0021-7557/04/80-01/77
Jornal de Pediatria

A rare variant of neuroenteric cyst: split notochord syndrome

Lisieux E. Jesus[1], Cristiano G. França[2]

Abstract

Objective: We present a case of split notochord syndrome, an extremely rare form of spinal dysraphism.

Description: We treated a 2 month-old boy presenting with an extensive lumbosacral deformity, hydrocephalus and apparent enteric segments in the dorsal midline, accompanied by an enteric fistula and imperforated anus. The malformation was diagnosed as split notochord syndrome. The baby died as a result of sepsis before surgical treatment could be attempted.

Comments: Split notochord syndrome is the rarest form of neuroenteric cyst described until this moment (< 25 cases in the literature). It is frequently associated with anorectal malformation, intestinal fistulae and hydrocephalus. Prognosis is not necessarily poor and survival is possible if digestive malformations, hydrocephalus and the dysraphism itself are treated simultaneously.

J Pediatr (Rio J). 2004;80(1):77-80: Split notochord syndrome, spinal dysraphism, myelodysplasia, neuroenteric cyst.

Introduction

We describe a case of split notochord syndrome (SNS), which is a rare and usually unknown neural tube malformation. An extensive search using both MEDLINE and LILACS revealed that up to now only 25 cases have been described in the literature. To our knowledge, this is the first case described in Brazil and the second one in Latin America: Baeza–Guerrera et al.[1] mention two cases in Mexico, but the second case has some characteristics that suggest incomplete twinning. These characteristics are often arguable with respect to mature forms of teratoma.

1. Pediatric surgeon, Service of Pediatric Surgery, Hospital Municipal Jesus and Hospital Universitário Antônio Pedro, Universidade Federal Fluminense, Rio de Janeiro, RJ, Brazil.
2. Neurosurgeon, Service of Pediatric Neurosurgery, Hospital Municipal Jesus, Rio de Janeiro, RJ, Brazil.

Manuscript received 30.07.03, accepted for publication 30.09.03.

Patient care followed the guidelines established by the Research and Ethics Committee of Hospital Municipal Jesus.

Case Report

A 2-month-old male patient weighing 1,800g, born to a teenage mother (14 years old), was admitted to Hospital Municipal Jesus, Rio de Janeiro. The mother, who received no prenatal care, reported having vaginal discharge during pregnancy, which was not treated, and urinary tract infection. She denied the use of any drugs or exposure to teratogenic agents. The child has necessitated total parental nutrition since birth, had recurrent episodes of sepsis, and was submitted to various antibiotic treatments during his stay in the intensive care unit.

On admission, the patient was jaundiced and underfed, presenting anorectal malformation (imperforated anus without perineal or urinary fistula) and diastasis of the posterior midline in the thoracolumbar region of the spine with extensive palpable bone deformity on lesion margins. No clinical signs of intestinal obstruction were observed, but the patient showed various small intestinal loops coming through the thoracolumbar deformity, with formation of a spontaneous intestinal stoma through which intestinal fluids were discharged (Figure 1). There was no leakage of cerebrospinal fluid or visible exposure of neural tissue elements. The patient was awake and responsive to environmental stimuli. He could move his lower limbs, but presented with hypoesthesia and paresis (predominantly distal), muscle atrophy, and reduction of tendon reflexes. Neither urinary retention or dribbling nor a palpable bladder was described. Macrocephaly and enlarged fontanelles, which indicate hydrocephalus, were noted.

