

Ochoa Syndrome—A Twins Case Report

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Abstract

Urofacial syndrome, known as Ochoa syndrome, is a very rare autosomal recessive disorder described as a clinical setting in which there is an association of a lower urinary tract and bowel dysfunction with a typical facial expression: the patient seems to be grimacing or crying when attempting to smile. If the diagnosis and treatment are delayed, the patients might have a poor outcome with continuous upper urinary tract deterioration, which makes the early diagnosis of this condition of vital importance. We report a case of two females identical twins, who are, nowadays, 19 years old, which had the Ochoa Syndrome diagnosis made when they were 10 years old, but discontinued medical follow up during treatment. They have developed chronic renal failure with hemodialysis need and are in a waiting list for kidney transplantation. In this rare disorder, even more unusual in twins and with high risks on progressing with renal failure, a simple attempt to smile may save lives if the condition is known by doctors such as pediatricians and pediatric urologists. Conclusion: We believe that unknowing this condition is the main explanation for this fact. Early diagnosis and close follow up are the key points to a successful outcome.

Keywords

Urofacial Syndrome, Ochoa Syndrome, Urinary Bladder, Neurogenic

1. Introduction

In the early 1960s, the Colombian physician Bernardo Ochoa followed a group of patients with lower urinary tract dysfunction and an inversion of facial expression, where the patient seems to be crying when attempting to laugh [1] [2]. This clinical association is known as the Urofacial Syndrome (UFS), or, in regard to its first de-

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scriber, as the Ochoa Syndrome. More than 100 cases have been reported in the last 50 years all around the globe [3], but none from Brazil. The UFS is a rare autosomal recessive disorder that occurs equally in both sexes but seems to be more frequent when parents are consanguineously related, and a mutation in the Heparanase gene (*HPSE2*), located on chromosome 10q24 is the cause of this disease [4]. The urinary symptomatology is variable and may consist of the recurrent urinary tract infection presenting with urgency and dysuria [5], incontinence, enuresis, constipation, bladder trabeculation and even renal failure symptoms [6]. The emotional (laughing and crying) center is close to the micturition center, reason why it has been speculated that this is a possible pathophysiological explanation for the association of the urinary tract dysfunction and typical facial expression.

Early diagnosis and treatment are essential to increase the chances of having a healthy relative normal life [7]. We report here a case of twins, which is even more unusual, who had their first symptoms after turning 4 years old and the UFS diagnosed when they were 10 years old. They lost follow up in regard to social conditions and underwent into chronic renal failure, what makes very clear how severe might the outcome of this condition be, and the importance of an early diagnosis and a regular follow up.

2. Case Report

Two young twin girls, “A” and “B”, from Ribeirao-Preto—São Paulo-Brazil, who were born at term following a normal vaginal delivery, adopted right after born, had been brought to our pediatric urology service by their adoption mother. No information on the biological parents was known. Development was normal in the first 4 years of life. The adoption mother told our crew that she had noticed, after the children turned 4 years old, that both of them were developing a lot of abdominal effort to urinate, had enuresis and urinary urgency.

Twin “A”: When she was 4 years and 10 months old, this girl had an episode of urinary retention, bilateral eyelid edema and a convulsive crisis. A vesical catheterism was needed and acute renal failure was diagnosed. At that point, she underwent a vesicostomy in another medical center. She was brought to our pediatric urology service for the first time, 4 days after the vesicostomy was performed, along with her twin sister. On our physical exam it was easy to notice that both girls had a typical facial expression, seeming to be crying when attempting to smile. Having the urofacial syndrome in mind, this diagnose was immediately considered. Renal ecography showed moderate bilateral hidronefrosis and bilateral renal rotation abnormality. Voiding cystography showed a trabeculated bladder and right vesico-uretral reflux. Lumbar x-ray was normal and no neurological symptom was noticed. On a close follow up the creatinine and urea levels went back to normal. Two years later, after having a close medical follow up on our pediatric urology service, the child underwent a new surgery to close the vesicostomy. An urodynamic study was performed showing a low-compliance small bladder and suggesting a lack of coordination between detrusor muscle contraction and urethral sphincter relaxation with significant residual urine after voiding. Adding the urodynamic findings of a non-neurogenic neurogenic bladder to the typical facial expression, the Ochoa Syndrome diagnosis was confirmed. She was treated with antibiotic prophylaxis and intermittent catheterism.

