Lumbosacral Agenesis in an Infant of a Diabetic Mother: A Case Report

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ABSTRACT

Sacral agenesis, known also as caudal regression syndrome, is a rare congenital abnormality characterized by absence of a variable amount of sacrum, lumbar spine and associated neural elements. It is mostly seen in infants of diabetic mothers. Nearly all patients have genitourinary anomalies, neurogenic bladder being the most common. Other associated anomalies include cardiac, gastrointestinal and orthopedic. Here, we present a rare type of lumbosacral agenesis in an infant of a diabetic mother. It was diagnosed antenatally at 26 weeks of gestation.

Key words: Caudal regression, Caudal dysplasia, Sacral agenesis.

Case Report

Our patient was born to a 24 year old mother who was discovered to be diabetic two years previously. She was on insulin and metformin with poor glycemic control. She was referred at 26 weeks of gestation for detailed fetal scan and was diagnosed antenatally to have a baby with sacral agenesis. The ultrasound showed sudden termination of the spine below the lower thoracic region. The fetal legs did not move during examination and were in an abnormal “frog like” position. The pelvis was hypoplastic while other organs were normal.

The patient was followed in our antenatal clinic and was growing below the 3rd centile. She gave birth to a full term female baby by caesarian section due to transverse lie at term. The newborn was 1.7 kg in weight, 27cm in length and 27cm in head circumference, all below the normal ranges. She was in respiratory distress and needed oxygen since birth. On examination, she had no dysmorphic features but her rib cage was small. She has ejection systolic murmur which was due to an atrial septal defect (ASD) that was diagnosed by 2D-echocardiogram. Her abdomen was soft and lax with no organomegaly. Neurological examination of the lower limbs showed hypoplastic and spastic legs, no response to painful stimuli and absent deep tendon reflexes. Upper limbs were normal with good strength and normal reflexes. See Fig. 1, 2, 3, 4.

Skeletal radiographic examination showed missing ribs, absence of lower thoracic, lumbar and sacral vertebrae. She had hypoplastic pelvis and lower limbs, severe flexion and abduction of the hips, severe flexion of the knees and bilateral club feet (Fig 5).

Her investigations showed normal renal and brain ultrasound. She died at the age of two days due to respiratory failure.

Discussion

Sacral agenesis is a severe congenital abnormality that includes complete or partial agenesis of the sacrum and lumbar vertebrae. It is
Fig. 1: Hypoplastic legs

Fig. 2: Lower limbs are flexed and externally rotated

Fig. 3: Frog like position.

Fig. 4: Dimples at sites of hypoplastic hips.

Fig. 5: Skeletal X-ray showed missing ribs and hypoplastic lower limbs bones.
a rare disease that occurs in one per 25,000 live births to 1-2.5 per 100,000 newborns. It occurs most commonly in infants of diabetic mothers, not gestational, at a rate of one in 350, making caudal regression syndrome as the most characteristic fetal abnormality of diabetic embryopathy, although this estimation seems to be high; not all cases of sacral agenesis were born to diabetic mothers, only 8-22% of them were born to those pregnancies. Many etiological factors were contributed to sacral agenesis like retinoic acid, organic solvents and folic acid deficiency, but the most important factor is hyperglycemia where the risk of caudal regression syndrome increases when it is poorly controlled, as in our case. Most cases are sporadic, but multiple genetic factors play a role in determining the risk of developing this abnormality.

A mutation in the HLXB9 gene, which is inherited in an autosomal dominant manner, was described in Currarino syndrome or triad which consists of sacral agenesis, mass in the presacral space and malformation of the anus and rectum. This mutation was specific to this syndrome and was not seen in other forms of sacral agenesis.

Caudal regression syndrome is associated with other multiple congenital abnormalities. Cardiac anomalies are not common, but have been described like in our patient who had ASD. Central nervous system anomalies like hydrocephalus and myelomeningocele are not rare but were not encountered in our patient. Orthopedic anomalies like scoliosis, limb contracture, club feet and rib hypoplasia are common findings which all were present in our patient. They may have imperforate anus, inguinal hernia and malrotation which was not the case here.

The most common and most significant anomalies are the genitourinary where the neurogenic bladder is the commonest. Renal defects include: unilateral renal agenesis, horseshoe kidney, ureteral duplication and bladder extrophy. Renal ultrasound in our patient was normal but she, unfortunately, didn’t live long to evaluate the function as most of them would have abnormal bladder and urethral dysfunction with incontinence.

Respiratory complication could occur due to instability between the pelvis and the spine causing the pelvis to migrate up the abdominal cavity and affect the pulmonary function, which we believe it was the case in our patient who died due to respiratory failure.

**Conclusion**

Adequate control of hyperglycemia in diabetic mothers is important to avoid sacral agenesis. Prognosis depends on the severity of the case and the associated anomalies. 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.

**References**

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