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Urological outcome in patients with Currarino syndrome

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ABSTRACT

Introduction: Currarino syndrome is a type of caudal regression syndrome characterized by the association of hemisacrum, anorectal malformation and presacral mass. Only few studies on small series report the incidence of urinary dysfunction in Currarino syndrome. Our aim was to evaluate the urological outcome in patients with Currarino syndrome.
Patients and methods: We retrospectively reviewed all Currarino syndrome patients treated in our institution. Of 20 patients, we could evaluate the urological outcome in 16. This group of patients underwent clinical, radiological and urodynamic evaluation.
Results: All 16 patients had a sacral defect, fourteen of them presenting a presacral mass (87.5%), eight a tethered cord (50%), and 7 anorectal malformations (43.7%). Eight patients underwent neurosurgical treatment for neural tube defects. In 14 patients, the presacral mass was resected. One case presented detrusor overactivity, 2 recurrent urinary tract infections and 2 vesicoureteral refluxes. Both patients with lipomyeloschisis had a neuropathic bladder. All the other patients could void the bladder spontaneously. Renal function was normal in all.
Conclusion: Currarino syndrome is a rare congenital disorder presenting a variable phenotype. Urological outcome is good in the majority of patients.

Currarino syndrome (CS) was first described by Currarino et al. [1] as a congenital disorder characterized by the triad of anorectal malformation (ARM), sacrococcygeal defect, and presacral mass. The only mandatory clinical feature for diagnosis of CS is the sacral anomaly. Mutations of *MNX1* gene have been reported in nearly all familial CS patients, with a 30% of CS sporadic patients. Current studies reported a mutation of *MNX1* that is detected in about 50% of affected patients [2]. The presacral mass can be a teratoma, a dermoid cyst, a rectal duplication, or a combination of them. Different spinal abnormalities can be associated, such as anterior meningocele, tethered cord, or lipoma.

The most frequent type of ARM in CS is rectoperineal fistula [3]. However, different types of ARM were described in CS (rectourethral fistula, rectovestibular fistula, cloacal malformation).

Among the many different types of sacral agenesis described [4] and classified into 5 categories, the sacral anomaly in CS belongs to the 4th group (hemisacrum). Many associated congenital spinal abnormalities were described [5]. Neural tube malformations, particularly tethered cord, can be associated with CS, and spinal magnetic resonance imaging (MRI) is the gold standard for detection of these anomalies. CS has variable phenotypes: some patients presented with only asymptomatic hemisacrum without other anomalies, others presented the complete triad and other associated gynecologic or urologic malformations. Due to this great variability of penetrance, the name of Currarino syndrome is more appropriate than Currarino triad [6].

Associated gynecologic malformations include bicornuate uterus, septate vagina, and bifid clitoris [7]. Urologic involvement may be characterized by duplex ureter, horseshoe or duplex kidney, vesicoureteral reflux (VUR), dysplastic kidney, or hypospadias.

There are only few reports about the therapeutic management and long term follow-up [8,9] and even less analyzing the urological outcome in patients with CS. In the largest reported series including 205 CS patients, 35% had urinary tract dysfunction [4], in other series the incidence of urological complications was about 20% [9]. A study performed by Crétolle reported an incidence of 3.4% of neuropathic bladder and 14% of urinary incontinence in a series of 29 patients [8]. Another study performed by Lee reported an incidence of 36% of urinary incontinence in a series of 14 patients [9]. However, in none of these studies it was possible to identify individual risk factors for urological dysfunction. In particular, owing to the small number of patients, no significant relation among the type of sacrococcygeal defect, treatment of presacral mass or anorectal anomaly, and urological sequelae was reported. Another confounding factor is the treatment received, that can

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Table 1
Patients collected data.

