Late Diagnosis of Rare Spinal Defect in a Girl with Urinary Incontinence

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Abstract

Urinary incontinence is a common problem in paediatric population and can be caused by many disturbances. To diagnose isolated nocturnal enuresis it is important to exclude physical anomalies that can lead to urinary incontinence. Sacral agenesis is a rare defect of vertebral column which can occur as an isolated defect or coexist with other anomalies. Mild clinical symptoms of the defect may delay the diagnosis and can keep patients at risk of severe complications including chronic kidney disease. Patients with caudal agenesis are characterised by higher prevalence of kidney defects and vesico-ureteric reflux and they are considered to be a group at higher risk of kidney damage. In this paper we present the case study of a 5-year-old girl with intermittent urinary incontinence admitted to the hospital for investigation of observed symptoms and further treatment. In the course of investigation, on the basis of magnetic resonance imaging and voiding cystography, the diagnosis of sacral agenesis was established. Urodynamic examination revealed overactive neurogenic bladder with reduced cystometric capacity but no obstruction. Treatment of constipation and anticholinergic agents enhancing bladder function, and physical rehabilitation were recommended as well as psychological care for the child and her parents. No neurosurgery was possible at this stage of sacral agenesis. Mild clinical symptoms in rare cases of caudal agenesis may delay the diagnosis that keep patients at risk of severe complication of chronic kidney disease.

Key words

Caudal regression syndrome; Children; Enuresis; Sacral agenesis; Urinary incontinence

Introduction

Sacral agenesis belongs to the group of rare defects of distal part of spine with incidence of 0.01-0.05/1000 births. It can occur as an isolated defect or coexisting with other anomalies in caudal agenesis syndrome (CA) which is also commonly called caudal regression syndrome. The clinical picture of CA is not homogenous – it can be associated with total or partial agenesis of spinal cord, anal imperforation, genital anomalies, bilateral renal dysplasia or aplasia, hypoplastic lungs or anomalies of lower limbs. CA can also
be a part of other complex syndromes (OEIS - omphalocele-extrrophy-imperforate anus-spinal defect; VACTERL association – vertebral anomalies, anal atresia, cardiac abnormalities, tracheo-oesophageal fistula, oesophageal atresia, renal abnormalities or radial dysplasia, limb abnormalities; Currarino triad – sacrum abnormalities, mass in presacral space, malformations of anus or rectum).

In this paper we present the case of a girl with isolated sacral agenesis and intermittent urinary incontinence who had delayed clinical diagnosis because of mild clinical presentation.

**Case Report**

A 5-year-old girl with urinary incontinence (intermittent) was admitted into hospital for investigation of the observed disorders and to establish further treatment. According to the mother, the girl was wet during the day and every night from birth until the day of admission. Anamnesis revealed one episode of urinary tract infection without fever. Because the paediatrician was not satisfied with the child's condition and felt that her development was delayed, she was referred to see a neurologist and psychologist who diagnosed mental retardation of mild severity. She started to walk by herself at 18 months of age. Detailed analysis of perinatal history revealed that the girl was born at term, from 1st pregnancy, delivered naturally with 7 points by Apgar. The pregnancy was uncomplicated and diabetes was excluded.

On admission the girl was in good condition and reported no complaints. On careful physical examination, abnormal appearance of sacro-lumbar area (Figure 1), malformation of feet and yawning anus were noticed. Mother had earlier reported to the paediatrician that in her opinion the buttocks area of her daughter looked abnormal but there were no further diagnostic work-up. Neurological examination revealed bilateral lack of Achilles tendon reflex and lack of anus sphincter reflex. There were no other abnormalities of neurological function. Detailed orthopaedic examination revealed equino-varus feet with contracture of Achilles tendon – dorsal flexion of both feet was of range of 5°. Toes IV of both feet overlapped on toes III.

Laboratory test results showed normal kidney function. Urinary tract infection was excluded. Ultrasound examination of abdominal cavity, aside from widened rectal ampulla that was a reflection of chronic constipation, did not show anomalies of the structure of the urinary system. Voiding cysto-urethrogram also did not reveal defect of urinary system. However, it visualised abnormal construction of the sacral part of spine – the image suggested sacral bone hypoplasia. Further diagnostics – magnetic resonance imaging of lumbo-sacral part of vertebral column showed normally developed sacral bone only in the S1 area with vertebra S2 partially visible. Spinal cord ended with atypical bulb shape at the level of Th12/L1 (Figure 2).

