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CLINICAL CASE

Reconstruction of anatomofunctional in a patient with short urethra, genital dysgenesis, and total incontinence with Turner syndrome in mosaic

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Abstract

The case of a 34-year-old female patient with Turner syndrome (TS) mosaicism (45X, 46XX) and severe urogenital malformations is presented. These malformations include clitoral agenesis, hypoplasia of the labia minora, and a short, patulous urethra that resulted in total urinary incontinence. After a multidisciplinary evaluation and a series of advanced surgical interventions, a significant improvement in urinary function and the patient's quality of life was achieved. This case highlights the clinical and surgical complexity associated with TS mosaicism, a condition that can present atypical and severe manifestations in the urogenital tract. Congenital short patulous urethra in women is a rare condition that delays diagnosis and treatment. Clinical identification, such as total incontinence, is a key sign for precise diagnosis and detailed surgical planning, essential for the effective management of patients with congenital anomalies. Advanced surgical techniques, such as subtrigonal sling placement and mobilization of vaginal wall flaps, have proven to be effective in the treatment of stress urinary incontinence, significantly improving quality of life.

Keywords: *Turner syndrome mosaicism. Urogenital malformations. Urinary incontinence. Genitourinary reconstruction. Subtrigonal sling. Congenital anomalies of the urinary tract.*

Introduction

Turner syndrome (TS) is a disease that affects women. The genetic background of the phenotype is highly variable, and karyotype analysis can improve disease understanding[1]. Previous reports indicate that the most common clinical manifestations are short stature, gonadal dysfunction, renal malformations, and certain phenotypic traits in external sexual organs[2]. When the syndrome presents as a mosaic karyotype, clinical presentations are usually uncommon[3]. Here, we describe the case of a female patient with TS mosaicism (45X, 46XX) with a patulous urethra leading to total urinary incontinence.

Congenital short patulous urethra in women is a rare condition and has been previously reported in association with epispadias. Congenital anomalies affecting the distal segment of the urogenital sinus (which gives rise to the female urethra and vagina) can result in abnormal urethral development ranging from an absent urethra to a markedly deficient urethra, leading to total urinary incontinence[4].

Clinical case

A 34-year-old female patient presented to the clinic with continuous urinary leakage that worsened with effort. As relevant history, she requires diaper

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changes 5 times a day, accompanied by a lack of urinary urge. The patient has two full-term parturient, which is not common in TS in the literature; however, due to mosaicism, it is highly probable that her uterus is healthy and she was able to conceive two children. Throughout her illness, she denied episodes of hematuria or dysuria. The patient was initially evaluated through history taking and physical examination, revealing that the condition began in childhood. Physical examination showed urogenital malformations, such as clitoral agenesis, hypoplasia of the labia minora, and abnormalities in the size of the urethra (Fig. 1).

A urinary tract evaluation was performed through urethrocystography, revealing separation of both pubic branches, a bladder volume of approximately 270 mL prevoiding with onset of incontinence, and spontaneous urine leakage. The micturition phase could not be evaluated due to involuntary urine leakage at 270 mL, and complete emptying was seen in the post-micturition phase (Fig. 2).

In the protocol in patient's TS is necessary a second study to rule out kidney malformations, a computed tomography scan with contrast of the chest and pelvis was performed, showing a decrease in the size of the urethra, fluid exiting through the vagina with no evidence of bladder wall defect, and a normal path of the ureters to the bladder and subsequently to the vagina (Fig. 3).

In collaboration with the genetics department, and due to the abnormal findings, a karyotype analysis was performed, revealing mosaic TS (45X, 46XX) (Fig. 4).

