Case Report

Urofacial syndrome: A subset of neurogenic bladder dysfunction syndromes?

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ABSTRACT

The urofacial syndrome is probably a subset of neurogenic bladder dysfunction syndromes characterized by detrusor-sphincter discoordination along with a characteristic inversion of facial expression with laughing. This characteristic facial expression can facilitate early detection of this disorder, which leads to poor bladder emptying with high residual urine, hydro-nephrosis with vesico-ureteral reflux and potentially renal failure if left untreated. The etiology of the urofacial syndrome is unknown. In our case, a 12-year-old boy of Middle-Eastern origin presented to the Outpatient Department of our hospital with left pyelonephritis, hydronephrosis and bladder dilatation. Voiding cystourethrography performed 15 days later revealed left vesicoureteral reflux. Cystoscopy revealed bladder trabeculation however an anatomic urethral obstruction was not noticed. Both, neurological examination and radiography of the lumbosacral spine were normal. Urodynamic evaluation revealed the typical findings of detrusor-sphincter discoordination.

Key words: Neurogenic bladder dysfunction, enuresis, recurrent urinary tract infections, residual urine

INTRODUCTION

In 1987, Ochoa and Gorlin [1] suggested the term urofacial syndrome (UFS) to describe a syndrome characterized by a non-neurogenic bladder dysfunction and a characteristic facial feature which consists on the inversion of facial expression with laughing. Less than 50 cases have been reported since. According to the existent literature, the first signs of the disorder are the recurrent urinary tract infections, nocturnal enuresis and constipation while in most of the cases patients have severe obstructive symptoms and significant bladder dysfunction at the time of diagnosis.[2] Although rare, this disorder has to be considered in the differential diagnosis of enuresis in children with recurrent urinary tract infections and residual urine. Our aim is to further document the existence of this syndrome and to increase awareness among practicing pediatricians of the classical facial characteristics that facilitate diagnosis.

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CASE PRESENTATION

A 12-year-old boy of Middle-Eastern origin presented to outpatient department of our hospital with left flank pain, dysouria, fever (up to 38.5°C), persistent vomiting (hyperemesis) and severe dehydration for four days. On physical examination, he presented with tenderness on left pleuro-vertebral angle and a palpable bladder. A characteristic inversion of facial expression with smiling was also noticed [Figure 1].

