A Rare Case Report: Caudal Regression Syndrome in the Baby of a Mother with Hypothyroidism

Yusuf Atakan Baltrak,1 Seniha Esin Söğüt,1 Onursal Varlıklı,1 Nazan Karadeniz2

1Department of Pediatric Surgery, Health Sciences University Kocaeli Derince Training and Research Hospital, Kocaeli, Turkey
2Department of Obstetrics and Gynecology, Health Sciences University Kocaeli Derince Training and Research Hospital, Kocaeli, Turkey

ABSTRACT

Caudal regression syndrome (CRS) is part of a spectrum of conditions that include imperforate anus, sacral agenesis and syringomyelia. Most cases of caudal regression are sporadic or associated with gestational diabetes. In this case study, we describe a case of characteristic CRS in a patient of a mother with hypothyroidism with anal atresia, flexion contracture on the hips and knees, pes equinovarus, and the absence of a sacrum and coccx.

Keywords: Anal atresia, congenital abnormalities, hypothyroidism, imperforate anus

INTRODUCTION

Caudal regression syndrome (CRS) is a rare sporadic disorder characterized by congenital abnormalities of the lower limbs, gastrointestinal system and urogenital system, and affects the terminal spinal segment.1-3 Although the exact cause remains unknown, some environmental factors, genetic tendency and vascular hypoperfusion were suggested as possible causes.3,4 Many studies reported that CRS occurs 200 to 250-fold more in the babies of mothers with insulin-dependent diabetes mellitus.1,2,5

This case report aims to present a rare case with CRS and anal atresia detected in the baby of a mother with hypothyroidism.

CASE REPORT

The case was delivered as the first male baby of a 20-year old primigravida mother through caesarean section at 39th gestational week. The mother had a history of levothyroxine sodium use at a dose of 25 mg because of hypothyroidism. The history taken from the mother revealed that she was followed without any problem during the pregnancy, however, perinatological examination was not conducted. APGAR scores of the patient at minutes 1 and 5 were 9 and 10, respectively. The birth weight of the baby was 2,410 g, his length was 45 cm (<3%) and head circumference was 34.5 cm (25-50p). The first physical examination revealed flexion contracture of the hip and knees as well as anal atresia (Fig. 1). The direct x-ray imaging demonstrated that there was not any sacrum and coccx (Fig. 2a). Since anal concavity did not exist and meconium was detected in the urine analysis, the colostomy procedure was decided. A small secundum atrial septal defect was diagnosed in cardiac Echocardiography. No sacrum...
was observed in spinal ultrasonography for differential diagnosis. A cyst of 2.5 cm was detected at the end of the lower lumbar vertebra. Magnetic resonance imaging of the lumbosacral area was suggested for CRS evaluation. Magnetic resonance imaging was also reported to end at the T8 level of the medulla spinalis (Fig. 2b). The case was diagnosed with CRS after clinical and imaging findings were evaluated. The current treatment of the patient was carried out together with pediatric neurology, neurosurgery and physical therapy clinics. Colostomy care and physical exercise training were carried out by the family.

**DISCUSSION**

CRS was first identified by Duhamel. CRS is a developmental abnormality of the caudal vertebra, neural tube, urogenital system, gastrointestinal system and lower limbs originated from the caudal canal during the intrauterine period. Various types of anorectal malformations, spinal segment agenesis, multiple visceral abnormalities and musculoskeletal system abnormalities may be detected. The incidence of this condition is 1 per 60,000 births with a male/female ratio of 2.7/1. The risk of recurrence is quite low.

Although the aetiology and pathogenic mechanism could not be clarified, maternal diabetes, genetic predisposition, and vascular hypoperfusion are considered as possible causes. There is evidence that gestational diabetes may contribute to CRS. Experimental studies demonstrated that maternal use of retinoic acid, minoxidil and trimethoprim-sulphamethoxasole caused CRS. Chromosome analyses were found normal beyond some exemptions.

Intellectual functions are usually unaffected. Such patients would have a normal life except problems on the lower limb and neuromuscular deficit of the urinary bladder sphincter. The prognosis is poor in much-affected patients. Colostomy procedure was decided in our case due to the clinical presentation of anal atresia concomitant with urethral fistula.

The abnormalities detected in this syndrome include pes equinovarus and calcaneovarus, flexion contracture on the hips and knees, hip dislocation, agenesis of the sacrum and/or coccyx, agenesis of the fibula, rib fusion, syndactyly, polydactyly, cleft lip, cleft palate, tracheoesophageal fistula, abdominal wall defect, inguinal hernia, duodenal atresia, imperforate anus, renal agenesis or dysplasia, hydronephrosis, transposition of the external genitalia, hypoplasia, microphthalmia, hydrocephalus, meningomyelocele, partial agenesis of the corpus callosum and partial lobar holoprosencephaly.

Our case had anal atresia, flexion contracture of the hip and knees, sacral and coccygeal agenesis, external rotation of the right lower limb and left pes equinovarus. The case was diagnosed with CRS due to the findings obtained.

The expected life period is long in such cases if vital organs are not affected. Prenatal diagnosis may be established by sudden spinal interruption due to vertebral agenesis and frog leg-like position of lower limbs in the ultrasound scan at 20th gestational week. However, the cases with severe CRS (sirenomelic fetus) may be diagnosed with transvaginal ultrasound at 16th to 19th gestational weeks.

The present case with CRS was presented to draw attention to the prenatal diagnosis of the syndrome and to show the existence of this syndrome in the baby of a mother with hypothyroidism. This syndrome may be diagnosed by prenatal diagnosis, and pathological pregnancy may be terminated.

**Disclosures**

**Informed Consent:** Written informed consent was obtained from the patient’s family to publish the case report and accompanying images.
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REFERENCES