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Bilateral Congenital Anorchidism: Case Study

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Abstract **Case Report**

Embryonic testicular regression syndrome (ETRS) or congenital bilateral anorchidism (CBA) is a highly rare syndrome defined by the complete absence of testicular tissue in a patient with a male karyotype. The phenotype varies depending on when gonadal regression occurs in utero. Clinically, a micropenis, hypoplastic scrotum, empty, without perceived testicles are observed. Hormonal assessments reveal collapsed or completely absent testosterone levels and AMH (anti-Mullerian hormone) levels. Diagnosis is confirmed by laparoscopic exploration. Management relies on androgen supplementation and placement of testicular prostheses.

Keywords: Anorchidism, Testicle, Regression, AMH, Laparoscopy, Androgen Therapy.

INTRODUCTION

Embryonic testicular regression syndrome or congenital bilateral anorchidism is defined by the complete absence of testicular tissue in a patient with a normal male karyotype [1]. Its prevalence is estimated at 1 case per 20,000 male subjects but is often underestimated and clinically often mistaken for bilateral intra-abdominal cryptorchidism [2]. Its etiology is still debated but its familial occurrence suggests a genetic etiology [2]. Diagnosis requires paraclinical investigations and may involve exploratory laparoscopy to confirm the diagnosis. Management involves androgen therapy as well as placement of testicular prostheses [3].

We report a case of a young patient who was diagnosed with congenital bilateral testicular agenesis. The patient was placed on androgen supplementation and testicular prostheses were implanted.

The aim of this work is to describe the diagnostic, therapeutic, and evolutionary modalities of a case of congenital bilateral anorchidism.

CASE PRESENTATION

This concerns a 17-year-old patient with no particular medical or surgical history, who, initially at birth, was referred to Pediatric Surgery for suspicion of bilateral cryptorchidism. Neonatal examination revealed a micropenis, hypoplastic scrotum without palpable testicles on both sides. Hormonal assessment showed a collapse in testosterone, AMH, and inhibin B levels. Magnetic Resonance Imaging (MRI), which was performed, did not find any testicles. Exploration through laparoscopy confirmed the diagnosis of congenital bilateral testicular agenesis. The child was placed on delayed-release testosterone, and the evolution was satisfactory with a normal-sized penis, a well-placed urethral meatus, and normal secondary sexual characteristics according to age.

At the age of 15, delayed-release testosterone was replaced by androgen supplementation with Androtardyl, 1 intramuscular injection monthly, and the evolution remained normal.

At the age of 17, the patient was seen in urological consultation to discuss the placement of testicular prostheses. Physical examination revealed a patient in good general condition, with a height of 1.68 meters, a normal-sized penis, empty scrotum (Photo A), facial hair, and presence of pubic and axillary hair. Surgical intervention was arranged to place bilateral testicular prostheses via an inguinal approach. The intervention proceeded without any incidents or complications intraoperatively or postoperatively.

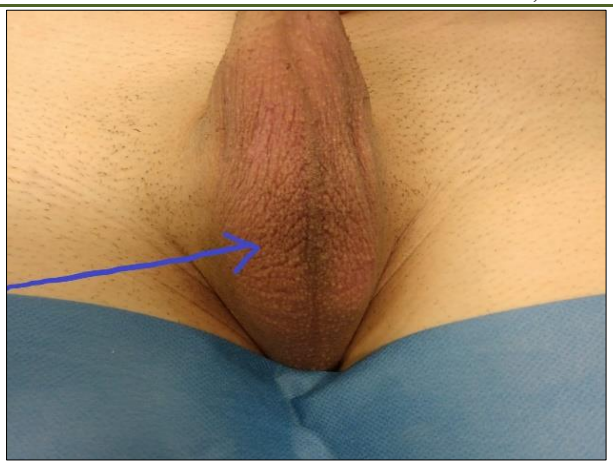

Photo A: Empty scrotum (preoperative)

Following the placement of the testicular prosthesis, the patient regained a normal scrotal morphology (Photo B).

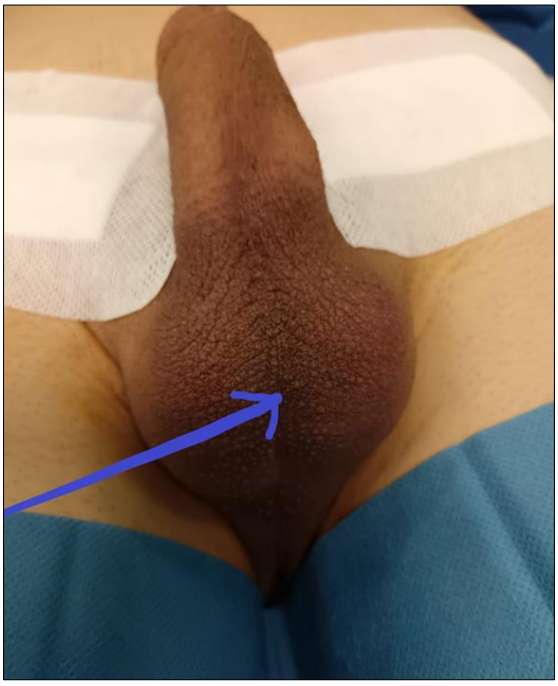

Photo B: Testicular prostheses in place (postoperative)

