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A RARE CONGENITAL ANOMALY: SPLIT NOTOCHORD SYNDROME WITH RECTAL DUPLICATION AND UTERUS DIDELPHUS

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ABSTRACT:
Split notochord syndrome (SNS) is an extremely rare spinal anomaly, associated with anomalies of the vertebrae, central nervous system, and gastrointestinal tract. So far less than 30 neonatal cases with SNS have been published in the literature. Duplications of the gastrointestinal tract are rarely seen and rectal duplications (RDs) account for 5% of alimentary tract duplications. This case of SNS was associated with RD, posterior stenotic anus, meningomyelocele, Arnold Chiari malformation, hemivertebrae, short neck, and pes equinovarus. Postmortem investigations have also revealed uterus didelphus and bilateral hypoplastic kidneys. This case was presented to highlight the fact that there are rare associations with SNS, together with a literature review.
Key words: Split Notochord Syndrome, Rectal Duplication

NADİR BİR KONJENİTAL ANOMALİ: SPLİT NOTOKORD SENDROMU, REKTAL DUPLİKASYON VE UTERUS DİDELFİS BİRLİKTELİĞİ
ÖZ:
Split notokord sendromu (SNS) nadir bir spinal anomali olup, vertebra, santral sinir sistemi ve gastrointestinal sistem anomalilerine eşlik eder. Literatürde 30'dan az yenidoğan olgusu yayınlanmıştır. Gastrointestinal sistemin duplikasyonları nadirdir ve rektal duplikasyonlar tüm duplikasyonların %5'ini oluşturur. Split notokord sendromlu bu olguda, rektal duplikasyon, posterior stenotik anüs, meningomyelosel, Arnold Chiari malformasyonu, hemivertebra, kısa boyun ve pes ekinovarus bulunmaktadır. Postmortem incelemeler ile uterus didelfis ve bilateral hipoplastik böbrekler saptandı. Bu olgu, Split Notokord sendromuna nadir anomalilerin eşlik etmesi nedeniyle tüm literatür gözden geçirilerek sunuldu.
Anahtar Kelimeler Split Notokord Sendromu, Rektal Duplikasyon

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INTRODUCTION

Alimentary tract duplications are rare congenital malformations. Although the lesions can occur at any site of the gastrointestinal tract, only 5% of them occur in the rectum. Of the approximately 100 reports of rectal duplication (RD) in the literature, the majority are located in the retrorectal space.[1,2] Associated congenital abnormalities occur in up to 50% of cases.[3] Split notochord syndrome (SNS), a rare congenital anomaly, is simply defined as a form of spinal dysraphism in which there is a persistent communication between ectoderm and endoderm. The defect takes place during the 2nd and 3rd weeks of embryogenesis when the notochordal plate is formed. Although the etiology is unclear, the pathogenesis is explained as anomalous splitting and disturbed separation of the notochord from the endoderm and primitive gut, which normally should take place during development, resulting in different levels of spinal and gastrointestinal anomalies.[4] A case of SNS and RD associated with urinary and vertebral anomalies is presented here.