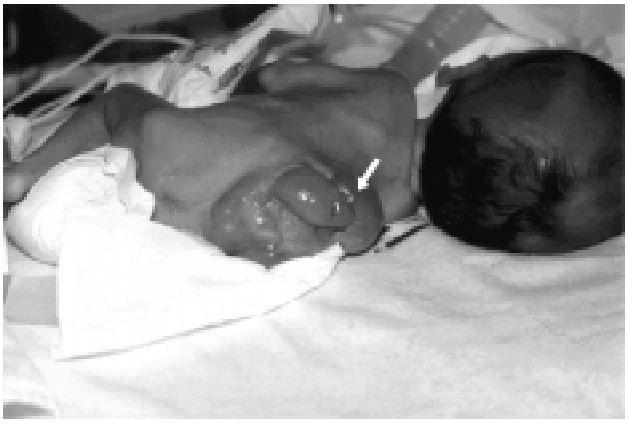

Figure 1 - Pacient's dorsal region. Note the intestinal loops coming through the thoracolumbar deformity, with enteric fistula (arrow) and hydrocephalus.

Chest and abdominal CT scans showed extensive vertebral fusion anomalies in the lumbar region, extending from the 11th thoracic vertebra to the sacral spine. The bone deformity was freely permeated by intestinal loops. The cranial ultrasound showed type II Arnold Chiari malformation, hydrocephalus, and cerebral cortex abnormal development.

The diagnosis of SNS, severe hydrocephalus, anorectal malformation and spontaneous congenital intestinal fistula was initially considered. Abdominal nuclear magnetic resonance and fistulography were considered for surgical planning, but the patient developed sepsis and died after three days in hospital. Necropsy could not be performed because the family did not authorize it.

Discussion

The SNS, as proposed by Bentley and Smith,[2] (also known as posterior *spina bifida*, combined *spina bifida*, neurenteric fistula, dorsal enteric fistula) is an extremely rare form of dysraphism (less than 25 cases have been described in the literature so far). It was first described by Rembe in 1887.[3] In this syndrome, vertebral anomalies (anterior and posterior *spina bifida,* butterfly vertebrae), central nervous system abnormalities (diastematomyelia, diplomyelia, myelomeningocele) and intestinal anomalies (fistulas, dermal sinus tract, diverticula and enteric cysts) are associated with each other. The syndrome manifests as a cleft in the dorsal midline of the body through which intestinal segments are exteriorized (often with an associated fistula), myelomeningocele, and occasionally as a teratoma. Central nervous system abnormalities are always present: hydrocephalus and diastematomyelia/ diplomyelia are constant findings. However, babies do not necessarily present with functional spinal cord defects: in some reported cases, the motor function of the lower limbs is normal. The presence or absence of intestinal and urinary sphincter dysfunction is difficult to evaluate given the high frequency of anorectal malformations associated with the wide variation in the clinical expressions of neurogenic bladder, thus including cases without urinary retention or continuous urinary dribbling, mainly bladder-sphincter dyssynergia and some forms of detrusor overactivity, which are difficult to detect in newborns, especially if a formal urodynamic evaluation is not performed. No data regarding immediate or long-term urodynamic function were found among surviving patients.[4-7] *Talipes equinovarus* is a common finding.

The location of the intestinal fistula may vary from case to case, and may be found either in the distal ileum/cecum or in the large intestine (most of the cases). The disease affects both males and females, and there exists a high incidence of urogenital malformations (3/25 cases described in the literature) and anorectal malformations (7/25 cases described in the literature, in addition to the current case) (Table1).

In most cases, there is no history of exposure to teratogenic agents or family history of congenital defects. The embryological origin of this anomaly is discussed and its cause is still unknown. The oldest theory suggests the persistence of a primitive neurenteric canal connecting the amniotic cavity to the dorsum of the embryo in the third week of gestation. This theory is refutable on the grounds that this connection occurs in a normal embryo with the infracoccygeal region, whereas in the SNS, the vertebral deformity is constantly proximal (cervical, thoracic or lumbosacral). Some authors attribute this discrepancy to the varied positions of Hensen's node or to the presence of an accessory neurenteric canal.[16] Currently, the most widely accepted theory suggests a primary notochord defect (the notochord is split, but not completely separated from the primitive intestine), resulting in secondary changes to the paraxial mesoderm, which is responsible for the

formation of the spinal column, giving rise to a medial interosseous space. Through this space, the endoderm and the underlying primitive intestine herniate, adhere to the dorsal ectoderm, and eventually rupture.[2,9,17] This way, the SNS represents an extreme end of the spectrum of neurenteric cysts.[17-18] Extensive studies on the role of the Sonic Hedgehog gene and its possible defects in the formation of spinal dysraphism have been underway with the aim of finding a genetic etiology for the disease. The available data, in their vast majority, originate from experimental studies using mouse models manipulated through the administration of adriamycin.[19-20] The data are still preliminary, and some researchers are not sure of the essential role of this gene in the final organization of neural tube closure.[21]