The patient underwent surgery by the urology service. The urethra was reconstructed and the urethral sphincter was increased using autologous fascia lata as a subtrigonal sling. During this process, dense fibrous scar tissue between the urethra and vagina was dissected. After mobilizing the remaining urethra, a flap of the vaginal wall was used to lengthen the urethra and reconstruct the external urethral meatus. Finally, a subtrigonal sling was placed. Synthetic slings are the most common primary surgical treatment for incontinence. Placing a subtrigonal sling is an effective surgical intervention for treating stress urinary incontinence in women. This procedure is performed through two incisions, one vaginal and one abdominal. In the vagina, an opening is created and the tissue is dissected to access the trigonal space formed by the urethra and both ureters, through which a sling is passed that adheres to the adjacent tissue. Following this, reconstruction of the clitoris, labia, and mons pubis was performed. The technique used for the mons pubis was plication with 1 Prolene of the aponeurosis of the rectus abdomen, increasing the subcutaneous cellular tissue in the mons pubis. For clitoris reconstruction, the internal face of the right and left half clitoris was sectioned, fusing with monocryl. Finally, for labial reconstruction, a minor labiaplasty was performed. Subsequently, the patient showed a drastic improvement and was completely dry at the 3-month follow-up (Fig. 5).

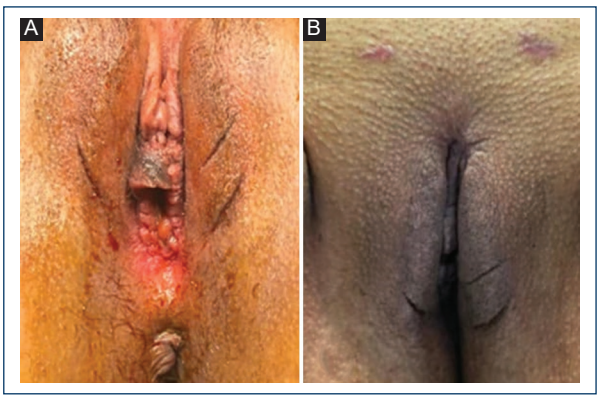

Figure 1. A: pre-operative findings show malformation of the labia majora, unfused clitoris, and hypoplasia of the labia minora. **B:** post-operative findings at 3-month follow-up show fused labia, intact clitoris, with proper healing of the fascia lata sling forming the mons pubis.

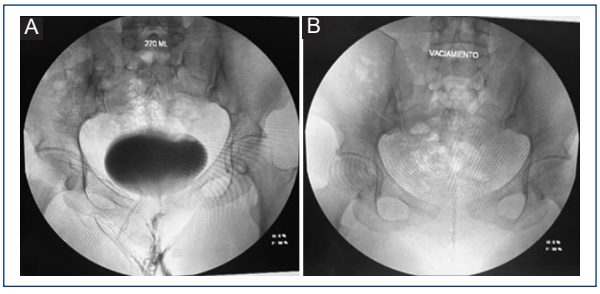

Figure 2. Urethrocystography. **A:** bladder with an approximate volume of 270 mL pre-micturition, well-defined edges, and spontaneous urine output was confirmed. **B:** the voiding phase was not assessable due to the involuntary leakage of urine, although complete emptying was evident in the post-micturition phase.

Discussion

TS can be classified according to the form of X chromosome deletion as follows: 50% of X chromosome deletions are in the classical form of TS (45,X), the mosaic form (45,X/46,XX) represents 15-25%, and the remaining cases of TS have structural abnormalities in the X chromosome[5]. The mosaic form of TS may

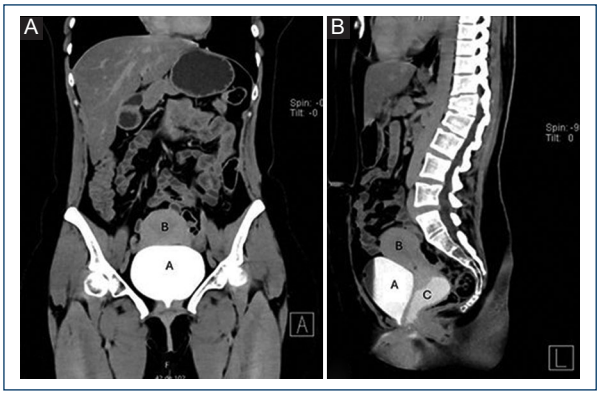

Figure 3. CT urography in coronal A the entire bladder **A:** is observed in the elimination phase with the proximal urethra above the uterus **B:** urography in sagital we observe the bladder (A) above this the uterus (B) in the usual way, on the left side of the image we observe the vagina (C) in which there is passage of contrast medium from the urethra in dorsal decubitus position showing the deficiency of the urethra to retain urine.

