

Case Report

An Incidentally Detected Sacral Agenesis and Associated Anomalies: Case Report

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ABSTRACT

Sacral agenesis, being a part of caudal regression syndrome, is a congenital spinal defect with an unknown etiopathogenesis, consisting of total or partial underdevelopment of sacrum and tethered cord, and can be accompanied by lumbar spinal defects and neurologic problems. It is thought to be caused by a developmental defect, occurred at the early phases of gestation. Filum terminale is tethered due to adhesions and as a result of this, the genitourinary system in the first place, and then the musculoskeletal system and gastrointestinal system can also be affected particularly. Co-existing cardiac and respiratory defects can be found. The morbidity level can be determined by the level of sacrum's agenesis and accompanying dysfunction degree of the organs and systems. In our article, we have presented the case that was brought by the family to the pediatrics clinic with the symptoms of tethered filum terminale, recurrent urinary system infection and intractable constipation.

Keywords: Sacral Agenesis, Neurogenic Bladder, Caudal Regression Syndrome, Hydronephrosis.

ÖZET

İnsidental Olarak Saptanan Sakral Ajenezi ve Eşlik Eden Anomaliler: Olgu Sunumu

Sakral ajenezi, kaudal regresyon sendromunun bir parçası olarak lomber omurga defektlerinin de eşlik edebileceği, sakrumun kısmen ya da tamamen olmadığı, gergin omurlilik ile birlikte nörolojik sorunlar ile seyreden ve etiopatolojisi kesin olarak ortaya konulamamış konjenital spinal bir defektir. Gestasyonun erken dönemlerinde oluşan gelişimsel bir hatadan kaynaklandığı düşünülmektedir. Yapışıklık nedeni ile girilen filum terminale ve buna bağlı olarak genitouriner sistem başta olmak üzere, kas-iskelet sistemi ve gastrointestinal sistem özellikle etkilenebilir. Birlikte kardiyak ve respiratuar defektler bulunabilir. Morbidite düzeyini; sakrumun ajenezi seviyesi ve eşlik eden organ ve sistemlerin disfonksiyon derecesi belirler. Biz yazımızda gergin filum terminale ile birlikte rekürren idrar yolu enfeksiyonu ve tedaviye yanıt vermeyen dirençli kabızlık şikayetleri ile pediatri kliniğine ailesi tarafından getirilen olguyu sunduk.

Anahtar Sözcükler: Sakral Ajenezi, Nörojenik Mesane, Kaudal Regresyon Sendromu, Hidronefroz.

Sacral agenesis is a congenital spinal anomaly, which is a component of caudal regression syndrome where the complete or partial absence of sacral spine can be accompanied by neurologic, urologic, orthopedic pathologies and intra-abdominal and thoracic organ defects (1). Due to an embryological developmental defect of cloaca in intrauterine period, sacral agenesis consists of anomalies of caudal vertebra where caudal canal, neural tube, genito-urinary and intestinal systems, and lower extremities are originated. Generally, it can be diagnosed in intrauterine life, but the unnoticed cases may apply to pediatrics clinics with various clinical findings. Its approximate incidence is between 0.1-0.25/10.000 (2). While most of the cases are associated with maternal diabetes, in some cases, there can also be a genetic disposition (2, 3). Renal ectopia and agenesis, imperforate anus, anorectal atresia, neural

tube defects such as; tethered cord, and diastematomyelia and orthopedic disorders such as; hip dislocation, articular contractures and club-foot can co-exist with the illness (4). The diagnosis of the accompanying pathologies at an early phase is important in terms of treatment at an early stage. In this article, we have presented the case who were brought to our clinic with the symptoms of recurrent urinary system infection and intractable constipation and is characterized with the absence of 3rd to 5th sacrum vertebrae.

CASE REPORT

One-year-old girl who was brought to our clinic for resistant constipation and recurrent urinary system infection was in generally good health. Her neuromotor development was behind her coevals. She could sit without any support, but she was not crawling, and walking. Deep tendon reflex was normoactive and

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anocutaneous reflex was negative. Urination was thought to be abnormal as an overflow urinary incontinence was present. Gluteal flattening and disappearance of the gluteal cleft were found (Figure 1).



