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LONG TERM FOLLOW UP ON CONTINENCE AND RENAL FUNCTION IN 50 PATIENTS WITH CAUDAL REGRESSION SYNDROME. A MULTICENTER STUDY.

Aims of Study

Caudal Regression Syndrome (CRS) is a rare condition. It is a heterogeneous constellation of caudal anomalies (agenesis of the caudal spinal tract, anal imperforation, genital anomalies, dysplastic lower limbs, renal and pulmonary dysplasia, in variable association). It belongs to closed spinal dysraphism and may escape early diagnosis when anorectal malformation, cutaneous stigmata or severe lower limbs malformation are absent. Neurogenic involvement of micturition is very frequent and the risk of both upper urinary tract derangement and urinary incontinence is high.

We developed a single database, with protected on-line access, to collect clinical data of patients with closed spinal dysraphism from 5 Italian paediatric centres, in order to collect as many cases as possible with a follow up long enough to assess the fate of upper urinary tract and the possibilities to treat urinary incontinence.

This project started on January 2002.

We present the results in 50 patients, focusing on those with severe impairment of continence and/or renal failure.

Methods

The inclusion criteria were the following:

1. Diagnosis of CRS (only sacral or lumbo-sacral defects were considered essential).
2. Imaging about spinal defect and spinal cord available.
3. Available description of urodynamic observations associated with characteristic symptoms and signs.
4. Serum creatinine, renal sonography and/or scanning available.
5. Follow up longer than 10 years.

The database, powered by a professional agency, is accessible on-line from the five Italian centres and protected by passwords. Patient's family name is shown only cryptic, but the system is able to recognise if a new patient has been previously inserted by another centre, allowing to append new clinical observations without duplicating the record (one patient – one record). Each operator can modify or implement only the data belonging to his own patients, but is able to know clinical features of all the patients. The entire database can be analysed through a number of queries.

Results

Fifty patients, 27 females and 23 males, were considered eligible. Mean age was 13.3 years (range: 11.2 to 35.9). An anorectal malformation was found in 14 patients, 2 of which had a cloaca. Unilateral renal agenesis was found in 8, while 1 patient underwent nephrectomy for multicystic kidney.

Seven patients (14%) have normally innervated bladder and are continent, only 2 of which complaining of episodes of nocturnal enuresis.

Of the remaining 43 patients with neurogenic bladder, 21 (48.9%) are dry between voiding obtained either by intermittent self-catheterisation or by bladder expression, 5 (11.6%) are usually dry up to 2 ½ hours (with dribbling linked mainly to their physical activity, amount of fluid uptake and emotional status) and 14 patients (32.5%) are moderately to severely incontinent; the remaining 3 (6.9%) had permanent urinary diversion due to complex pelvic malformations in which urinary and digestive tract could not be reconstructed in infancy. One of these last has recently undergone bladder reconstruction by ileocecal reservoir with Mitrofanoff continent conduit (appendix).

Upper urinary tract is mildly dilated in 12 (24%) and moderately dilated or scarred in 6. Severe derangement was found in only one case and led to transplantation.

Vesico-renal reflux was found in 24 renal units of 16 patients, all but one successfully treated.

Overall renal function is slightly impaired in 4 (serum creatinine between 1.2 and 1.4 mg/dL, all young adults), moderately impaired in one (1.9 mg/dL, 25 years old) while a patient who underwent multiple surgical procedures including temporary diversion, developed an end stage renal failure and underwent successful renal grafting.
One patient recently died, due to unrelated cause.

Conclusions

Matching urological data and imaging of all patients with impaired renal function, we could observe that bony defect exceeded 3 sacral bodies except in a patient with complicated multi stage surgical treatment and in another one who came too late to intermittent catheterisation practice.
Extensive bony defect is also present in all the patients with dilatation of upper urinary tract except in one with complex pelvic malformation (cloaca).
The great majority of severely affected patients shows club shaped cord(high lying and without the normal taper), with double bundle shape of the cauda equina and no lipomatous tissue or tethering
Renal agenesis has an exceedingly high rate (16%) and vesico-renal reflux affects one third of the patients.
Only a minority of patients escape neurological involvement of micturition (14%) and barely half of those with neurogenic bladder do not need diapers unless undergoing bladder neck surgery.

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