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Case Series
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Testicular Regression Syndrome: A Series of 7 Cases

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Abstract

Introduction: Testicular regression syndrome (TRS) is a rare cause of disorders of sex development in 46,XY individuals. It corresponds to the partial or complete disappearance of testicular tissue following initially normal male differentiation.

Objective: To describe the clinical, biological, radiological, and evolutionary characteristics of a series of 7 patients followed for TRS, and to compare them with recent literature data.

Materials and Methods: Retrospective descriptive study conducted on 7 46,XY patients followed at an endocrinology consultation center in Rabat. Clinical, hormonal, morphological, and surgical data were analyzed.

Results: The mean age at diagnosis of TRS was 9.4 ± 3.6 years (range: 2–12 years). All patients had absent palpable gonads, low or undetectable testosterone levels, a hypergonadotropic hypogonadism profile, and a 46,XY karyotype. Imaging was negative in 100% of cases. Laparoscopy, performed in all patients, confirmed the absence of functional testicular tissue. Androgen replacement therapy was initiated in all patients with good clinical response.

Conclusion: TRS should be considered in any case of bilateral anorchia in a 46,XY individual. Laparoscopy remains the reference examination to confirm diagnosis. Management is based on androgen replacement therapy and psychological support.

Keywords: Testicular Regression Syndrome (TRS); Disorders of Sex Development (DSD); 46,XY Karyotype; Bilateral Anorchia; Gonadal Dysgenesis; Hypergonadotropic Hypogonadism

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Introduction

Testicular regression syndrome, also known as "vanishing testis syndrome," is defined by the congenital or acquired absence of testicular tissue in 46,XY individuals with normally differentiated male external genitalia. It accounts for approximately 3 to 5% of disorders of sex development in boys [1, 2].

The most accepted pathophysiology involves a late antenatal or perinatal vascular accident, such as testicular torsion or thrombosis, leading to progressive atrophy and subsequent disappearance of gonadal tissue [3–5].

Materials and Methods

This is a retrospective descriptive study including 7 patients followed for confirmed TRS. For each patient, the following data were collected:

- Age at diagnosis of TRS,
- Clinical data (height, penis size, body hair, presence or absence of micropenis),
- Hormonal assays (FSH, LH, basal and post-hCG testosterone, AMH),
- Imaging results (ultrasound, CT scan),
- Surgical exploration data (laparoscopy),
- Evolution under androgen replacement therapy.

Clinical Observations

Case 1: A 15-year-old patient with no notable history, consulting for delayed puberty. Clinical examination showed tall stature (178 cm), macroorchidism, normal penis size, and absence

Table 1: Clinical, Biological, and Morphological Characteristics of Our Patients.

Case	Age at Diagnosis (years)	Karyotype	Penis Size	Basal Testosterone	hCG Test	Laparoscopy	Androgen Replacement Therapy
1	15	46,XY	Normal	Low	Negative	Absence of gonads	Yes, good evolution
2	2	46,XY	Micropenis	Low	Negative	Testicular remnant	Yes, good evolution
3	17	46,XY	Normal	Low	Negative	Absence of gonads	Yes, good evolution
4	11	46,XY	Normal	Very low	Negative	Absence of gonads	Yes, good evolution
5	6	46,XY	Micropenis	<0.1 ng/mL	Negative	Absence of gonads	Yes, good evolution
6	11	46,XY	Borderline	Undetectable	Negative	Absence of gonads	Yes, good evolution
7	12	46,XY	Normal	Undetectable	Negative	Absence of gonads	Yes, good evolution

of palpable gonads in the scrotal or inguinal position. Axillary, pubic, and facial hair were absent. Hormonal assessment revealed hypergonadotropic hypogonadism with low testosterone, FSH 40 mIU/mL, and LH 8 mIU/mL. Karyotype was 46,XY. Imaging and laparoscopy did not identify testicular tissue.

Case 2: An 18-year-old patient followed for TRS diagnosed at 2 years of age. Initial clinical examination showed micropenis and bilateral anorchia. Hormonal profile was consistent with hypergonadotropic hypogonadism. Laparoscopy revealed a 13 mm right inguinal testicular remnant without spermatic cord and a left cystic formation. The patient was placed on androgen replacement therapy, with currently favorable clinical evolution: normal penis size and grade 5 body hair. Testicular prostheses placement and psychological support are planned.

Case 3: A 17-year-old patient followed for delayed puberty. He measured 1.92 m, had macroorchidism, normal penis size, and P2 pubic hair. Gonads were absent on palpation. Hormonal profile showed hypergonadotropic hypogonadism with a negative hCG test. Karyotype was 46,XY. Exploratory laparotomy found no gonads.