Urodynamic examination revealed overactive neurogenic bladder with reduced cystometric capacity (55 ml) with no obstruction. The presence of detrusor sphincter dyssynergia and detrusor overactivity was recorded as well. Treatment of constipation, anticholinergic agents to enhance bladder function, and physical rehabilitation were recommended as well as psychological care for the child and her parents. No neurosurgery was possible at this stage of sacral agenesis.

**Discussion**

Etiopathogenesis of sacral agenesis still is uncertain. It is supposed that defect occurs as a result of mesoderm forming before 4th week of gestational life and can be connected with toxic, infectious or ischaemic injury of neuronal and mesodermal cells migration. Some association can also be found in genetic disorders (chromosome 7q36 defect). The most documented connection is for maternal diabetes – it is estimated that in 15-20% mothers of children with sacral agenesis – isolated or coexisting with other syndrome, insulin-dependent diabetes is observed.³

Several classification systems of sacral agenesis exist. According to Renshaw's classification from 1978, sacral agenesis is classified into one of four groups according to severity of disturbance.⁴ Grading is based on the amount of sacral bone and a kind of junction between spine and pelvis. Clinical picture of the defect is variable depending of the type of agenesis. In mild cases there can be no clinical signs or only external stature anomalies are visible – unproperly developed buttocks and anomalies of feet. In more severe cases related to further types of defect (type III or IV) – aside from external body disturbances, impairment of genito-urinary system, respiratory and nervous system functions can be observed. Such a variable clinical manifestation of the defect can be a cause of late diagnosis. Early diagnosis of sacral agenesis however is exceedingly important for the
child’s development and for taking care of his general condition. The first manifestation in ultrasound examination of first trimester is shortened crown rump length in prenatal ultrasound examination. The defect can appear as a sudden rupture of spine due to lack of vertebrae and frog-like position of lower limbs. If there are external features of agenesis – proper physical examination of newborn after birth should be sufficient to suspect the defect and refer the child for further diagnosis and under specialist care. Children with sacral agenesis with neurologic and orthopaedic disturbances most often get such care rapidly. Abnormalities of urinary system function, however, may be missed at early age when there are no recurent urinary tract infections and no external malformations.

Urinary incontinence may be defined as uncontrolled urine leak that can be continuous or intermittent. Intermittent incontinence during sleep is called nocturnal enuresis and can be diagnosed in a child above 5 years of age. Only at this age that most of parents would ask health professionals to diagnose and solve the problem. Enuresis diagnosis is based on precisely performed anamnesis, physical examination, with special consideration of appearance of genito-urinary area and lower limbs, assessment of perineum sensation and performing accessory diagnostic tests. Presence of abnormalities in body composition should be suggestive of spinal anomalies, however as it was mentioned earlier, sacral agenesis can be present with no external defects.

In a child with urinary incontinence, detection of sacral structure disorders sometimes might be accidental – associated with performing other diagnostic tests. Moritoki et al described a case of a 2-year-old girl with grade IV vesico-ureteric reflux (VUR) in whom, through various diagnostic procedures, sacral agenesis was revealed. The child presented no external signs of the defect. After urodynamic examination constrictor-detrusor dissynergy was detected and in order to minimise risk of kidney damage the child was recommended intermittent bladder catheterisation. In the present case, defect of sacral spine was suspected only on voidnig urethrocystogram that was performed due to incontinence.

As caudal agenesis belongs to the group of closed neural tube defects it is not always connected with neurogenic bladder – symptoms concerning urine excretory functions can be very variable. Early recognition of defect is extremely important due to possible complications. Torre
et al assessed that in patients with caudal agenesis significantly more frequent congenital kidneys anomalies, are present – the most frequent is kidney agenesis.9 Also commonly in about 37% is VUR. Similar results were obtained by Wilmshurst et al.10 Because patients with caudal agenesis are characterised by higher prevalence of kidney defects and VUR, they are considered to be at higher risk of kidney damage.9,10 Therefore, it is extremely important to diagnose the defect as soon as possible and introduce treatment in order to make bladder empty more efficiently. In the present case we found no urine retention in the bladder after voiding and there was no vesico-ureteral reflux as well. Thus this kind of neurogenic bladder has low risk of progression to kidney insufficiency.

Conclusions

As a conclusion it has to be emphasised that mild clinical symptoms in rare cases of caudal agenesis may delay the diagnosis that keep patients at risk of severe complication of chronic kidney disease.

Conflict of Interest

Authors declare no conflict of interest.

References