CASE REPORT

A 1-day-old female neonate was admitted to Gazi University Hospital Neonatal Intensive Care Unit (NICU) with thoracolumbar meningomyelocele. The mother was 18 years old, gravida 1, and had received no antenatal care. The neonate, weighing 2600 g, was delivered by vaginal delivery in a community hospital at term gestation and was transferred after the defect was noticed. Antenatal history was unremarkable for drug ingestion or health problems; however, no antenatal care was provided. There was no consanguinity between the parents and no history of children with neural tube defects in the family. The infant had required no resuscitation and the transfer took place without problems. On admission she was noted to have enlarged anterior and posterior fontanelles (6 x 6 cm, 5 x 5 cm, respectively), anteverted nostrils, a small mouth, a short neck, covered meningomyelocele about 5 cm in diameter at the thoracolumbar region between thoracal 10 and lumbar 2 vertabrae, pes equinovarus, and lack of movements of the lower limbs. Stool passage was present despite the stenotic and posteriorly placed anus. The external genitalia and urethral meatus were normally placed. Laboratory findings revealed normal complete blood cell count, biochemistry, and urine analysis. However, urine output was borderline, with 1 cc/kg/h. Echocardiography was normal. However, vertebral X-ray showed cervical hemivertebrae, vertebral fusion anomalies in the thoracal region between 7 and 12 thoracal vertebrae, and lumbar hemivertebrae. Cranial ultrasound revealed hydrocephalus, Arnold Chiari type II malformation, and cranial magnetic resonance imaging (MRI) showed a dilated foramen magnum, cerebellar tonsillar herniation, hypoplasia of the cerebellar hemispheres, lateral intraventricular hemorrhage and dilatation, and parenchymal hemorrhage. Abdominal ultrasound showed bilateral hypoplastic kidneys (right kidney 32 x 14 mm, left kidney 29 x 19 mm). The decision was made to repair

the meningomelocele first and she was operated on to close the meningomyelocele and place a ventriculoperioneal shunt on the 2nd and 3rd days of life, respectively. However, the child could not get off the ventilator post-surgery because of severe apnea, during that time urine output diminished further to 0.5 cc/kg/h, with elevated renal function tests. She died on the 11th day of life due to cerebellar herniation. The autopsy revealed the following additional findings: uterus didelphis (double uterus, double cervix, vaginal septum)(Figure 1), multiple bilateral ovarian cysts, severely hypoplastic kidneys (23 g), and blind rectal duplication 3 x 0.5 x 0.5 cm in the retrorectal region with no connection to the lumen. Based on the autopsy and laboratory findings the diagnosis of SNS with RD was made.

Figure 1: Autopsy photograph: Uterus didelphus

DISCUSSION

Duplication anomalies of the gastrointestinal tract may occur anywhere from the mouth to the anus. Associated congenital abnormalities occur in up to 50% of cases and include genitourinary anomalies, vertebral defects, and rarely cardiac malformations. [3] Less than 100 cases of RD have been reported in the literature. The causes of enteric duplications include incomplete separation of the notochord from the endodermal tube, defects of recanalization, caudal twining, and persistent epithelial buds.[5] A rectal duplication can manifest as fistulization, infection within the duplication, symptoms caused by the mass effect, ectopic mucosa, or malignant degeneration. The differential diagnosis of rectal duplication must include tail gut cysts, which have a potential relationship with adenocarcinoma formation, teratomas, dermoid cysts, and meningocele.

Duplicated bowel is rarely associated with SNS. The spectrum of SNS includes a wide variety of congenital anomalies of the spine, the nervous system, and the gastrointestinal tract. A brief review of the literature is presented in Tablo 1. During the third week of gestation, the human embryo consists of 3 embryological layers: ectoderm, mesoderm, and endoderm. By day 20, the notochordal process has appeared as a tube in the mesodermal layer. The ventral wall of the notochordal process then begins to fuse with the endoderm to form the notochordal plate. During a brief period, an open neuroenteric canal is formed between the yolk sac cavity and the amniotic cavity. The final remnants of this canal are located at the tip of the coccyx. Then the notochordal process creates the notochord. Somitomeres appear in the paraxial mesoderm, which form sclerotomes. These eventually give rise to the vertebral bodies, vertebral arches, and part of the back of the skull. During the fourth week, the neural plate converts into the neural tube. Inadequate closure of the neural tube can result in vertebral anomalies. SNS includes a cleft of vertebral column associated with gastrointestinal anomalies mostly dorsal enteric fistula and central nervous system anomalies. So far less than 30 neonatal cases have been reported in the English literature with a wide spectrum of anomalies including spina bifida, butterfly vertebrae, diastematomyelia/diplomyelia, meningomyelocele, sacral agenesis, dorsal enteric fistula, imperforated anus, intestinal duplications, enteric cysts, and talipes equinovarus.[4,6-14,15-23]

RD should be considered in patients with neural tube defects, vertebral anomalies, and gastrointestinal malformations. More than 50% of patients with RD also have associated anomalies. The patient is presented as a rare example of a rare anomaly and also to emphasize the importance of a complete evaluation of the patient with major congenital anomalies and the importance of autopsy.

