Residência RP Pediátrica
Publicação Oficial da Sociedade Brasileira de Pediatria
ISSN-Online: 2236-6814

Submitted on: 12/26/2018
Approved on: 03/07/2019

CASE REPORT

Complications of renal transplantation in patients with Ochoa urofacial syndrome: case report

Bruna Weis Jovino[1], Ylara Liza Porto de Carvalho[1], Victor Batitucci[1], Cássia Caroline Emilio[1], Stela Maria Tavolieri de Oliveira[1]

Keywords:
Cystitis,
Kidney Failure,
Chronic,
Transplants.

Abstract

Ochoa urofacial syndrome (UFS) is a rare condition with an autosomal recessive inheritance, characterized by the presence of functional obstructive uropathy associated with the inversion of facial expression when attempting to smile, similar to crying facies. From an eminently clinical diagnosis, facial expression abnormality is the main characteristic of UFS and its relation with voiding dysfunction is still controversial. Patients with UFS, who present with recurrent urinary tract infection, voiding dysfunction and vesicoureteral reflux, if not diagnosed early, may present progressive deterioration of the upper urinary tract with evolution to end-stage renal failure. The authors report a case of patients with UFS, kidney transplant, poor adherence to the treatment that evolved with severe urinary tract infection, stage 2 arterial hypertension and renal graft loss.

[1] University Hospital of the School of Medicine of Jundiaí, Pediatrics Department - Jundiaí - São Paulo - Brazil.

Correspondence to:
Bruna Weis Jovino.
Hospital Universitário da Faculdade de Medicina de Jundiaí. Praça Rotatória, s/n - Jardim Messina, Jundiaí - SP, Brazil. CEP: 13207-450. E-mail: bruna_js_@hotmail.com

INTRODUCTION

Ochoa syndrome (OS) or urofacial syndrome was described for the first time by Rafael Elejalde in 1979.[1] Patients with Ochoa syndrome (OS), a rare autosomal recessive congenital disorder, present with functional obstructive uropathy associated with an inversion of facial expressions when they attempt to smile, at which time they appear to be crying)[2](Figure 1). In 1987, Ochoa and Gorlin described 36 cases of children with enuresis and recurrent urinary tract infection (UTI) exhibiting inverted smiles.[3]

None of the patients had neurologic disorders or mechanical blocks to the outflow of urine that might explain the urinary findings.[4] OS equally affects individuals of both sexes and is more frequently seen in children of consanguineous parents.[3] Recent studies mapped the disorder to chromosome 10 q23-24 and described the production of defective Heparanase 2 protein.[1] Abnormal facial expressions are the hallmark of OS, and its association with voiding dysfunction is still controversial.[1] The most widely accepted explanation indicates that the laughing and crying centers lie close to the centers for micturition in the upper pons of the midbrain and the site from which the seventh cranial nerve (facial nerve) originates. Injuries to this area may produce the two clinical manifestations.[4]

Diagnosis of the condition is eminently clinical. If not diagnosed in the early stages of the condition, patients with OS presenting with recurrent UTI, voiding dysfunction, and vesicoureteral reflux may develop progressive deterioration of the upper urinary tract and ultimately progress to end-stage renal disease.[5]



Figure 1. Patient presenting an inverted smile.

CASE REPORT

The patient described in this case is a 15-year-old young man of Roma ethnicity born in Sorocaba, SP, diagnosed with Ochoa syndrome with an augmented neurogenic bladder and a continent urinary diversion (Mitrofanoff) on clean intermittent catheterization (CIC). He had received a kidney from a deceased donor at Hospital Pequeno Príncipe four years prior to this report and lost the graft two years after transplantation for poor compliance to prescribed therapy. He was prescribed renal replacement therapy and had been on hemodialysis for a year.

The patient was admitted to the pediatric emergency unit complaining of abdominal pain in the hypogastric region. He had fever and had been vomiting for three days, and a purulent effusion was oozing through his shunt during catheterization. The patient was constipated and hypertensive.

Physical examination revealed he was dehydrated and had stage 2 high blood pressure (140X100mmHg). His abdomen was distended with low abdominal sounds and was painful to palpation in the right hypochondrium and hypogastric region. Workup results were as follows: Hg: 10.1 g/dl; Ht: 33.4 g/dl; white blood cells: 6.47 mil/mm3; blood urea nitrogen: 215.2 mg/dl; creatinine: 11.6 mg/dl; K: 5.9mmol/L; urine sediment examination revealed leukocyturia with > 1 million WBC/ml; Red blood cells: 400,000/ml. Abdomen computed tomography (CT) scans showed moderate distension of the small bowel loops without signs of obstruction. Two-dimensional echocardiography with Doppler showed mild pulmonary hypertension and concentric left ventricular hypertrophy. The patient was suspected for dialysis-dependent chronic kidney disease (CKD) caused by kidney graft loss and inflammatory acute abdomen associated with febrile UTI. The patient was prescribed hemodialysis, had an indwelling urinary catheter and an open nasogastric tube implanted, and took antibiotic therapy with ceftriaxone before he improved from infection and resumed oral feeding. Urine culture results showed the patient had ceftriaxone-resistant Klebsiella pneumoniae > 100,000 CFU/ml sensitive to amikacin; his antibiotic

therapy was changed and doses were adjusted to meet the requirements of individuals with stage 2 CKD. The patient was discharged after spending 12 days in hospital and was asked to continue treatment with antibiotics at home, and return for additional hemodialysis sessions. He was also referred to the renal transplantation service. Compliance to treatment has been less than ideal since.