A poor prognosis for survival has been described in the literature, with only four survivors being reported (Table 1), but many of these studies are obsolete, and much better results are likely to be obtained with present-day surgical technology and neonatal intensive therapy. Some authors suggest a two-step approach: correction of the intestinal lesion and the subsequent correction of spinal dysraphism. This approach prevents fecal contamination during the neurosurgical procedure; however, no clearly defined technical standard exists for the management of these patients. Appropriate and

Table 1 - Cases described in the literature

Author	Age/sex	Associated problems	Defect localization	Fistula connection	Anus	Treatment	Result
Bentley, 1960[2]	4d, F	NTM	L2-sacro	Rectum	ARMF	NP	Death
Akgur, 1998[3]	Neonate, M	MMC, HC, cengenital clubfoot	T10-L5	Cecum	Normal	Fistulectomy, intestinal anastomosis, MMC treatment	Death 7th POD, sepsis
Gupta, 1987[4]	16d, M	HC	L5-sacro	Rectum	Normal	Fistulectomy, lumboplasty	Alive
Kiristioglu, 1998[5]	2h, M	Microcolon, HC	L1-sacro	Rectum	ARMF	MMC treatment and HC, ARMF correction	Alive
Meller, 1989[6]	Neonate, F	HC, ceccum duplication, malrotation	?	Cecum	Ectopia	Fistulectomy, cecostomy, MMC treatment	Alive
Razack, 1995[7]	8d, F	Teratoma, HC, encephalocele, congenital clubfoot	L2-sacro	Rectum	Normal	Colostomy, teratoma ressection, MMC treatment and HC	Alive
Faris, 1975[8]	1d, M	HC, hypospadias, microcolon	T10-sacro	Colon	ARMF	MMC treatment, colostomy, ileostomy	Death
Saunders, 1943[9]	2d, F	Microcolon, urogenital MF, HC	L1-S2	Cecum and Rectum	Normal	NP	Death
Rosselet, 1955[10]	1d, M	Ambiguous genitalia, HC	T12-sacro	Sigmoid	Normal	NP	Death
Singh, 1982[11]	2d, M	HC, ear anomaly	T10-sacro	Colon	Normal	NP	Death 12th AD, meningitis
Kheradpir, 1983[12]	Neonate, M	HC	T11-sacro	Colon	ARMF	NP	Death
Kramer, 1988[13]	Neonate, M	HC	T10-sacro	Cecum	ARMF	NP	Death
Hoffman, 1993[14]	Neonate, F	Cloaca exstrophy, HC	T12-sacro	?	ARMF	NP	Death
Kanmaz, 2002[15]	8m, M	Inguinal herniation, syringomyelia, HC, malrotation	T11-L4	Distal descending colon	Normal	Fistulectomy and colostomy	Death 1st POD
Jesus, 2003	2m, M	HC	T11-sacro	?	ARMF	NP	Death 3rd AD, sepsis

F: female; M: male; MMC: meningomyelocele; MF: malformation; ARMF: anorectal malformation; HC: hydrocephalus; NP: not performed; NTM: nothing to mention; POD: post-operative day; AD: admission day.

detailed preoperative imaging studies, combining nuclear magnetic resonance and contrast fistulography, are of utmost importance for proper surgical planning. The constant treatment of hydrocephalus is necessary as a complementary measure.

