# **CASE REPORT**



# Relationship between Turner Syndrome and Congenital Malformations of the Kidney and Urinary Tract

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#### ABSTRACT

We present the clinical case of a 20-year-old woman with horseshoe kidney who attends periodic check-ups at the Urology department. He had a history of Turner Syndrome and numerous pathologies associated with the genetic disease. Malformations of the urinary tract, characteristics of Turner syndrome and the association between both are reviewed. Finally, some advices are given for the best control and follow-up of this patient.

Key words: Turner syndrome, kidney malformations, MAG3 renogram scan, horseshoe kidney.

## **CASE REPORT**

2 <sup>0</sup>-year-old female patient with no allergies is presented. She had Turner syndrome (45X0), moderate mitral regurgitation due to myxomatous mitral valve, hirsutism, peripheral vertigo, low to moderate intellectual disability, dysembryoplastic neuroepithelial tumor (DNET) in the left inferior frontal gyrus, horseshoe kidney, nystagmus and eyelid ptosis surgical removed and left pyeloplasty for ureteropelvic junction stenosis at 9 months of age. She follows chronic treatment with progesterone 100 mg every 24 hours, finasteride 5 mg every 24 hours, estradiol 75 mcg in transdermal patches every 72 hours, cholecalciferol 25,000 IU/2.5 ml every month.

The patient undergoes regular annual check-ups at Urology department. At the last visit she was asymptomatic, with no recurrent urinary infections, and with normal kidney function. The results of the most recent complementary explorations are described below.

Cardiac doppler ultrasound: non-dilated left ventricle, ejection fraction of 65%, normally functioning three-leaflet aortic valve, mitral valve with thickened and myxomatous leaflets with moderate-severe insufficiency at the expense of 3 jets. Moderate-severe mitral and tricuspide valve regurgitation, and moderately dilated left atrium.

Abdominal and pelvic CT (figures1-7): right kidney malrotated and descended in the right lower quadrant, with a parenchymal bridge between its lower pole and the lower pole of the contralateral kidney. Complete double urinary excretory system of the right urinary tract. Left kidney also malrotated and descended, with dilation of the renal calyces suggestive of left congenital megakaliosis. Unique left urinary excretory system. Bicornuate uterus.

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Figure 1. Horseshoe kidney fused in the caudal area



Figures 2, 3. Left megakaliosis

![](_page_1_Picture_5.jpeg)

Figures 4, 5. Double left urinary excretory system

![](_page_2_Picture_1.jpeg)

Figure 6. Ectopic and rotated pelvic kidneys

MAG3 renogram scan (figures 8-10): horseshoe kidney pattern with lower viability of the parenchyma in the lower isthmus and bilateral pyelocalicillary ectasia especially in the left upper calicillary group, of a functional, non-obstructive.

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![](_page_2_Picture_5.jpeg)

Figure 7. Bicornuate uterus

The response to the administration of the diuretic was suitable. Contribution to global renal function was 52% in the right hemi-kidney and 48% in the left.

![](_page_2_Figure_8.jpeg)

![](_page_2_Figure_9.jpeg)

![](_page_2_Figure_10.jpeg)

Figure 10. contribution to global renal function was 52% in the right hemi-kidney and 48% in the left

Brain MRI (figures 11-13): presence of neuroepithelial dysembryoplastic tumor with small microcysts, located in the gyrus of the left lower frontal area, without contrast enhancement or edema, without changes in morphology and size, similar to previous MRI 2 years before. DNET is a low-grade, slow-growing brain tumor and contains properties of both glial and neuronal cells. It is a surgically curable neuroepithelial neoplasia. The prognosis is generally good with 80% of 5-year survival rate.

![](_page_3_Picture_3.jpeg)

Figure 11,12. Microcysts, located in the gyrus of the left lower frontal area.

![](_page_3_Picture_5.jpeg)

Figure 13. Neuroepithelial dysembryoplastic tumor

The patient was asymptomatic and without changes in the complementary urological tests, so it was decided to perform annual control with blood test, urine culture and renal ultrasound.

### DISCUSSION

Congenital malformations of the kidney and urinary tract represent 20-30% of all disorders identified in prenatal control, appear in 3-6/1,000 live births and are responsible for 34-59% of chronic kidney disease and 31% of cases of end-stage

kidney disease. Disorders in the development of the urinary system can affect the renal parenchyma (dysplasia, hypoplasia and renal agenesis, tubular dysplasia and polycystosis), the embryological migration of the kidney (renal ectopia, fusion defects and horseshoe kidney) or the collecting system (ureteropelvic junction stenosis, megaureter, ectopic ureter, vesico-ureteral reflux, bladder exstrophy and posterior urethral valves)(1,2).

