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Turner Syndrome and the SRY Gene: Report of Two Cases

Gorgi K[1] and Chaouche M[2]*

[1]*Department of Endocrinology and Metabolic Diseases, Ibn Sina University Hospital, Rabat, Morocco*

[2]*Department of Dermatology, Mohammed VI University Hospital, Agadir, Morocco*

Abstract

Turner syndrome (TS) is a rare chromosomal disorder characterized by partial or complete monosomy X. The presence of Y chromosome material, particularly the sex-determining region Y (SRY) gene, has important clinical implications due to the increased risk of gonadoblastoma. We report two cases of young women with Turner syndrome harboring Y chromosome material detected by fluorescence in situ hybridization (FISH), illustrating the phenotypic variability, diagnostic challenges, and therapeutic management, particularly regarding prophylactic gonadectomy.

Keywords: Turner Syndrome; SRY Gene; Y Chromosome; Gonadoblastoma; Prophylactic Gonadectomy; Disorders of Sex Development

Introduction

Turner syndrome (TS) is a chromosomal abnormality affecting approximately 1 in 2,500 live female births and is characterized by the complete or partial absence of one X chromosome [1]. The most common karyotype is 45,X, although mosaic forms such as 45,X/46,XX or 45,X/46,XY are frequently observed [2].

The presence of Y chromosome material in patients with TS has major clinical relevance because it is associated with an increased risk of gonadoblastoma, estimated between 10% and 30% [3, 4]. The sex-determining region Y (SRY) gene plays a central role in male sex differentiation and its detection in TS patients raises important issues regarding gonadal management and oncological surveillance [5].

We report two cases of Turner syndrome with Y chromosome material and SRY gene positivity, highlighting differences in gonadal anatomy and management strategies.

Case Reports

Case 1

A 19-year-old woman was followed for mosaic Turner syndrome (45,X/46,XY) with the presence of the SRY gene detected by fluorescence in situ hybridization (FISH).

Clinical examination revealed a weight of 60 kg and a height of 1.43 m. Pubertal development was classified as Tanner stage S3P3, with no sexual ambiguity.

Pelvic magnetic resonance imaging (MRI) demonstrated a markedly hypoplastic uterus and absence of visible gonadal structures (ovaries or testes).

Laparoscopic exploration revealed a very hypoplastic uterus, and the gonads were reduced to fibrous streaks. After multidisciplinary discussion and detailed explanation to the patient and her family, an ovarian biopsy was performed. Histopathological examination showed fibroadipose tissue with residual gonadal remnants, consistent with complete ovarian atrophy.

Despite the recommendation for prophylactic gonadectomy due to the presence of Y chromosome material, the procedure was refused by both the patient and her family. The patient was subsequently placed on estrogen-progestin hormone replacement therapy, with regular clinical and imaging follow-up.

Case 2

A 20-year-old woman was followed for Turner syndrome with Y chromosome material detected by FISH, including the SRY gene.

Clinical examination showed a patient in good general condition, with a weight of 48 kg, height of 1.41 m, and a body mass index of 25 kg/m². Blood pressure was 110/70 mmHg and heart rate was

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Correspondence:
Dr. Gorgi Khaoula, Department of Endocrinology and Metabolic Diseases, Ibn Sina University Hospital, Rabat, Morocco; Tel: 0615591874; E-mail: khaoulagorgi@gmail.com

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70 beats per minute. Pubertal development was Tanner stage S2P2, with no sexual ambiguity.

Pelvic MRI revealed a normally sized uterus measuring 57 × 17 × 33 mm, with regular contours, an arcuate fundus, thin endometrium (7 mm), and absence of latero-uterine masses. Gonadal structures were not visualized.

Laparoscopic exploration identified gonadal structures suggestive of dysgenetic testes. Prophylactic gonadectomy was performed, with uncomplicated postoperative recovery. Histopathological examination demonstrated stromal cell clusters and germ cells arising from two dysgenetic testes, confirming the diagnosis.

The patient was started on hormone replacement therapy combining estrogen and progestin, with favorable clinical and biological evolution. Annual pelvic MRI surveillance was recommended.

Discussion

Turner Syndrome and Y Chromosome Material

The prevalence of Y chromosome material in Turner syndrome varies widely across studies, ranging from 0% to 61%, depending largely on the sensitivity of the detection methods used, such as conventional karyotyping, PCR, or FISH [6, 7]. The presence of cryptic Y material may be underestimated when sensitive molecular techniques are not systematically employed [8].

Clinical Implications of the SRY Gene

The detection of the SRY gene in TS patients is clinically significant, as it is associated with varying degrees of gonadal differentiation and an increased risk of gonadal tumors, particularly gonadoblastoma [9]. Phenotypic expression can range from normal female external genitalia to ambiguous genitalia or virilization, although many patients, as in our cases, present without sexual ambiguity [10].

Risk of Gonadoblastoma and Management

Multiple studies recommend prophylactic gonadectomy in TS patients with Y chromosome material due to the substantial risk of gonadoblastoma, even in the absence of clinical or radiological abnormalities [11–13]. In our second case, gonadectomy confirmed dysgenetic testes, validating the indication. In the first case, refusal of surgery necessitated careful long-term surveillance, although this approach carries inherent oncological risks.

Hormone Replacement Therapy

Hormone replacement therapy (HRT) is essential in TS patients to induce and maintain secondary sexual characteristics, optimize bone mineral density, and improve quality of life [14]. Both patients received estrogen-progestin therapy with good clinical outcomes.

Conclusion

Turner syndrome associated with Y chromosome material and SRY gene positivity presents significant diagnostic and therapeutic challenges. Systematic screening for Y chromosome material using sensitive molecular techniques should be considered in all patients with Turner syndrome. Prophylactic gonadectomy remains the recommended management to prevent gonadoblastoma, although patient and family preferences must be respected. Long-term multidisciplinary follow-up is essential to optimize outcomes.

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