Case Report

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Sitting Buddha position: Sacral agenesis case

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ABSTRACT
Sacral agenesis syndrome (Caudal regression syndrome) is a neural tube defect that is characterized by absence of the vertebral segment that constitutes the sacrum. It is very rarely seen and generally develops sporadically. Its etiology is influenced by maternal diabetes, genetic factors, teratogenic agents and vascular hypo-perfusion. It is important to make a diagnosis in the prenatal period. This paper presents a newborn diagnosed with sacral agenesis as a case and discusses this disease in the light of the latest literature information.

INTRODUCTION

Sacral agenesis syndrome (Caudal regression syndrome) is a neural tube defect that is characterized by the absence of vertebral segment that forms the sacrum and it constitutes a rarely seen congenital and heterogeneous group of diseases (1). It generally develops sporadically and it also has autosomal-dominant inheritance family types. For its etiology, several reasons genetic factors, maternal diabetes, teratogenic agents such as retinoic acid and vascular hypo-perfusion are blamed (2, 3). The diagnosis may be made in the prenatal period. The disease may be accompanied by neurological, orthopedic, genitourinary, gastrointestinal, cardiac and respiratory problems at varying degrees per the abnormality level. This paper presents a newborn diagnosed with sacral agenesis as a case and discusses this rarely seen disease in the light of the latest literature information.

CASE REPORT

A male patient delivered by a 23-year-old mother, who was not regularly followed up during her pregnancy and did not have any chronic diseases, at week 38 and with a

birth-weight of 2100 g through C-section was hospitalized at the newborn service since deformity was observed in his legs. It was learned that his mother and father did not have kin marriage, he had three healthy and living siblings, his relatives did not have any similar diseases and his physical examination showed a good overall condition. His body temperature was measured at 36.6 C°, heart rate 114 beat/minutes, blood pressure 57/26 mmHg, oxygen saturation in room atmosphere 97% and respiratory count 48/min. The patient was identified to have pes equinovarus, bilateral internal rotation at the calcaneus, varus and club deformity. His neurological and genital examinations as well as other system examinations showed normal results as assessed. Cranial and abdominal ultrasonography and echocardiography examinations performed to detect any additional abnormalities gave normal results. The patient was observed to have a sitting Buddha position in his direct x-ray examination (Figure 1) and his lumbosacral MRI evaluation was performed. Sections were obtained in the T1A and T2A sagittal as well as T2A axial plan, cystic expansion in the spinal canal between L2-L5 at the lumbar site and sudden interruption at the level of L2 in the spinal cord were observed. Conus medullaris and sacral vertebrae were not observed and the signs were assessed to be consistent with sacral agenesis (Figure 2). Meningomyelocele was not detected. The orthopedics department was consulted and the patient's lower extremities were encased in plaster. His diet, weight gain, urinary and stool outputs were normal, so he was discharged with his outpatient follow-up planned.

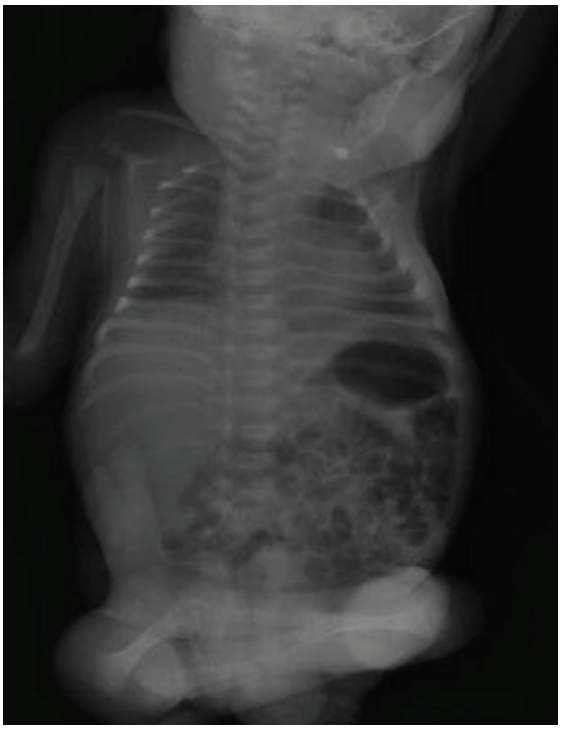

Figure 1: *The antero-posterior radiography showed the 'sitting Buddha-like position', that was the result of the flexion-abduction of the hip joints, flexion of the knees, and equinocavovarus deformity.*

