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Early elimination dysfunction associated with cephalic anomalies: Is there a link?

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Abstract Elimination dysfunction in children can be related to three main aetiologies: 1) spinal cord anomalies, 2) social and environmental disorders, and 3) syndromic elimination disorders. From this last group, we report cases of a previously undescribed combination of elimination disorders and cephalic anomalies symptoms which may constitute a proper entity for which conventional treatments may fail. A comprehensive review of congenital elimination disorders is given.

Patients and methods: Six patients (four boys, two girls) presenting with early elimination dysfunction associated with cephalic anomalies were assessed and treated between 1994 and 2005. None presented with identified lower urinary tract obstruction or spinal cord anomalies. Follow up ranged between 5.5 and 11.5 (mean 6.7) years.

Results: All six had early elimination disorders, represented by urine retention, urinary tract infections, constipation and soiling. All had facial dysmorphy and cerebral anomalies with developmental delay of varying severity. All had a dilated urinary tract, with severe vesicoureteral reflux in five and one megaureter without reflux. All had abnormal renal isotope scans, two associated with chronic renal failure. The family medical history was significant in some cases. Treatment included early urinary diversion, and there was a high failure rate for ureteral reimplantation.

Conclusion: The combination of congenital elimination dysfunction with facial anomalies, developmental retardation, cephalic anomalies, abnormal urinary tracts, without identified spinal cord disorders or lower urinary tract obstruction, may represent a defined population of children. Identification may lead to early elimination support measures including temporary bladder diversion, Mitrofanoff diversion, alpha blockers and bowel transit medications.

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Introduction

Dysfunction of urinary and fecal elimination is common in children and may be divided into three different aetiological groups which are not clearly defined and may overlap: 1) neurogenic elimination dysfunction mainly due to spinal cord anomalies; 2) social elimination dysfunction which includes a wide spectrum of acquired abnormal behaviour, from phobia of public toilets to sexual abuse and Hinman syndrome; 3) non-obstructive elimination dysfunction without spinal or peripheral detectable neurological anomalies. It is within the last group that several entities have been outlined since Beer's first description in 1915 [1]: intrinsic organ disorders like chronic intestinal pseudo obstruction (CIPO), visceral myopathy and megacystis microcolon [2—5].

Our attention was drawn to a group of six patients with precocious severe elimination disorders (urine retention and severe constipation) and distended urinary tracts associated with facial anomalies. All six patients received ureteric surgery in the first few years of life with disappointing outcomes mainly due to persistent bladder dysfunction. None of these cases could enter one of the three above-mentioned categories or be linked to the Ochoa syndrome, which combines an abnormal facial expression and urinary tract distension related to an identified gene defect [6]. The observations of these patients are reported and discussed in the light of other reported syndromes.

Patients

Six patients (four males, two females) presented during the neonatal period (4/6) or the first year of life (2/6) with early elimination disorders including urine retention (6/6) and severe constipation (6/6) (Table 1). Two had severe urosepsis, one associated with acute neonatal renal failure. The age of presentation of these six patients ranged from 1 day to 13 months (mean 4 months) with four during the neonatal period. Ultrasound and radiological investigations showed a dilated urinary tract with large ureters and a distended bladder (3/6) or a small trabeculated bladder (3/6) associated with severe bilateral grade 4—5 reflux (5/6) (Figs. 1 and 2). MRI scan of the spinal cord was normal as well as the endoscopic examination of the urethra in boys. Rectal biopsies allowed us to rule out Hirschsprung's disease in all cases. DMSA isotopic studies were abnormal (6/6) and associated with increased serum creatinine in three patients. Urodynamic studies (Fig. 3) confirmed bladder dysfunction with vesico-sphincteric dyssynergia and post micturition urine residue in all except one with a large acontractile bladder.

Cranial and facial anomalies were noticed in all, including macrocephaly and hydrocephalus (3/6); arched palate with macroglossia (1/6); hypertelorism and a flat nose root (1/6); forehead prominence, bilateral epicanthi, big and abnormally positioned ears, triangular mouth and facial hypoplasia (1/6). Three had associated axial hypotonia and five poor psychomotor development. Cerebral MRI showed various anomalies including ventricular septal defect (1/6), hydrocephalus (3/6), craniostenosis and plagiocephalia (1/6), and cortical atrophy (3/6).

Associated symptoms included cardiac and respiratory failure at birth (2/6) and clynodactylia (1/6). Four had a normal karyotype, whereas one had a deletion on q11 chromosome 22 and another X-linked inheritance Golabi Rosen syndrome.

Family history was remarkable in several patients, including two cases of Golabi Rosen syndrome in one family with Wilms' tumour found in both our patient and his cousin; two early deaths (infantile spinal amyotrophy with deletion exon 7 of gene SMN) and several miscarriages. One patient's sister presented with similar symptoms but was followed elsewhere.

Table 1 Patient data.

Sex	Age at presentation/Age at latest evaluation	Urinary symptoms	Constipation	Cephalic anomalies
M	Neonate/11 years	Urine retention, VUR, urosepsis, megabladder	(+)	Macroglossia, big face, narrow highly arched palate, large nostrils, axial hypotonia, hydrocephalus
M	Neonate/6.5 years	Renal failure, dysuria, megabladder, VUR	(+)	Macrocephaly, frontal bossing, forehead prominence, hydrocephalus
M	Neonate/5.5 years	Renal failure, VUR, small trabeculated bladder	(+)	Square-shaped ears, small mouth, cerebral cortical atrophy
F	Neonate/5 years	Urosepsis, detrusor sphinter dyssynergia, megaureter	(+)	Hypertelorism, flat and large nose root, palpebral fissures antimongoloid slant, forehead prominence, fine hair, axial hypotonia, temporal and frontal cortical atrophy
M	13 months/11.5 years	Urosepsis, VUR, dysuria	(+)	Dental anomalies, craniostenosis, amblyopia, plagiocephalia
F	11 months/7.5 years	Urinary retention, VUR, megabladder	(+)	Forehead prominence, bilateral epicanthi, big ears abnormal in position, triangular mouth, middle floor hypoplasia, fifth finger clinodactyly, nipples apart, axial hypotonia, hydrocephaly, mild sustentorial atrophy, mega citerna magna