Figure 1: Sacral flattening and low gluteal cleft

There were no other pathologies as a result of the examination of other organs and systems. There was not diabetes or gestational diabetes in the patient's mother and there was not a history of drug usage, serious infection or exposure to X-ray during the pregnancy. The family history of sacral agenesis was not present. There was a consanguineous marriage of second-degree cousins between the parents. No pathology was diagnosed in the antenatal follow-ups during the pregnancy. At the magnetic resonance imaging (MRI) for the spinal canal of the patient, sacral 3-5 vertebrae were not observed. It was detected that conus medullaris ended in a blunt-sharp way at the level of thoracic 12 vertebra and a coarse filum terminale was tethered because of an adhesion at the level of lumbar 5 vertebra. In T1-T2 weighted magnetic resonance imaging, a 8x4 hypointense, irregular limited solid bulk was detected in filum terminale (Figure 2a, b).



Figure 2: (a) Solid mass at the filum terminale (b) Disrupted spinal cord at the level of T12 vertebrae

No anomaly was detected at the middle and lower thoracic, lumbar vertebra corpus and at its posterior side that can be screened in the imaging area. In the cervical MRI, there was not any pathology. In the abdominal ultrasonography and MRI, there was Grade-2 hydronephrosis in the right kidney (Figure 3a, b).

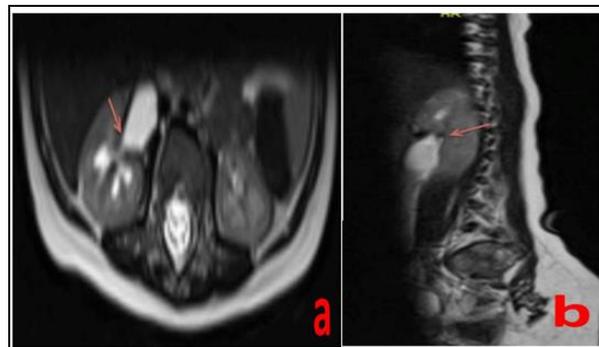


Figure 3: (a, b) Right kidney, grade 2 hydronephrosis.

Although voiding cysto ureterogram (VCUG) of patient did not reveal any vesicoureteral reflux in both kidneys; a small and trabeculated bladder was found in VCUG. Maximum detrusor pressure was 54 cmH₂O, which is in normal range but bladder capacity was low at about 40 cc in the urodynamic study. Post-micturition residual urine was high, at about 35 cc (Figure 4).

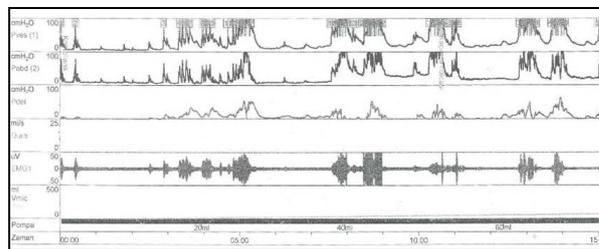


Figure 4: Urodynamic test result: Normal detrusor pressure and low bladder capacity.

The family was informed about the urgency of the surgical treatment of the tethered filum terminale, and the patient was referred to a higher-level of care hospital. The patient was given prophylactic antibiotic treatment for recurrent urinary system infection and regular disimpaction for the constipation.

DISCUSSION

Caudal regression syndrome includes malformations ranging from the absence of coccyx to the absence of lumbar vertebra and sometimes can be together with neurologic, urogenital, gastrointestinal and orthopedic anomalies (5, 6). The clinical symptom spectrums of the patients generally originate from the level of the vertebra defects (7). In the etiology, maternal diabetes, usage of minoxidil or retinoic acid during the pregnancy and genetic predisposition are the factors that are blamed (2, 8). In our case, there was no history of maternal diabetes.

