CASE REPORT

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Prenatal Diagnosis of Caudal Regression Syndrome in Non-Diabetic Maternal Fetus

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ABSTRACT Caudal regression syndrome is a rare complex abnormality. It is characterized by a series of congenital abnormalities, including complete or partial agenesis of the sacrum and lumbar vertebrae associated with urinary, cardiovascular and gastrointestinal malformations. The incidence is estimated to be approximately 1:60,000 births, with a male:female ratio of 2.7:1. The etiopathogenetic mechanism of caudal regression syndrome is unknown. Caudal regression syndrome is more frequent in patients with diabetes than in non-diabetic women. We report a case of caudal regression syndrome in non-diabetic woman. Prenatal ultrasonographic examination and fetal MR images are the most useful for diagnosed the caudal regression syndrome.

Keywords: Caudal regression syndrome; prenatal diagnosis; ultrasonography, prenatal

Caudal regression syndrome (CRS), also referred to as sacral agenesis syndrome or caudal dysplasia. CRS presents varying degrees of vertebral anomalies from partial sacral agenesis to complete absence of the lumbosacral spine with urinary, cardiovascular and gastrointestinal malformations.[1] The malformations are due to defects in neuralization around the 28th day of fetal development. The pathogenesis of CRS is not clear. Genetic predisposition, vascular hypoperfusion and maternal diabetes have been suggested as possible causative factors; however, maternal hyperglycemia has been recognized as the most common teratogen in this syndrome.[2] CRS is more frequent in patients with diabetes than in non-diabetic women. Therefore, we report a case of caudal regression syndrome in non-diabetic woman.

CASE REPORT

A 27-year-old gravida 2, para 1 woman was referred to our perinatology unit at 31 weeks of gestation. She previously had a c-section at 39 weeks. Her child is healthy. Her medical history was unremarkable. Her oral glucose tolerance test and glycosylated hemoglobin (HbA1C) level were normal. Ultrasonographic examination showed a singleton fetus with normal amniotic fluid volume. The posterior fossa was seen abnormally with enlarged lateral ventricles (Figure 1). In addition thoracale meningocele and

kyphoscoliosis, a single pelvic kidney and CRS were visualized. There was absence of lumbosacral vertebrae and hypoplasia of the lower limbs. The lower extremities crossed each other in a "frog-like" position. Lower limb movements were absent during ultrasonographic examination. Then, a fetal MR imaging was performed. MR images were obtained in the fetal coronal, transverse and sagittal planes. These images showed ventriculomegaly, cerebellar herniation, thoracale meningocele, a single kidney, a single umbilical artery (Figure 2). Based on abnormality, CRS was diagnosed and the parents were offered both perinatal karyotype analysis and termination of pregnancy, but refused.

At 38 weeks, c-section was performed due to uterine contractions. The Apgar scores were 7 and 8 at 1 and 5 min, respectively. The infant weighed 2560 g , female gender. Examination by a neonatologist specialized in congenital malformations revealed a thoracale meningocele and CRS with the typical frog-like position (Figure 3). She is still alive at neonatal intensive care unit.

DISCUSSION

CRS is a rare complex abnormality. It is characterized by a series of congenital abnormalities, including complete or partial agenesis of the sacrum and lumbar vertebrae associated with pelvic deformity. Hypoplasty and flexion contractures of the lower extremities and clubbed feet are commonly seen.[3] Patients with CRS lack motor function below the level of the remaining normal spine. CRS is often associated with neural tube defects, gastrointestinal, cardiovascular, genitourinary anomalies. The crown-rump length of CRS fetuses in the first trimester is shorter than the normal fetuses.[4]

CRS is about 200-400 times more frequent in patients with diabetes than in non-diabetic women. The incidence is estimated to be approximately 1:60,000 births, with a male:female ratio of 2.7:1.[2] Therefore CRS is the most specific fetal malformation in maternal diabetes. However, CRS has occurred in nondiabetic women also, therefore confounding etiologic factors.[5]

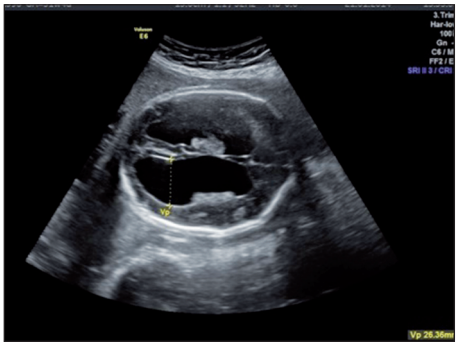

FIGURE 1: Ultrasonographic examination showed enlarged lateral ventricles.

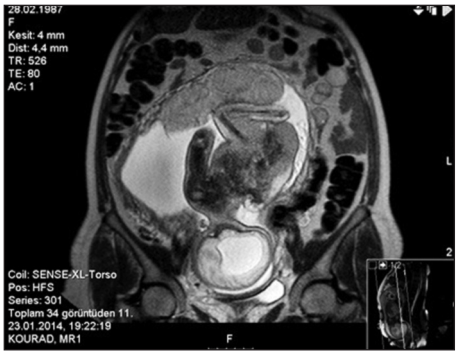

FIGURE 2: MR images showed the lower portion of body was small compared with the midbody and chest.

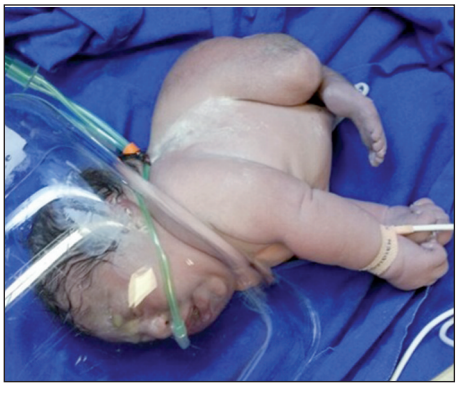

FIGURE 3: The newborn infant with CRS.

The etiopathogenetic mechanism of CRS is unknown. Defective development of caudal portions of fetus, originating from the axial mesoderm is probably the common pathway.[6] It is different from Sirenomelia which is thought to result from a vascular compression, obstruction or malformation causing severe ischemia of fetal caudal portions.[3] CRS is related to maternal diabetes,

sirenomelia being essentially linked to vascular abnormalities.[5,7]

The case presented here was additionally complicated by a severe thoracale meningocele with type-II Arnold- Chiary malformation.

In our case, the mother was not diabetic however, diabetes, particularly with poor glycemic control is the most recognized risk factor. The prognosis for fetus with CRS depends on the severity of the lesion and the presence of associated anomalies. Prenatal ultrasonographic examination and fetal MR images are the most useful for diagnosed the CRS.

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Conflict of Interest

No conflicts of interest between the authors and / or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

Authorship Contributions

Idea/Concept: *Yasin Ceylan;* ***Design:*** *Yasemin Doğan;* ***Control/Supervision:*** *Sebiha Özkan;* ***Data Collection and/or Processing:*** *Yasin Ceylan;* ***Analysis and/or Interpretation:*** *Sebiha Özkan;* ***Literature Review:*** *Yasin Ceylan;* ***Writing the Article:*** *Yasin Ceylan;* ***Critical Review:*** *Yasemin Doğan;* ***References and Fundings:*** *Yasemin Doğan;* ***Materials:*** *Yasin Ceylan.*

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