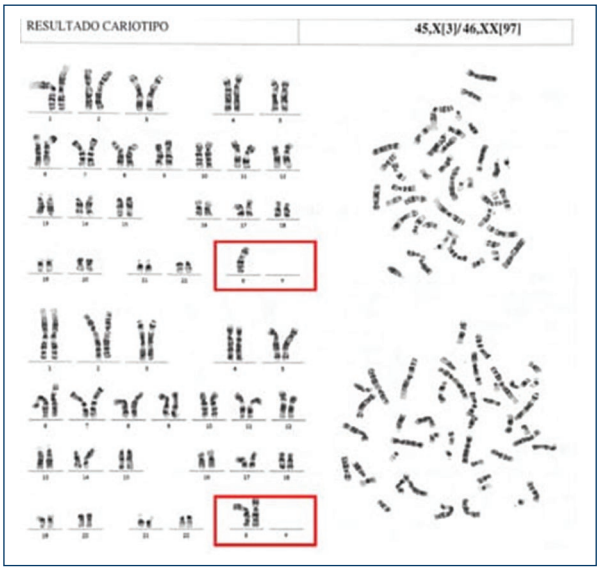

Figure 4. Karyotype results: mosaic with 3 cell lines of monosomy of the affected chromosome (45X), and a chromosome according to normalcy (46XX) in peripheral blood samples.

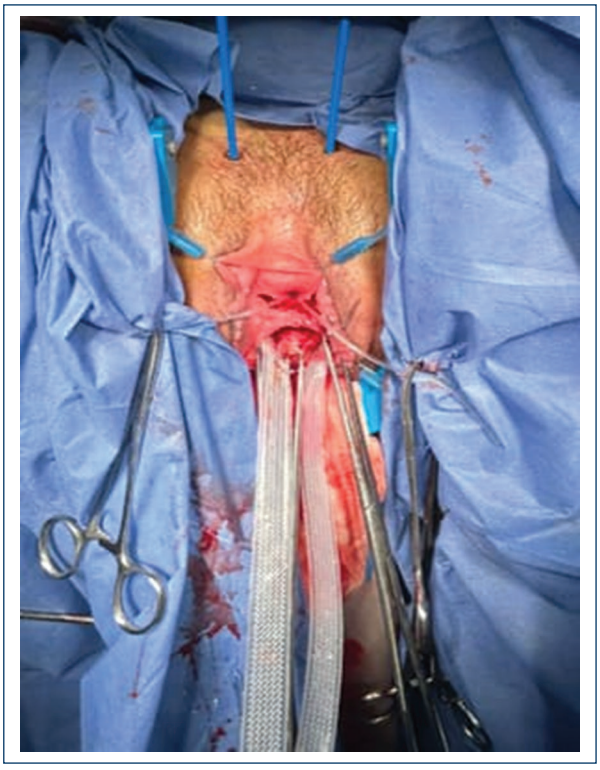

Figure 5. Urethra reconstruction with increased urethral sphincter using autologous polypropylene sling. Dense fibrous scar tissue used as a flap of the vaginal wall to lengthen the urethra and reconstruct the external urethral meatus.

overlook many of the known features of TS, leading to a delayed diagnosis similar to our patient's.

The most common urological clinical manifestations associated with TS include horseshoe kidney, with a frequency of 7-14%. It is also known that in TS, there can be a duplicated collecting system and unilateral renal agenesis. These anomalies are collectively referred to as congenital anomalies of the kidney and urinary tract (CAKUT), which are one of the leading causes of pediatric chronic kidney disease[6]. Although renal function in TS is considered normal, it has not been thoroughly studied. Studies reported in the literature indicate that the frequency of CAKUT in mosaic karyotype is 25.0%[6].

