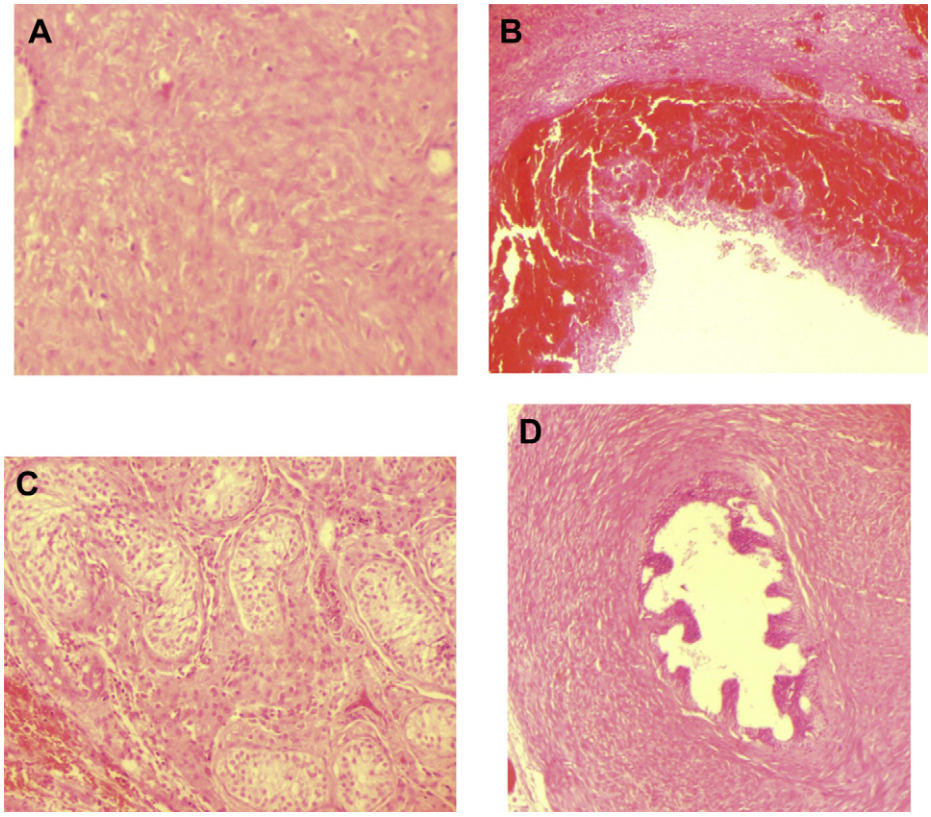

Figure 4 A. H and E of normal ovarian parenchyma. B. Focal area of hemorrhage due to a bleeding follicle inside the ovariansegment of the ovotestes. C. Testicular parenchyma of the ovotestes with lack of germ cells and atrophic Sertoli cells and significant peritubular fibrosis. D. Vas deferens.

She was immediately taken to the OR, and a scrotal exploration was performed. No torsion was identified but instead the gonad with no defined anatomy presented a discolored area compatible with hemorrhage. Rudimentary fallopian tube was seen macroscopically. A gonadectomy was performed at that moment given the possibility of a gonadal tumor (Figs. 2A and B, 3). Laboratory studies performed in the immediate postoperative period showed an alphafetoprotein level of 3.52 ng/mL, βHCG less than 1.2 mIU/mL, LDH 353 U/L and a testosterone level of 0.276 ng/mL.

Final pathology report after gonadectomy was compatible with a 50% normally developed ovary, with a bleeding follicle. The rest of the pathology showed atrophic testicular parenchyma. A well developed vas deferens was also reported (Fig. 4A, B, C and D).

There were no postoperative complications. Plans are to follow up on the patient's masculine characters (clitoromegaly, palpable gonad on the right side) by physical examination, and evaluate for changes since there is no longer testicular parenchyma remaining.

Discussion

Ovotesticular syndrome (OTS) is a rare condition characterized by the presence of testicular as well as ovarian parenchyma in the same individual. The most common genotype is 46,XX. Since the available information comes from case reports and case series, the true incidence of this condition is unknown. The global incidence of DSD is reported to be around 1/5500 newborns [1].

After an extensive review of the current literature on EMBASE, Medline and PubMed, scarce information on similar cases was found. One such case was identified by Bani et al. [2]. Another case reported by Eberez et al. [3] presented a 15-year-old boy with gynecomastia and testicular pain who was diagnosed with OTS. After he underwent bilateral partial gonadectomy, he presented back to the ER with acute scrotum. A second exploration was performed and multiple cysts were identified and excised. No gonadal torsion was described and nor was a bleeding follicle identified. The third case was reported by Aparicio et al. [4]: a 50-day-old male-assigned boy presented to the ER with irritability and labio-scrotal edema and erythema. Surgical excision was performed and a hemorrhage was identified inside an ovotestes. Another patient with complete insensitivity to androgen syndrome who presented with testicular torsion was reported by Arikan et al. [5]. The only similarity between their case and ours is the acute onset of pain and the necessity for urgent surgical intervention in a patient with DSD. Their case did not develop acute labio-scrotal pain and the main condition was not OTS. There are other similar cases documented [6—8]. Our case represents one of a few such cases reported in the literature. From our experience and after reviewing the literature, it is important to keep in mind that acute labio-scrotal pain in a patient with OTS can be due to a bleeding follicle and is not always because of gonadal torsion. There is no relation between age and clinical presentation as well as laterality.

Conclusions

Ovotesticular syndrome is a very rare condition, and rarely presents with acute labioscrotal pain. Given the complexity of OTS patients, it is recommended to evaluate these patients with a multidisciplinary team even in the acute clinical setting. There is little information about surgical management in these complex cases, and the few published articles are all case reports.

If a tumor is a possibility, the surgical approach should be done by an inguinal incision and a frozen biopsy should then be sent to pathology in order to define whether a gonadectomy is necessary or not. Conservative management is indicated in the case of a bleeding cyst, but the incidence of subsequent development of gonadal tumors in conservatively managed patients is unknown.

Conflict of interest/funding

None.

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