ID	Gender	Age at presentation (months)	Genetics	Neural tube defect	Age at NST (months)	Presacral mass	Age at OST (months)	ARM	Age at GIT (months)	Number of surgical intervention	Bowel outcome	Urological outcome	Follow-up (months)
1	F	48	Sporadic	Tethered cord, lipomyelocele	42	Lipoma	42			2	Constipation	Neuropathic bladder, VUR	21
2	F	60	Sporadic	Tethered cord	54					1			93
3	F	1	Sporadic			Lipoma	10	Rectoperineal fistula	10	2			85
4	F	3	Sporadic	Tethered cord, lipomyelocele	23	Sacral dermoid	26			2	Constipation	Neuropathic bladder	18
5	M	24	Sporadic	Tethered cord	26	Teratoma	32			2	Constipation	Detrusor overactivity	74
6	F	72	Sporadic	Tethered cord	76	Anterior meningocele	88			2	Constipation		55
7	F	144	Sporadic (mutation)	Tethered cord	135	Anterior meningocele	135			2			133
8	M	1	Sporadic	Myelocystocele				Rectoperineal fistula	4	1			64
9	M	1	Sporadic	Tethered cord	13	Teratoma	33			2	Fecal incontinence	UTI	36
10	F	12	Sporadic (mutation)			Anterior meningocele	22	Rectal stenosis	22	2	Fecal incontinence	VUR	35
11	F	12	Sporadic			Teratoma	9			1	Constipation		94
12	F	2	Sporadic	Diastematomyelia		Teratoma	2	Rectoperineal fistula	2	2	Constipation		40
13	F	36	Sporadic			Teratoma	51	Rectoperineal fistula	38	2	Constipation		213
14	F	24	Sporadic	Tethered cord	30	Sacral dermoid	32			2	Constipation	UTI	82
15	F	24	Familiar (mutation)			Anterior meningocele	32	Rectoperineal fistula	33	2	Constipation		98
16	F	24	Familiar (mutation)			Anterior meningocele	27	Rectoperineal fistula	27	2			220

NST: Neurosurgical treatment; OST: Oncological surgical treatment; ARM: Anorectal malformation; GIT: Gastrointestinal treatment; VUR: Vesicoureteral reflux; UTI: Urinary tract infection.

obviously influence the final urological outcome. In patients treated for sacrococcygeal teratomas long-term sequelae, including neuropathic bladder or bowel abnormalities, have been observed in 11%–41% of survivors [10–13].

Our aim was to evaluate the urological outcome in a series of CS and try to elucidate if bladder function disorders could be owing to a congenital malformation or be the sequelae of surgical treatment.

1. Materials and methods

We studied retrospectively 20 CS patients (13 girls and 7 boys) evaluated in a single center from 1991 to 2010 (Table 1).

Inclusion criterion for diagnosis of CS was the presence of hemisacrum. Every case underwent history and physical evaluation, renal function assessment, urinalysis, ultrasound examination, sacrum radiograph, genetic molecular analysis of the *MNX1* gene, cystogram, magnetic resonance and urodynamic studies.

Sacrum radiograph was performed in 2 projections (anteroposterior and lateral). MRI was always performed in patients with diagnosis of hemisacrum.

Urodynamics was performed in children more than 2 years old with suspect of neuropathic bladder owing to their voiding habits (dropping, using diapers, not toilet trained) or some radiological signs (bladder profile at cystogram or ultrasound) after the last surgical treatment. In toilet trained children without any voiding trouble urodynamics and in infants, urodynamics was not performed in order to avoid any undue invasiveness. Urodynamics study included cystometrography and uroflowmetry, electromyography (EMG) was not performed.

The decision to perform detethering surgery was ultimately made by the neurosurgeon and based on MRI findings, somatosensory evoked potential and symptoms. Cases of tethered cord were identified by magnetic resonance imaging, which revealed the conus medullaris below the L2 vertebra. Presacral masses and anorectal malformations, when present, were treated by posterior sagittal anorectoplasty (PSARP) as described by Peña in 1982 [14] (Fig. 1). To reduce the risk of recurrence of presacral masses, excision of the coccyx was always performed.

The surgical timing was decided by the two surgical teams. In case of neurosurgical treatment, this was the first to be performed, followed by PSARP in case of presence of ARM and, finally, excision of presacral mass.

All the patients have been operated by the same two surgical teams: spinal cord surgery was performed by neurosurgical team, anorectal and presacral mass surgery by the general surgeon team.

After surgery, patients were followed by a multidisciplinary team including neurosurgeons, pediatric surgeons and urologists at regular intervals (at least 3, 6, and 12 months after each surgery and then once per year, more often in case of symptoms).

To evaluate postsurgical urinary outcomes, we considered: renal function, presence of urinary infections, vesicoureteral reflux, ultrasound pattern, voiding pattern. All patients were evaluated clinically, by ultrasound, renal function using Schwartz creatinine clearance formula [15] and urinalysis, in case of anomalies cystogram, nuclear scan and/or urodynamic testing were performed.

Regarding voiding pattern, each patient was categorized as continent, intermittently incontinent, or continuously incontinent, according to the Report from the Standardisation Committee of the International Children's Continence Society [16]. Bladder was defined neuropathic if the patient had clinical or urodynamic signs of voiding disorder or trouble of urinary continence.

Fisher exact test was used for statistical analysis to evaluate the correlation between neuropathic bladder and the associated spinal abnormalities, age at presentation, age at surgery, type of mass resected, type of ARM, type of neural tube defect and number of surgical treatment. A $p < 0.05$ was considered statistically significant.