Early diagnosis of gastrointestinal and genitourinary anomalies may be helpful both in decision making for treatment and also for prevention of potential complications.

Author, year	Age, sex	Anomalies	Treatment	Prognosis
Van Ramshorst, 2006[23]	4 d, M	Agenesis of corpus callosum, epispadias, intestinal malrotation	Enteroplasty, resection of the neuroenteric fistula	Alive
Hishiki, 2006[13]	Neonate, F	Thoracoabdominal duplication congenital diaphragmatic hernia, portoazygous shunt	Repair of diaphragmatic hernia	Dead
Agangi, 2005[4]	Neonate, M	Lipomyelomeningocele, tethered cord, rectourethral fistula	Colostomy, anorectoplasty, lipomyelomeningocele treatment	Alive
Dindar, 1999[9]	Neonate, M	Meningomyelocele, dorsal enteric fistula, wandering spleen	Colostomy, meningomyelocele treatment	Alive
Kiristioglu, 1998[15]	2 h, M	Hydrocephalus, meningocele dorsal enteric fistula, a foreshortened colon	Hydrocephalus and meningomyelocele treatment, anorectal malformation correction	Alive
Akgur, 1998[7]	Neonate, M	Meningomyelocele, hydrocephalus congenital clubfoot, dorsal enteric fistula	Fistulectomy, intestinal anastomosis, meningomyelocele treatment	Dead
Fowler , 1998[11]	Neonate, F	Ompholocele, rib and vertebral anomalies, colonic duplication, bicornuate uterus, tethered cord	Ileostomy, duplication resection	Alive
Razack, 1995[19]	8 d, F	Hydrocephalus, encephalocele, dorsal enteric fistula, teratoma, congenital clubfoot	Colostomy, teratoma resection, hydrocephakus and meningomyelocele treatment	Alive
Hoffman, 1993[14]	Neonate,	Hydrocephalus, cloacal extrophy, dorsal enteric fistula	None	Dead
Meller, 1989[18]	Neonate, F	Hydrocephalus, cecum duplication, malrotation, dorsal enteric fistula, anteriorly displaced anus, equinovarus	Fistulectomy, cecostomy, hydrocephalus treatment	Alive
Kramer, 1988[17]	Neonate, M	Hydrocephalus, dorsal enteric fistula, meningomyelocele, sacral agenesis	None	Dead
Gupta, 1987[12]	16 d, M	Meningomyelocele, dorsal enteric fistula, hydrocephalus	Meningomyelocele treatment, fistulectomy	Alive
Kheradpir, 1983[16]	Neonate, M	Hydrocephalus, dorsal enteric fistula, meningomyelocele, anal atresia	None	Dead
Singh, 1982[18]	2d, M	Hydrocephalus, ear anomaly, dorsal enteric fistula	No	Death
Singh, 1982[22]	2 d, M	Hydrocephalus, ear anomaly, dorsal enteric fistula	None	Dead
Faris, 1975[10]	1 d, M	Meningomyelocele, hydrocephalus, dorsal enteric fistula, hypospadias, microcolon	Meningomyelocele treatment, colectomy, ileostomy	Dead
Bentley, 1960[8]	4 d, F	Dorsal enteric fistula	None	Dead
Rosselet, 1955[20]	1 d, M	Meningomyelocele, hydrocephalus, ambiguous genitalia	None	Dead
Saunders, 1943[21]	2 d, F	Hydrocephalus, agenesis transverse colon, malrotation, dorsal enteric fistula	None	Dead

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