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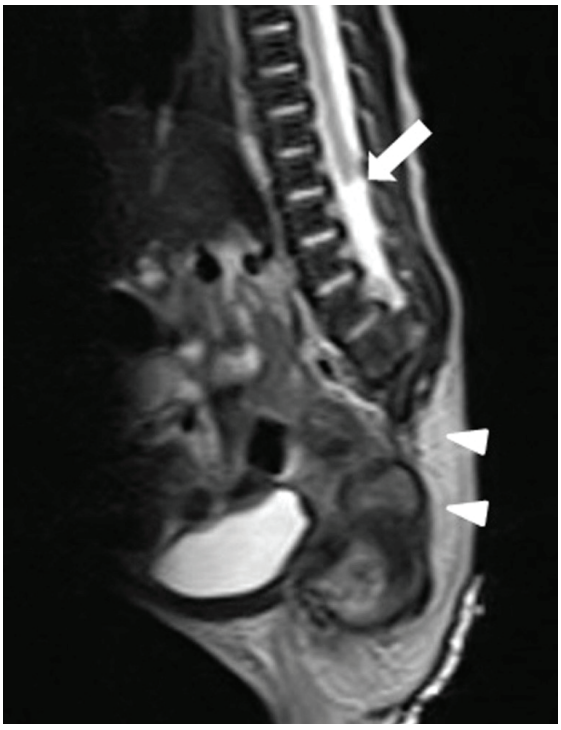

Figure 2: *MRI scan of the sagittal plan. The conus medullaris was absent and the spinal cord was ending abruptly at the level of the L2 vertebrae corpus (thin arrow). The sacral vertebrae were also absent (thick arrow).*

DISCUSSION

Sacral agenesis syndrome is seen at a frequency of approximately 0.1-0.2 in every 10000 normal pregnancies (2). It is seen 200 times more especially in the infants born of diabetic mothers; however, approximately one fifth of sacral agenesis patients have history of maternal diabetes (4, 5). Even though many factors such as genetic factors, teratogenic agents and vascular hypo-perfusion are blamed, the pathological mechanism of disease is still not entirely known (3, 4). In a case report presented in the literature, it was stated that sacral agenesis had developed in the infant of a non-diabetic mother who took minoxidil and trimethoprim-sulfamethoxazole during her pregnancy and these agents might have caused this disease (6, 7). Lower extremities, lumbar and coccygeal vertebrae and related segments of the spinal cord may be involved. Patients may be observed to have neurological sequelae such as paresis, sensation disorder, neurogenic bladder, urinary and fecal incontinence and orthopedic, gastro-intestinal, genitourinary, respiratory and cardiac abnormalities at varying degrees of severity, as well (2, 3). Imperforate anus, duodenal atresia, tracheoesophageal fistula, genital abnormalities, inguinal and diaphragmatic hernia, hydrocephaly, cleft palate and cleft lip, renal dysplasia and agenesis, hydronephrosis and congenital heart diseases may be seen (2, 5). It was learned that our patient's mother did not have diabetes or other chronic diseases, did not use any medicines or substances during her pregnancy, was not exposed to radiation and none of his relatives had been born with a similar disease.

Depending on the location and degree or vertebral agenesis, several orthopedic deformities may emerge. They include flexion contracture at the hip and knee, hip dislocation, narrow pelvis, pelvic tilt, lack of flexion and abduction in the hip, pes equinovarus, frog leg position as a result of fusion and extreme level of external rotation in lower extremities (sitting Buddha position), lack of sacrum, coccyx and/or fibula, scoliosis, polydactyly and syndactyly, inter alia. These mentioned bone deformities may be seen in direct radiographies; as necessary, patients may also be evaluated via computerized tomography or MRI, as well (5, 8, 9). Our patient had sitting Buddha and frog leg position due to deformities in the lower extremity and sacral agenesis syndrome was diagnosed on the basis of these signs.

The first and primary action for the management of this syndrome is to make an accurate and timely prenatal diagnosis. It is important to carefully examine especially patients with diabetic mothers and delivery history with abnormalities at antenatal diagnostic centers. The diagnosis is made based on the observation of lack of vertebrate in the ultrasonographic examination, sudden interruption of the spine and frog leg position in lower extremities in Week 20 of gestation (4, 5). The family needs to be informed about potential sequelae and preparation should be made to assess the patient immediately after birth in consultation with several departments including pediatric surgery, neurosurgery, orthopedics, urology, nephrology, gastroenterology and cardiology (2).

Respiratory problems are frequently seen in infants with diabetic mothers and patients with thorax deformity due to surfactant deficiency and some patients may require long-term mechanical ventilator support. The patients should also undergo echocardiographic examination for congenital heart diseases and ultrasonographic evaluation for nephrological malformations, as well. Caution should also be taken with respect to recurrent urinary infections, renal failure and gastrointestinal abnormalities (2, 10). It was identified that our patient did not have congenital abnormalities or respiratory trouble related to cardiac, nephrological and gastrointestinal system, the patient's diet and urinary and stool outputs were normal.

No definitive treatments are yet present for this disease; supportive treatments are performed in a multidisciplinary way. If there is no involvement of vital systems, the lifetime is long, intelligence has not been affected; however, patients require medical care throughout their life (5). The patients need to receive neurological, orthopedic, nephrological and urinary system interventional procedures and psychological support should be given to the family. It is important not to delay the revision surgeries and rehabilitation of patients diagnosed in the early period following birth with respect to the quality of life (9).

In conclusion, it is important to make a diagnosis of the sacral agenesis syndrome, which is rarely seen and does not have a definitive treatment, in the prenatal period and to inform the family for termination of pregnancy. Supportive treatments are provided by several departments for infants born with this disease.

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