Case 4: An 11-year-old patient presenting with normal penis size and bilateral absence of palpable gonads. Karyotype was 46,XY. Basal testosterone was very low (0.01 ng/mL) and remained low after hCG stimulation. Laparoscopy did not identify testicular tissue.

Case 5: A 26-year-old patient followed for TRS diagnosed at age 6. Initial examination revealed micropenis (2 cm length, 0.5 cm width) and bilateral anorchia. Testosterone was <0.1 ng/mL with no response to hCG test. AMH was low (0.7 ng/mL). Laparoscopy confirmed absence of gonads.

Case 6: A 20-year-old patient followed since age 11 for TRS. Penis size was borderline (3 cm length, 2 cm width). Basal testosterone was undetectable and remained <0.5 ng/mL after hCG. AMH was low (0.2 ng/mL). Laparoscopy confirmed absence of testicular tissue.

Case 7: A 23-year-old patient followed for TRS diagnosed at age 12. Penis size was normal. Testosterone was undetectable, with no response to hCG test. Ultrasound and laparoscopy visualized no gonads.

Global Clinical Summary

Seven male patients, all with 46,XY karyotype, were included. The mean age at diagnosis was 9.4 ± 3.6 years (range: 2–12 years). All patients had bilateral absence of palpable gonads. Micropenis was found in 3 patients (42.8%). All exhibited hypergonadotropic hypogonadism with low or undetectable testosterone and no response to hCG stimulation. AMH, measured in some patients, was low or undetectable. Ultrasound imaging was negative in all cases. Laparoscopy confirmed the absence of testicular tissue in all patients. All patients received androgen replacement therapy with good clinical response (Table 1) (Figure 1).

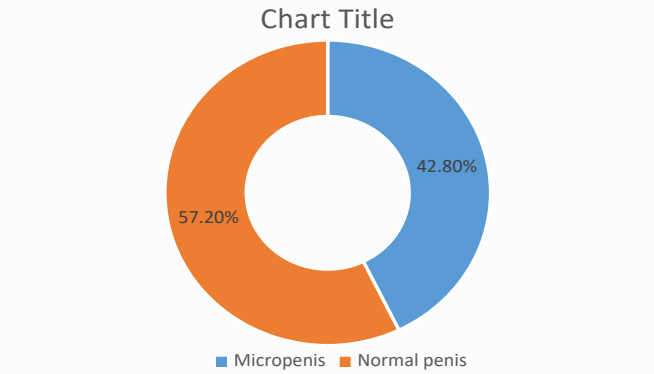

Figure 1: Clinical distribution of patients with micropenis versus normal penis size.

Discussion

Testicular regression syndrome (TRS) is a rare form of 46,XY disorders of sex development characterized by partial or complete disappearance of testicular tissue after initially normal male differentiation [1–3].

In our series, the mean age at diagnosis was 9.4 years, consistent with the literature which reports diagnosis usually made between late childhood and adolescence [4–6]. Clinical presentation depends on the timing of regression: early regression may cause micropenis or genital ambiguity, while late regression often manifests as delayed puberty or isolated bilateral anorchia [7, 8]. In our series, 42.8% of patients had micropenis, a proportion similar to other reports [5, 9].

All our patients had hypergonadotropic hypogonadism, with low or undetectable testosterone and no response to hCG stimulation, confirming the absence of functional testicular tissue, as described in the literature [10–12]. AMH is a sensitive marker of Sertoli cell function, and its absence confirms the severity of TRS [13, 14].

Ultrasound, although non-invasive, has limited sensitivity for detecting testicular remnants, consistent with our negative results in all cases [15]. Exploratory laparoscopy remains the reference examination to confirm absence of testicular tissue and to exclude intra-abdominal cryptorchidism [16–18]. In our patients, it confirmed the diagnosis in 100% of cases. TRS must be differentiated from bilateral intra-abdominal

cryptorchidism, congenital anorchia, or pure gonadal dysgenesis [19, 20]. The 46,XY karyotype excludes other syndromes such as Klinefelter syndrome.

Management is based on androgen replacement therapy, essential for inducing and maintaining secondary sexual characteristics, bone growth, and overall health, to prevent hypogonadism-related complications such as osteoporosis, and to improve patients' quality of life [21–23]. In our series, all patients benefited from this treatment with good clinical response, including development of pubic, axillary, and facial hair, and normal or borderline penile growth. Placement of testicular prostheses is often proposed to improve body image and psychological well-being [24]. Psychological support is essential to help patients cope with the psychosexual impact of this rare condition [25].

Conclusion

Testicular regression syndrome is a rare but essential cause of bilateral anorchia in 46,XY individuals. Diagnosis is based on a combination of clinical, hormonal, and surgical evidence, with laparoscopy as the reference examination. Early management with androgen replacement therapy and tailored psychological support are crucial to improve functional prognosis and quality of life.

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