Considering what is reported in the literature and as per protocol, a contrast-enhanced computed tomography was performed to rule out possible malformations associated with TS, the findings were observed the entire bladder is observed in the elimination phase with the proximal urethra above the uterus. The bladder above this the uterus in the usual way, we observe the vagina in which there is passage of contrast medium from the urethra in dorsal decubitus position showing the deficiency of the urethra to retain urine.

The spatiotemporal nature of molecular alterations defines the renal and urinary tract defects of individuals. Early embryonic development can lead to renal parenchymal malformations, while later interferences underlie ureteral anomalies[7].

The female urethral development occurs from the pelvic and phallic part of the urogenital sinus. The abnormal

division of the urogenital sinus with possible malrotation that occurs during development is the probable hypothesis in reported cases of short and wide urethras. The exact embryological defects are unknown[8].

The reported case shares similarities with the phenotype of Turner congenital malformations, such as clitoral agenesis and hypoplasia of the labia minora. However, there is no necessary information to directly link a patulous urethra to the syndrome. Congenital short patulous urethra in women is a rare condition and has been previously reported in association with epispadias, hypospadias, and urogenital sinus anomalies. Congenital anomalies affecting the distal segment of the urogenital sinus (which give rise to the female urethra and vagina) can lead to abnormal urethral development ranging from an absent urethra to a markedly deficient urethra[4].

Youthful women with epispadias will have a bifid clitoris, a patulous urethral meatus, a vaginal opening located anteriorly, and a malformed or absent mons pubis, poorly developed lips, and occasional symphyseal separation[8].

Epispadias is a rare and sporadic congenital defect with a worldwide estimated incidence of 2.4/100,000 live births[9]. The condition is more common in men than in women, with an estimated ratio of 1.4:1, but the incidence in girls is likely to be greatly underestimated due to the difficulty of diagnosis[10].

The retrosymphyseal form can present with complete incontinence and an extremely small bladder capacity. The most severe forms may demonstrate a urethral cleft affecting the bladder neck with prolapse of the bladder mucosa. Genetic, developmental, and environmental factors are believed to play a role in the etiology of epispadias, although the underlying cause is unknown[11]. In women, urinary incontinence is a typical finding in the clinical picture[12].

Urinary incontinence can be simply defined as loss of bladder control or involuntary urination.

Urinary stress incontinence (USI) is the involuntary loss of urine associated with physical effort that increases pressure in the abdomen, such as sneezing, coughing, laughing, or simply walking. It occurs when the intravesical pressure exceeds the urethral opening pressure, as a result of a failure in the mechanisms of urethral resistance, due to two non-exclusive causes: urethral hypermobility, secondary to the weakness of the structures that make up the pelvic floor and support the urethra, and intrinsic sphincter deficiency, caused by a defect in the urethral sphincter closure system, resulting in inadequate coaptation of its walls. Synthetic slings are the most common primary surgical treatment for USI. A recent systematic review reported cure rates of 65-98%[13].

The results demonstrate that the subtrigonal sling technique with abdominal fascia is useful for resolving complex USI, and its effectiveness is comparable to that obtained with the use of suburethral tapes, but with a lower rate of obstructive complications, including urinary retention[14]. This technique was chosen based on the patient's condition, as she had a short urethra that did not have the necessary diameter for a sling. In addition, urethral elongation was performed, so if a synthetic material were to be placed in the reconstructed area, there could be a risk of fistula formation; hence, an independent working area from the urethra was selected.

Female intimate surgery has developed significantly in recent years.

The main objective of genital esthetic surgery in women, as well as in men, is to improve the subjective appearance of the external genital organs and potentially provide psychological or functional improvement in sexual satisfaction[15].

Conclusion

This case highlights the complexity and challenges in genitourinary reconstruction of a patient with TS mosaicism (45X, 46XX) and congenital anomalies in short urethra and urinary incontinence. The treatment of short urethra and severe urinary incontinence due to a deteriorated embryological defect, as part of a developmental anomaly, justifies multiple surgical reconstructions. In this case, socially acceptable continent periods were achieved through advanced surgical interventions and a multidisciplinary approach. However, more information and research are needed to reach more accurate conclusions about the optimal management and long-term implications of these conditions.