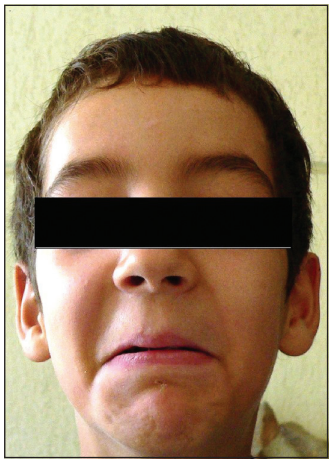

Figure 1: Inversion of facial expression with smiling

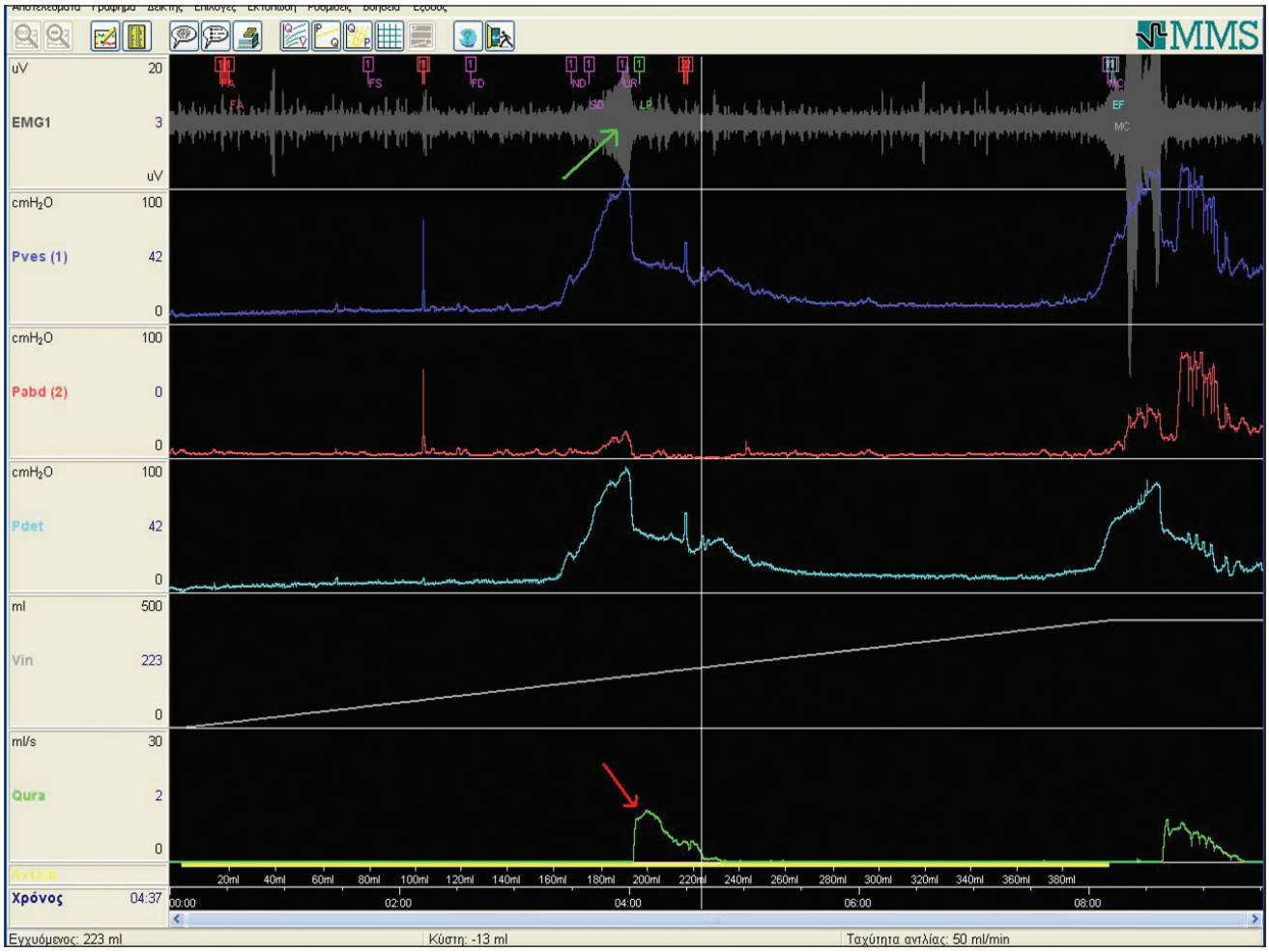

Figure 2: Detrusor-sphincter discoordination

The patient complained of urgency and with associated urge incontinence which has been initially considered as symptoms of acute pyelonephritis. Upon admittance, his white blood cell (WBC) count was elevated (14.5×10^9/L, granulocytes 85%), electrolytes were diminished (Na: 135, K: 3.1), while red blood cell count and hemoglobin concentration were normal (Hct: 37.5%, Hb: 12.5mg/dL, RBC: 4.26M/micoliter). The serum creatinine was normal. A history of constipation, enuresis and recurrent urinary tract infections was present.

His treatment included intravenous amoxicillin-clavulanic acid, intravenous fluid administration and paracetamol oral for pain relief. On the following four days his fever persisted at above 37°C, with spikes up to 38.5°C during evening hours. Voiding symptoms also persisted. Abdominal ultrasound on the fourth day of hospitalization confirmed a dilatation of renal pelvis and initial ureter on left side and a slight thickness of bladder wall. The terminal ureters were visible on both sides. Post urinating ultrasound examination of the bladder revealed a residual urine volume of up to 140 cc. Improvement was noticed in both his clinical picture and his lab results two days after the placement of a 14 Ch Foley catheter. Voiding cystourethrography performed 15 days later showed a narrowing of the urethra at the level of the external sphincter and left vesicoureteral reflux. Cystoscopy revealed bladder trabeculation, however, an anatomic urethral obstruction was not noticed. Both the neurological examination and radiography of the lumbosacral spine were normal. Urodynamic evaluation (pressure-flow study) revealed the typical findings of detrusor-sphincter discoordination [Figure 2]. More precisely, the filling phase (thick yellow line) revealed both phasic and final type detrusor overactivity and urine leakage (red arrow). The flow phase was characterized by increased detrusor pressure and low flow (pdet.max 89 cm H2O, pdetQmax 67 cm H_2O, and maximum flow (Q_{max}) value 7 ml/sec). EMG activity of the pelvic floor was significantly increased (green arrow). Although bladder augmentation and intermitted catheterization were suggested, patient's parents refused any intervention and the boy was discharged home.

DISCUSSION AND CONCLUSION

UFS is a rare disease characterized by detrusor-sphincter

discoordination (with the characteristic symptomatology consisting of recurrent urinary infections, incontinence, constipation, dysuria, frequency, and enuresis) in children and young adolescents with a characteristic facial expression consisting of inversion of facial expression with laughing. Patients have no apparent neurological or obstructive abnormality therefore; the syndrome may in fact represent a subgroup of the non-neurogenic bladder dysfunction.[2] According to other investigators, UFS represents a distinct entity. Since the laughing and crying centers are located in upper pons of the midbrain lying in close proximity to the micturition center, it has been speculated that a subtle neurologic lesion may simultaneously affect both regions therefore resulting in this clinical association.[1] Recent research, however, suggests that UFS probably represent a genetic inherited disease transmitted in an autosomal recessive fashion.[3]

Patients with UFS may develop urinary tract pathology having a wide spectrum of severity. Mild cases may simply present with the characteristic facial findings and mild or at times no urinary tract issues. Severe bladder dysfunction with a significant functional obstruction may in other cases lead to poor bladder emptying with high residual urine, hydroureteronephrosis with vesicoureteral reflux and potentially renal failure. Differential diagnosis may be difficult, especially in patients presenting with only symptoms suggesting infection (urgency, frequency and urgency incontinence) or elimination dysfunction (constipation and encopresis).[4] While other non-neurogenic bladder dysfunction conditions such as "lazy bladder syndrome" and "urge syndrome" are more prevalent in females, UFS equally affects both sexes.[1,5] The characteristic facial expression can facilitate early detection of this disorder. The early institution of prophylactic treatment in these cases may prevent the subsequent development of urinary tract deterioration. Failure to recognize patients with this disorder may lead to an unnecessarily complicated clinical course.[2] In addition, the positive family history for obstructive uropathy should have also raised suspicion for a possible hereditary form of bladder dysfunction. The basic therapeutic goals for cases detected early are the restoration of balanced bladder emptying and the prevention of upper urinary tract deterioration.

Biofeedback is the mainstay of treatment in such patients. Drug therapy is usually guided by urodynamic findings. Urinary diversion or bladder augmentation may provide the only alternative to prevent an ongoing deterioration of renal function.

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