DISCUSSION

Diagnosing individuals with early-stage OS is relevant, since voiding dysfunction may progress with upper urinary tract injury and result in hypertension and chronic kidney disease in the long run.[6] Late diagnosis and non-compliance to treatment contribute to the onset of CKD, the most severe complication of OS.[5]

Patients submitted to kidney transplants may experience surgical and clinical complications, in addition to developing infection. Surgical complications include vascular adverse events such as renal artery and renal vein thrombosis, lymphocele, and renal artery stenosis; and urinary events such as urinary fistulae, bladder fistulae, and urinary obstruction.[7] Infections include UTI, surgical site infection, respiratory tract infection, and bloodstream infection.[7] A publication from the North American Pediatric Renal Trials and Collaborative Studies (NAPRTCS) indicated that 3,066 kidney transplants were performed in 1995. Reported graft survival rates after three years were 83% in living-donor kidney transplants and 66% in deceased-donor kidney transplants.[7] Our patient underwent a kidney transplant and lost the graft two years after the procedure.

UTI is the most common infection to affect kidney transplant patients and accounts for 23-75% of all cases of healthcare-associated infection. More than 50% of UTI episodes occur during the first year after transplantation, with females and recipients of deceased-donor kidney transplants being preferentially affected (55-84%). The main pathogens involved are Klebsiella sp., Enterobacter sp., Enterococcus sp., E. coli, P. aeruginosa, and Candida sp.[8] Our patient had a positive urine culture for Klebsiella pneumoniae.

The patient described in this report was on clean intermittent catheterization to drain residual urine, a risk factor for recurrent UTI. He was admitted with purulent effusion seen during catheterization with a Mitrofanoff. Risk factors for kidney graft loss included the patient's Roma etiology, lack of fixed residence, low socioeconomic status, and the facts that he did not go to school and had an illiterate mother.

Given the severity of OS, pediatricians must be aware of the connections between voiding symptoms and presenting with an inverted smile, so as to prevent the complications arising from the disease.

REFERENCES

1. Bertolotti AF, González SGT, Etcheverry RM. Síndrome de Ochoa en Argentina. Cir Pediatr [Internet]. 2007; [citado 2018 out 19]; 20(1):54-6. Disponível em: https://studylib.es/doc/4692995/caso-cl%C3%ADnico---s%C3%ADndrome-de-ochoa-en-argentina
2. Guo C, Kaneko S, Sun Y, Huang Y, Vlodavsky I, Li X, et al. A mouse model of urofacial syndrome with dysfunctional urination. Hum Mol Genet [Internet]. 2015 Abr; [citado 2018 out 19]; 24(7):1991-9. Disponível em: https://www.ncbi.nlm.nih.gov/pubmed/25510506
3. Woolf AS, Stuart HM, Roberts NA, McKenzie EA, Hilton EN, Newman WG. Urofacial syndrome: a genetic and congenital disease of aberrant urinary bladder innervation. Pediatr Nephrol [Internet]. 2014 Abr; [citado 2018 out 19]; 29(4):513-8. Disponível em: https://www.ncbi.nlm.nih.gov/pubmed/23832138
4. Aydogdu O, Burgu B, Demirel F, Soygur T, Ozcakar ZB, Yalcinkaya F, et al. Ochoa syndrome: a spectrum of urofacial syndrome. Eur J Pediatr [Internet]. 2010 Abr; [citado 2018 out 19]; 169(4):431-5. Disponível em: https://www.ncbi.nlm.nih.gov/pubmed/19669792
5. Rondon AV, Leslie B, Bastos Netto JM, Freitas RG, Ortiz V, Macedo Junior A. et al. Síndrome urofacial de Ochoa: reconheça o sorriso peculiar e evite complicações urológicas e renais graves. Einstein [Internet]. 2013 Mai; [citado 2015 mai 1]; 13(2):279-82. Disponível em: http://www.scielo.br/scielo.php?pid=S1679-45082015005052990&script=sci_arttext&tlng=pt
6. Teebi AS, Farag TI, el-Khalifa MY, Besisso MS, al-Ansari AG. Urofacial syndrome. Am J Med Genet [Internet]. 1989 Dez; [citado 2018 out 19]; 34(4):608. Disponível em: https://www.ncbi.nlm.nih.gov/pubmed/2624278
7. Lima MR. Prevalência de complicações infecciosas em pacientes pediátricos submetidos a transplante renal no hospital irmandade da Santa Casa de Misericórdia de São Paulo [monografia]. São Paulo: Hospital Irmandade da Santa Casa de Misericórdia de São Paulo – Nefrologia Pediátrica; 2016.
8. Ochoa B, Gorlin RJ, Opitz JM, Reynolds JF. Urofacial (Ochoa) syndrome. Am J Med Genet [Internet]. 1987 Jul; [citado 2018 out 19]; 27(3):661-7. Disponível em: www.https://onlinelibrary.wiley.com/doi/abs/10.1002/ajmg.1320270320