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.

Keywords: Sacral agenesis; Caudal regression syndrome; Newborn.

INTRODUCTION
Sacral agenesis syndrome (Caudal regression syndrome) is a neural tube defect that is characterized by absence of the 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.
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 (3). 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.
REFERENCES