DISCUSSION

The syndrome of embryonic testicular regression is defined by a partial or complete absence of testicular tissue in the presence of a 46 XY karyotype [1]. Most patients with this condition present with a sexual differentiation anomaly or a micropenis with complete regression of testicular tissue, which can be unilateral or bilateral. The degree of masculinization of external and internal genital organs depends on the duration of testicular function before its collapse, explaining the phenotypic variability [4, 5]. Congenital testicular agenesis affects approximately 5% of cryptorchid children [6, 7], which accounts for the underestimation of this diagnosis, as it is not as rare as commonly thought. Measurement of anti-Mullerian hormone (AMH), as a marker of the presence of testicular tissue, shows extremely low or undetectable levels with no response of plasma testosterone to HCG (Human chorionic gonadotropin) stimulation [2-9].

Diagnostic confirmation can only be provided by laparoscopic exploration [6-12]. Treatment consists of substitutive androgen therapy [3], and the placement of testicular prostheses can be performed for psychosocial and aesthetic reasons [13].

CONCLUSION

Testicular agenesis is a very rare congenital malformation. An undetectable or collapsed AMH level associated with no response of plasma testosterone to HCG stimulation suggests a diagnosis of anorchidism, which is confirmed by laparoscopic exploration. Current

therapeutic options include androgen therapy combined with testicular prosthesis placement. Psychological support for parents and the patient himself is necessary to improve the experience of the situation into adulthood.

REFERENCES

1. Abeyaratne, W. A., Aherne, W. A., & Scott, J. E. (1969). The vanishing testis. *Lancet*, *2*(7625), 822-841.
2. Philibert, P., Zenaty, D., Lin, L., Soskin, S., Audran, F., Léger, J., ... & Sultan, C. (2007). Mutational analysis of steroidogenic factor 1 (NR5a1) in 24 boys with bilateral anorchia: a French collaborative study. *Human Reproduction*, *22*(12), 3255-3261.
3. Pierson, M., Vidailhet, M., Wuilbercq, L., Wendremaire-Palandri, Y., & Combescure, B. (1983). Total testicular regression syndrome or anorchidism. *Archives Francaises de Pediatrie*, *40*(10), 767-773.
4. Mendonca, B. B., Domenice, S., Arnhold, I. J., & Costa, E. M. (2009). 46, XY disorders of sex development (DSD). *Clinical endocrinology*, *70*(2), 173-187.
5. Josso, N., & Briard, M. L. (1980). Embryonic testicular regression syndrome: variable phenotypic expression in siblings. *The Journal of pediatrics*, *97*(2), 200-204.
6. Pirgon, Ö., & Dündar, B. N. (2012). Vanishing testes: a literature review. *Journal of Clinical Research in Pediatric Endocrinology*, *4*(3), 116.
7. Spires, S. E., Woolums, C. S., Pulito, A. R., & Spires, S. M. (2000). Testicular regression syndrome: a clinical and pathologic study of 11 cases. *Archives of pathology & laboratory medicine*, *124*(5), 694-698.
8. Huff, D. S., Wu, H. Y., Snyder III, H. M., Hadžiselimović, F., Blythe, B., & Duckett, J. W. (1991). Evidence in favor of the mechanical (intrauterine torsion) theory over the endocrinopathy (cryptorchidism) theory in the pathogenesis of testicular agenesis. *The Journal of urology*, *146*(2), 630-631.
9. Noel, M., Chevenne, D., Nicolas, M., Rigal, O., Kertsz, G., Drai, L., ... & Guibourdenche, J. (2003). Biochemical investigations of the testicular tissue: establishment of reference values and their clinical interest. *IMMUNOANALYSE ET BIOLOGIE SPECIALISEE*, *18*(5), 277-282.
10. Hegarty, P. K., Mushtaq, I., & Sebire, N. J. (2007). Natural history of testicular regression syndrome and consequences for clinical management. *Journal of pediatric urology*, *3*(3), 206-208.
11. Stéfaniu, M., Lefébure, B., Liard-Zmuda, A., & Bachy, B. (2004). Testicules impalpables. Place de la coelioscopie. *Archives de pédiatrie*, *11*(4), 315-318.
12. Hassan, M. E., & Mustafawi, A. (2010). Laparoscopic management of impalpable testis in children, new classification, lessons learned, and rare anomalies. *Journal of laparoendoscopic & advanced surgical techniques*, *20*(3), 265-269.
13. Sonne, S. B., Kristensen, D. M., Novotny, G. W., Olesen, I. A., Nielsen, J. E., Skakkebæk, N. E., ... & Leffers, H. (2008). Testicular dysgenesis syndrome and the origin of carcinoma in situ testis. *International journal of andrology*, *31*(2), 275-287.