Caudal Regression Syndrome: A Case Report of a Palestinian Children

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Abstract

Caudal Regression Syndrome (CRS), also referred to as caudal dysplasia and sacral agenesis syndrome, is defined as total or partial agenesis of the sacrum and lumbar spine, frequently associated with other developmental malformations characterized by varying degrees of developmental failure early in gestation. It involves the lower extremities, the lumbar and coccygeal vertebrae and corresponding segments of the spinal cord, and true pathogenesis is unclear. CRS affecting 0.1 to 0.25 per 10,000 pregnancies. Here, we report a male boy aged 4 years old, diagnosed as a case of CRS according to sacral agenesis, coccyx agenesis, iliac bone hypoplasia, the fifth lumbar spine agenesis, spina bifida occulta, hypoplastic lower limb, bladder incontinence, encopresis and associated with other anomalies.

Keywords: Caudal Regression Syndrome (CRS); Caudal Dysplasia; Sacrum; Lumbar Spine

Introduction

CRS is a complex, heterogeneous constellation of congenital caudal anomalies affecting the lower part of the body along with multiple systems involvement (neurological, gastrointestinal, renal and genitourinary defects) [1-3]. The term CRS was first used in 1961 by Duhamel [4]. CRS is a very rare and unusual disorder in the general population but occurs in about one in 350 infants of diabetic mothers, representing an increase of about 200-fold over the rate seen in the general population [5]. It occurs in one per 25,000 live births to 1 - 2.5 per 100,000 newborns and male to female ratio is 3:1 [6-9]. Many etiological factors were contributed to CRS like retinoic acid [10,11] organic solvents and folic acid deficiency, but the most important factor is hyperglycemia where the risk of CRS increases when it is poorly controlled [12]. The embryologic insult occurs at the mid-posterior axis mesoderm and the lesion originates before the fourth weeks of gestation [13]. CRS may range from absent coccyx as an isolated finding without neurological sequelae, to sacral or lumbosacral agenesis [14]. It can affect the lower extremities, the lumbar and coccygeal vertebrae and corresponding segments of the spinal cord. The neurologic, orthopedic, gastrointestinal, genitourinary and cardiac anomalies, imperforate anus are commonly seen [6,15].

Patient Report

A four years old boy was admitted to El-Doura pediatric hospital due to symptoms and signs of follicular tonsillitis that is unresponsive to oral treatment. History of three orthopedic surgery for talipes equinovarus, diurnal and nocturnal enuresis, encopresis, inability to walk. During physical examination the child was conscious, oriented with macrocephaly. Mass on the sacral area, lower limbs muscle hypotrophy, scars on both feet due to previous orthopedic surgery. Skeletal survey reveal agenesis of sacral, coccyx and the fifth lumbar vertebrae. Spina bifida occulta, hypoplasia of iliac bones, 13 pairs of ribs. Others, macrocephaly, ear deformity. Complete blood count, serum electrolytes, renal functions test, liver functions test, serum blood sugar were within normal averages.

Discussion

CRS is a rare and which is often sporadic congenital malformation of the lower vertebral column. It is characterized by partial or complete absence of sacrum and lumbar vertebrae. This syndrome is accompanied by severe lower extremity and pelvis deformities, neurological deficits, neural tube defects and genitourinary, gastrointestinal, cardiac anomalies [16,17].