Renal ectopia has an incidence of 1 every 1,000 autopsies, and occurs when kidney does not ascend from the pelvis

towards the renal fossa. Usually have poor position and lack of rotation from anterior to medial, therefore placing the renal pelvis more anterior. If it crosses the midline, it is called crossed renal ectopia, and can occur with or without fusion to the contralateral kidney. Arterial vascularization of the ectopic kidney is variable and can arrive from iliac artery, the aorta and sometimes from hypogastric artery or sacral arteries. The ectopic kidney usually has a decreased function, and can be associated with vesico-ureteral reflux and malformations of the genital organs and the skeleton(3-5).

Renal fusion abnormalities occur when a portion of one kidney fuses with the contralateral kidney. The most common fusion is the horseshoe kidney, with an incidence between 1/400 and 1/800 live newborns, so that the two kidneys are joined by an isthmus of parenchyma or fibrosis that crosses the midline. In 90% of cases they are joined at the lower pole and usually this union is located in the lower part of the abdomen, in front of the great vessels and below the union of the inferior mesenteric artery and the aorta. Fusion occurs before the kidneys ascend from the pelvis to their normal position in the renal fossa, usually before the fifth week of gestation. This fusion implies a lack of rotation, so the renal pelvises are located anteriorly. It is frequently associated with vesico-ureteral reflux and the presence of hydronephrosis, present in 26-32% and 8-15%, respectively. Hydronephrosis can become obstructive in 30% of cases. The vascular supply of the isthmus is provided by an isolated vessel and may come from the aorta, the common iliac artery, or the inferior mesenteric artery. A polar vessel that initially does not cause obstruction can, with the growth of the kidney, become difficult to pass through the pyeloureteral junction and behave obstructively, compromising the renal function(6-9).

In crossed renal ectopia the ectopic kidney crosses the midline and fuses with the contralateral kidney, but the ureter of the ectopic kidney maintains its insertion into the bladder. Its incidence is estimated at 1/2,000 births. In most cases, the ectopic kidney fuses to the lower pole of the contralateral kidney, which can be placed in its normal thoracolumbar or pelvic position. Its diagnosis is usually made with ultrasound, but sometimes other imaging tests such as CT-scan or uromagnetic resonance are required to understand the anatomy, mainly if the patient needs to undergo surgery(4).

Turner syndrome is a genetic disease that appears in 50-100 out of every 100,000 women, and is characterized by a set of phenotypic traits secondary to the partial or complete absence of the second X chromosome (45X0), being the most frequent cause of consultation, the short stature.

These patients are more predisposed to present diabetes mellitus, hypothyroidism, autoimmune thyroiditis, dyslipidemia, osteoporosis, strabismus, eyelid ptosis, palpabral cleft, disorders of visual memory and motor coordination,

cognitive deficit, significantly subaverage intellectual functioning, sexual infantilism, infertility, delayed menarche and puberty, amenorrhea, hypoplastic or inverted nipples, short neck or pterygium colli, disproportion between thorax and legs, hyperpigmented nevi, atopic dermatitis and keloids, hypoacusis, otitis media, cholesteatoma, epilepsy, psychotic disorders, micrognathia, high-arched palate, cubitus valgus and hypoplastic hands (Madelung deformity), convex nails, sleep apnea, obesity, respiratory failure, stroke, hypertension, dilated cardiomyopathy, atrial septal defect, aortic stenosis and dissection, tumors of the central nervous system, melanoma, bladder and endometrial tumor(10-12).Kidney malformations appear in Turner syndrome in 30-50% of cases. The most common is horseshoe kidney (10%), followed by duplication of collecting systems (15%), hydronephrosis (5-15%), renal aplasia (3%) and ectopic or multicystic kidney (<1%). They are between 2 and 3 times more likely to present bladder tumors than the general population. Recurrent urinary infections and vesicoureteral reflux are also described. For this reason, a renal ultrasound must always be requested in the initial diagnosis of Turner syndrome(13).

## CONCLUSION

The patient in this clinical case with Turner syndrome presents urinary malformations described as horseshoe kidney, duplication of collecting systems and megakaliosis. In addition, she underwent surgery for vesico-ureteral reflux in childhood. Due to this history, if the patient is asymptomatic, annual follow-up with blood tests, urine culture and ultrasound or abdominal CT-scan with contrast are recommended. It is important to remember that the management of these patients must be multidisciplinary due to the different organs that are affected in Turner syndrome.

80% of patients with Turner syndrome have a medium-high level of education with good integration into society, and can reach a life expectancy of 10 years less than the general population. Moreover, in adulthood the medical checks are irregular and scarce. For these reasons, we must insist on regular visits to the doctor, especially at endocrinology, cardiovascular and gynecology, and to the urologist if they present kidney malformations.

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