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Conflicts of interest

The authors declare no conflicts of interest.

Ethical considerations

Protection of humans and animals. The authors declare that no experiments involving humans or animals were conducted for this research.

Confidentiality, informed consent, and ethical approval. The authors have followed their institution's confidentiality protocols, obtained informed consent from patients, and received approval from the Ethics Committee. The SAGER guidelines were followed according to the nature of the study.

Declaration on the use of artificial intelligence. The authors declare that no generative artificial intelligence was used in the writing of this manuscript.

References

1. Cui X, Cui Y, Shi L, Luan J, Zhou X, Han J. A basic understanding of turner syndrome: incidence, complications, diagnosis, and treatment. Intractable Rare Dis Res. 2018;7:223-8.
2. Lippe B, Geffner ME, Dietrich RB, Boechat MI, Kangarloo H. Renal malformations in patients with turner syndrome: imaging in 141 patients. Pediatrics. 1988;82:852-6.
3. Kawanishi C, Kono M, Onishi H, Ishii N, Ishii K. A case of turner syndrome with schizophrenia: genetic relationship between turner syndrome and psychosis. Psychiatry Clin Neurosci. 1997;51:83-5.
4. Gahlawat S, Goel H, Khattar N. Management of female congenital short patulous urethra with urethral tapering and pubovaginal sling: a report of two cases. Indian J Urol. 2018;34:149-51.
5. Altalib A, AlSulmi E, Bokhari D, Alhalal Z, 2nd, Alismail M, Alzayyat R. Mosaic turner syndrome with multiple spontaneous pregnancies: a case report. Cureus. 2024;16:e53351.
6. Izumita Y, Nishigaki S, Satoh M, Takubo N, Numakura C, Takahashi I, et al. Retrospective study of the renal function using estimated glomerular filtration rate and congenital anomalies of the kidney-urinary tract in pediatric Turner syndrome. Congenit Anom (Kyoto). 2020;60:175-9.
7. Westland R, Renkema KY, Knoers NV. Clinical integration of genome diagnostics for congenital anomalies of the kidney and urinary tract. Clin J Am Soc Nephrol. 2020;16:128-37.
8. Rajamaheswari N, Agarwal S, Chhikara AB, Seethalakshmi K. Anterior bladder flap neo urethra as treatment for stress urinary incontinence due to developmental urogenital anomaly. Urol Ann. 2013;5:215-7.
9. Epidemiology of bladder exstrophy and epispadias: a communication from the international clearinghouse for birth defects monitoring systems. Teratology. 1987;36:221-7.
10. Gambhir L, Höller T, Müller M, Schott G, Vogt H, Detlefsen B, et al. Epidemiological survey of 214 families with bladder exstrophy-epispadias complex. J Urol. 2008;179:1539-43.
11. Ebert AK, Reutter H, Ludwig M, Rösch WH. The exstrophy-epispadias complex. Orphanet J Rare Dis. 2009;4:23.
12. Frimberger D. Diagnosis and management of epispadias. Semin Pediatr Surg. 2011;20:85-90.
13. Manjavacas PO, Canet FD, Benítez CA, Jaime JC. Urinary incontinence in women: evaluation and practical management in primary care. FMC. 2020;27:480-7.
14. Serrano-Brambila EA, Maldonado-Alcaraz E, Espinoza-Guerrero XA, Moreno-Palacios J, Ixquiac-Pineda GA, Cárdenas-Rodríguez E. Cabestrillo subtrigonal con aponeurosis abdominal en el tratamiento de incontinencia urinaria de esfuerzo compleja [Subtrigonal sling with abdominal fascia for treatment of complex stress urinary incontinence]. Cir Cir. 2009;77:193-200.
15. Guerid S, Boucher F, Delay E. Genital aesthetic surgery. EMC Reconstr Aesthet Plast Surg. 2016;24:1-8.