In our case there was sacrum, coccyx and the fifth lumbar vertebrae agenesis, spina bifida occulta of the fourth lumbar vertebrae, pes equinovarum deformity which was operated three times, hypoplasia of iliac bones, 13 pairs of ribs, macrocephaly, ear deformity, loss of gluteal fold, sacral mass, muscle wasting of the lower limbs, associated with enuresis and poor anal sphincter tone lead to encopresis. CRS is a rare anomaly, with about 300 cases reported, characterized by abnormal development of the caudal spine of the developing fetus [18,19]. CRS, also referred to as caudal dysplasia and sacral agenesis syndrome, is a rare congenital malformation characterized by varying degrees of developmental failure early in gestation. Of the 100,000 pregnancy occurred, only 1 to 2.5 is resulted in diagnosis of caudal regression syndrome [20]. The prevalence of CRS is estimated to be one in 25,000 live births [21]. The true pathogenesis is unclear, but in about 15 - 25% of mothers of children with caudal regression syndrome have insulin-dependent diabetes mellitus [22], most cases are sporadic, but multiple genetic factors play a role in determining the risk of developing this abnormality [11]. Genetic predisposition, vascular hypoperfusion and drug-related with minoxidil and trimethoprim-sulfamethoxazole have been proposed as possible causative factors [1,7,8,23]. Genetic mutations in the coding sequences of HOXD13, CYP26A1 and HLXB9 have been suspected in the pathogenesis of CRS [24]. Evaluation for suspected CRS prenatally is possible through fetal ultrasound (US) and magnetic resonance imaging (MRI), is possible starting at around 20 weeks gestational age [3,25]. Diagnosis in the first trimester is difficult because of incomplete sacral ossification. Prenatal ultrasound is the best method to diagnosis this anomaly. Ultrasonographic findings are variable and will depend on the extent and severity of the defect [17]. In first trimester we may suspect of such a case of CRS if we find: short crown-rump length, "protuberance" of lower spine, increased nuchal translucency and abnormal yolk sac. In the second and third trimester may appear: abrupt termination of spine (best seen on sagittal section), no spine visible on axial views of abdomen and short trunk [26,27]. The Pang's classification of lumbosacral agenesis with five types: where Types I and II represent total sacral agenesis with and without associated lumbar vertebral agenesis; Type III represents subtotal sacral agenesis with at least S1 present; Type IV has a hemisacrum and Type V includes coccygeal agenesis [28]. After birth, physical examinations and imaging studies are routinely performed to diagnose the type and extent of damage to lower parts of the body. CRS is associated with other multiple congenital anomalies include cardiac, gastrointestinal and orthopedic (See table 1).

<table>
<thead>
<tr>
<th>Orthopedic</th>
<th>Iliac wings approximated or fused, clubfeet, lower extremities contractures, rib hypoplasia, scoliosis [6,12,29].</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genitourinary</td>
<td>Cystic renal, dysplasia, hydronephrosis, penoscrotal inversion, penile agenesis, cryptorchidism, Vesico-ureteral reflux [6,10]</td>
</tr>
<tr>
<td>Gastrointestinal</td>
<td>Anorectal and duodenal atresia, malrotation, inguinal and umbilical hernia [6,10,12]</td>
</tr>
<tr>
<td>central nervous</td>
<td>Atrial septal defect, ventricular septal defect [10,16]</td>
</tr>
<tr>
<td>cardiovascular</td>
<td></td>
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</tbody>
</table>

Table 1: CRS associated with other congenital anomalies.

In differential diagnosis of CRS may includes, sirenomelia, segmental spinal dysgenesis (SSD), myelomeningocele and VACTERL association should be evaluated [29,30]. Sirenomelia, which is also known as mermaid syndrome, is a rare and fatal congenital defect characterized by varying degrees of lower limb fusion, thoracolumbar spinal anomalies, sacrococcygeal agenesis, genitourinary, and anorectal anomalies.
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atresia [18,29]. Currarino syndrome consists of sacral agenesis, mass in the presacral space and malformation of the anus and rectum. Occurs due to mutation in the HLXB9 gene, which is inherited in an autosomal dominant manner [31]. VACTERL association includes combination of abnormalities: vertebral, anorectal, cardiac, tracheoesophageal, renal and limbs [29].

Treatment and prognosis

Treatment is difficult, multidisciplinary and largely supportive [32]. The main goals of treatment include maintaining and improving renal, cardiac, pulmonary and GI function, preventing renal infection and achieving continence. Orthopedic intervention is necessary to correct the associated malformations. The prognosis for children with CRS largely depends on the severity of vertebral anomalies and associated malformations [13,30,33]. The prognosis is poor for patients with total sacral agenesis, since associated musculoskeletal, respiratory, cardiac, gastrointestinal, and genitourinary malformations predispose to early neonatal death [34]. Renal anomalies could lead to frequent urinary tract infection and progressive renal failure. As surviving patients would have normal mental function and no cognitive impairment; attention should be given to preserve kidney function with adequate psychological support [35].

Conclusion

Caudal regression syndrome is a rare entity with a known association with maternal diabetes. It is characterized by sacrococcygeal dysgenesis with an abrupt termination of a blunt-ending spinal cord. Clinicians must keep in mind that CRS may be seen without any risk factors. Adequate control of hyperglycemia in diabetic mothers is important to avoid sacral agenesis. Prenatal ultrasonographic diagnosis of CRS is possible at 22 weeks' of gestation. Early recognition is important because the imaging specialist may be the first to suggest the in-utero diagnosis of CRS to the obstetrical care provider.

Bibliography


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