Congenital anomalies of neural tube structures developed from caudal canal during embryological period such as caudal vertebra and urogenital system and lower extremities constitute the caudal regression syndrome (9). Caudal regression syndrome can be divided into two groups (10). In Group 1, spinal cord ends abruptly at above the first lumbar vertebra level and symptoms occur due to tethered filum terminale, usually caused by adhesions in the lower lumbar region and a lipoma may accompany. In Group 2, the conus ends below the normal level and a thick filum terminale and/or lipoma can accompany. While neurologic symptoms are heavier in Group 2, sacral deformities are more serious in Group 1. In our case conus was above at T-12 level and filum terminale was tethered due to adherence to L5 vertebra. Because of the concomitant embryological development of lower vertebra, colon and urinary system, we think that developmental defects of the kidneys, neurogenic bladder and constipation problems in our case are due to adhesions caused by tethered filum terminale. Our case was in concordance with the Group 1 caudal regression syndrome. Urinary and fecal incontinence were present and sacral deformity was at a severe level. In caudal regression system, besides the sacral agenesis level, tethered and adherent cord should be investigated and genitourinary system, musculoskeletal system and neurological system should be examined carefully (1, 5, 9). The clinical symptoms of pes equinovarus, contractures of hip and knee and congenital hip dislocation which cause orthopedic problems, symptoms of renal agenesis, hydro-nephrosis, hypospadias which cause urogenital problems, symptoms of hydrocephaly, meningomyelocele, tethered spinal cord and diastematomyelia which cause neurological problems should be examined carefully (9, 11). Especially in cases with tethered filum terminale caused by adhesions, as in our case, surgical treatment should be considered in no time. Parents must be informed that the patient will benefit from the surgery. In cases with neurogenic bladder and are diagnosed at a later stage, morbidities such as; hydro-nephrosis, vesicoureteral reflux and recurrent urinary system infection in an irreversible stage were reported (1). We also detected partial sacral agenesis, tethered filum terminale and related neurogenic bladder that form the basis of recurrent urinary system infection and constipation, which was resistant to the treatment. The family was informed about the urgency of the surgical treatment of the tethered filum terminale and the patient was transferred to a higher-level of care hospital. The patient was given prophylactic antibiotic treatment for the recurrent urinary system infection and regular disimpaction for the constipation.

For radiologic diagnosis, ultrasonography is the first option for infants. Other investigations that help to establish a diagnosis are computed tomography and magnetic resonance imaging (5). In sacral agenesis, the defects in the bones can be seen in the direct X-ray imaging. Detailed investigation in 3-D can be made via the images taken with computed tomography. The diffusion-weighted MRI is the golden standard to affirm the findings of the ultrasonography and to find additional anomalies. The images of the sagittal plane are important and especially the deficiencies in the lumbosacral vertebral structures, distal spinal cord and cord tension are evaluated. On the axial plane, spinal canal stenosis, diastematomyelia, the existence of hydromyelia and other related lesions are evaluated (9). We made the sacral agenesis diagnosis of our case with the help of direct x-ray graphy and diffusion-weighted MRI and detected that the conus was ended at T12 level, and filum terminale was tethered due to the adherence to L5 vertebra. We did not detect any additional pathology in the spinal cord at cervical and thoracic diffusion-weighted MRI.

The cases of sacral agenesis together with the other systematic problems create economic and social problems in the society. The detection of the cases with sacral agenesis during the pregnancy is possible at the 18th-22nd week by the ultrasonography of the mothers, especially diabetics, via the detection of the early ending of medulla spinalis in the spine and frog leg posture of the lower extremities of fetus (11). It is proper to end the pregnancy through talking to the family during the prenatal follow-ups. In the cases that are given birth to, there should also be cooperation with the family and by giving detailed information the patient should be evaluated in a multidisciplinary way, which orthopedists, neurosurgery specialists, urologists, physiatrists and pediatric surgery specialists attend. In the presence of tethered filum terminale due to adhesions, patient must be evaluated considering the aspects of clinical and radiological dynamics and surgical treatment should be offered to patients.

Sacral agenesis should be considered in the babies of whom sacral dimple, gluteal line abnormality and gluteal flattening, recurrent urinary system infection and constipation are detected in the physical examination during neonatal period and surgical treatment should be offered to these patients. The severity of the pathology should be examined via spinal diffusion-weighted MRI, VCUG, antenatal and postnatal ultrasonography, urodynamic tests. We think that both with early surgical treatment of tethered filum terminale and associated anomalies by the related clinical branches and with the help of rehabilitation, quality of life can be increased significantly.

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