References

1. Baeza-Gerrera C, Martinez-Aguilar G, Bravo-Becerra JM, Moralez-Velazco F. The split notochord syndrome: report of 2 cases. Bol Med Hosp Infant Mex. 1993;50(11):824-7.
2. Bentley JFR, Smith JR. Developmental posterior enteric remnants and spinal malformations. Arch Dis Child. 1960;35:76-84.
3. Akgur FM, Ozdemir T, Olguner M, Erbayrektar S, Ozer E, Aktug T. A case of split notochord syndrome: presence of dorsal enteric to the dorsal enteric fistula. J Pediatr Surg. 1998;33: 1317-9.
4. Gupta DK, Deodhar MC. Split notochord syndrome presenting with meningomyelocele and dorsal enteric fistula. J Pediatr Surg. 1987;22:382-3.
5. Kiristioglu I, Teitelbaum DH, Dogruyol H. Split notochord syndrome with prolapsed congenital colostomy. J Pediatr Surg. 1998;33:525-8.
6. Meller JL, Loeff DS, Reyes HM. Variant of the split notochord syndrome. J Pediatr Surg. 1989;24:733-5.
7. Razack N, Page LK. Split notochord syndrome: case report. Neurosurgery. 1995;37:1006-8.
8. Faris JC, Crowe JE. The split nothocord syndrome. J Pediatr Surg. 1975;10:467-72.
9. Sanders RL. Combined anterior and posterior spina bifida in a living neonatal human female. Anat Rec. 1943;87:255-78.
10. Roselet PJ. A rare case of rachischisis with multiple malformations. Am J Roentgenol Radium Ther Nucl Med. 1955;73:235-40.
11. Singh A, Singh R. Split notochord syndrome with dorsal enteric fistula. J Pediatr Surg. 1982;17:412-3.
12. Kheradpir MH, Ameri MR. Dorsal herniation of the gut with posterior opening of the terminal colon. A rare manifestation of the split notochord syndrome. Z Kinderchir. 1983;38:186-7.
13. Kramer EL, Giacoia GP, Say B, Jarolim KL, Miller-Hardy D. Split notochord syndrome with dorsal enteric fistula and sacral agenesis. Teratology. 1988;38:1-5.
14. Hoffman CH, Dietrich RB, Pais MJ. The split notochord me with dorsal enteric fistula. AJNR Am J Neuroradiol. 1993;14:622-7.
15. Kanmaz T, Demirbilek S, Ozturk A, Baykara S, Yucesan S. The split notochord syndrome with dorsal enteric fistula. Indian J Pediatr. 2002;69:729-30.
16. Bremer JL. Dorsal intestinal fistula, accessory neuroenteric canal, dyastematomielia. Arch Pathol. 1952;54:132-8.
17. Alrabeeah A, Gillis DA, Giacomantonio M, Lau H. Neuroenteric cysts – a spectrum. J Pediatr Surg. 1988, 23(8):752-4.
18. Rauzzino MJ, Tubbs RS, Alexander E III, Grabb PA, Oakes WJ. Spinal neuroenteric cysts and their relation to more common aspects of occult dysrafism. Neurosurg Focus [serial on the Internet] 2001 [cited 2004 Jan 19];10(1):[about 10 p.]. Available from: http://www.neurosurgery.org/focus.
19. Arsic D, Qi BD, Beasley SW. Hedgehog in the human: a possible explanation of the VATER association. J Paediatr Child Health. 2002;38(2):117-21.
20. Gillick J, Mooney E, Gilles S, Banningan J, Puri P. Notochord anomalies in the adryamicin rat model: a morphologic and molecular basis for the VACTERL association. J Pediatr Surg. 2003;38(3):469-73.
21. Ybot-Gonzalez P, Cogram P, Gerreli D, Copp AJ. Sonic Hedgehog and the molecular regulation of mouse neural tube closure. Development. 2002;129(10):2507-17.

Corresponding author:
Lisieux Eyer de Jesus
Rua Presidente Domiciano, 52/801
CEP 24210-270 – Niterói, RJ, Brazil