Twin “B”: This girl was brought to us along with her sister with a similar history of recurrent urinary tract infection, low urinary volume, night enuresis, constipation and already had creatinine levels alterations. Urinary tract ultrasound revealed an enlarged bladder, with bilateral ureteral dilatation and moderate bilateral hidronefrosis. Voiding cystourethrography revealed a trabeculated bladder but didn’t show signs of vesico-ureteral reflux. As she was unable to do clean intermittent catheterization a vesicostomy was performed, which was closed 2 years later. Urodynamic study showed a low-compliance small bladder and non-inhibited detrusor contraction with a 66 ml urine volume and 131.4 cm/H₂O pressure, also confirming the UFS diagnose. She was then treated with anticholinergic drug and intermittent vesical catheterism.

Both girls moved to a different town with their family, losing medical follow up and underwent chronicle renal failure. At the age of 19 years old (**Figure 1**), they moved back to our hospital and the contact with our crew was reestablished. They were reevaluated and urodynamic study and voiding cystourethrography were updated. Twin A: Bladder capacity and sensitivity were preserved with reduced compliance. No detrusor contraction (underactive bladder) and no effort to accomplish voiding were showed. Voiding cystourethrography: bladder with reduced dimensions and with pseudo-diverticulum and absence of vesico-ureteral reflux. Twin B: Urodynamics showed a decreased bladder capacity and borderline compliance, with no spontaneous urination. Maximum voiding detrusor pressure of 100 cm/H₂O suggesting vesico-sphincter dyssnergism. Patient had no urination during examination (anuric for 2 years). Voiding cystourethrography: bladder with low capacity and



Figure 1. Twin sister with Ochoa syndrome at the age of 19. Notice the grimacing expression when attempting to smile.

absence of vesicoureteral reflux. Both girls are on a hemodialysis program and are registered on a kidney transplant waiting list.

3. Conclusion

In conclusion, the Ochoa syndrome is a rare uro-genetic condition. Although unusual, this syndrome is a well-described entity that if not early diagnosed and adequately treated, may lead to significant morbidity and even mortality from lower and upper urinary tract deterioration. Colombia was the first country in the world where the cases were reported, but the Ochoa Syndrome has not been officially reported in any other South Americans countries except Argentina [8], which makes us to believe that there still are many patients with severe voiding dysfunction that haven't been properly diagnosed with the UFS. Spreading the knowledge about the clinical set of this syndrome, where you can diagnose a neurogenic bladder from a smile, emphasizing that bad outcomes might happen if it's not properly treated, and sharing our experiences with other urologists and pediatricians are the first step to accomplish this goal.

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Conflict of Interest

The authors declare that they have no conflicts of interest.

References

- [1] Akl, K.F. and Momany, H.M.A. (2012) Urofacial Syndrome. *Saudi Journal of Kidney Diseases and Transplantation*, **23**, 346-348.
- [2] Ochoa, B. and Gorlin, R.J. (1987) Urofacial (Ochoa) Syndrome. *American Journal of Medical Genetics*, **27**, 661-667. <http://dx.doi.org/10.1002/ajmg.1320270320>
- [3] Ochoa, B. (2004) Can a Congenital Dysfunctional Bladder Be Diagnosed from a Smile? The Ochoa Syndrome Update. *Pediatric Nephrology*, **19**, 6-12.
- [4] Wang, C., Davoodi-Semironi, A., Shi, J., Yang, P., Huang, Y., Agundez, J.A.G., Moran, J.M., Ochoa, B., Hawkins-Lee, B. and She, J. (2003) High Resolution Mapping and Mutation Analyses of Candidate Genes in the Urofacial Syndrome (UFS) Critical Region. *American Journal of Medical Genetics Part A*, **119A**, 9-14. <http://dx.doi.org/10.1002/ajmg.a.20042>

- [5] Stamatiou, K., Tyritzis, S., Karakos, C. and Skolarikos, A. (2011) Urofacial Syndrome: A Subset of Neurogenic Bladder Dysfunction Syndromes? *Urology*, **78**, 911-914. <http://dx.doi.org/10.1016/j.urology.2010.12.061>
- [6] Bacchetta, J. and Cochat, P. (2010) Severe Voiding Dysfunction: Ask the Child to Smile. *Kidney International*, **78**, 225-226. <http://dx.doi.org/10.1038/ki.2010.144>
- [7] Nicanor, F.A., Cook, A. and Pippi-Salle, J.L. (2005) Early Diagnosis of the Urofacial Syndrome Is Essential to Prevent Irreversible Renal Failure. *International Brazilian Journal of Urology*, **31**, 477-481. <http://dx.doi.org/10.1590/S1677-55382005000500012>
- [8] Bertolotti, A.F., González, S.G. and Etcheverry, R.M. (2007) Ochoa's Syndrome in Argentina. *Cirugía Pediátrica*, **20**, 54-56.

Abbreviation

UFS: Urofacial Syndrome