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CASE REPORT

A case of split notochord syndrome with congenital ileal atresia, the total absence of a colon, and a dorsal enteric cyst communicating to the retroperitoneal isolated ceca with a vesical fistula

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Abstract Split notochord syndrome (SNS) is an extremely rare anomaly. This report presents the case of a male infant with SNS associated with congenital ileal atresia and a dorsal enteric cyst communicating to the retroperitoneal isolated ceca with a vesical fistula. Dorsal fistulography and vesicography were useful and essential for the detailed study of the topology in this patient. The embryological mechanism and etiologic theories are discussed with a review of 19 cases reported in the literature.

Keywords Split notochord syndrome · Dorsal enteric fistula · Intestinal atresia

Introduction

Split notochord syndrome (SNS) is an extremely rare congenital malformation that results in spinal anomalies, associated with anomalies of the vertebrae, central nervous system, and gastrointestinal tract. The anomalies arise from a connection between endoderm and dorsal ectoderm. Nineteen cases of SNS with a dorsal enteric fistula had been previously reported [1–19]. Abnormal splitting of the notochord can result in a wide spectrum of malformations depending on its size, site, and point in time.

This report describes a unique variant case of SNS associated with congenital ileal atresia, the total absence of a colon, and a dorsal enteric cyst communicating with a retroperitoneal isolated ceca with a vesical fistula.

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Case report

A boy was born at 37 weeks 6 days of gestation by Caesarean section as the first child of a 33-year-old mother. He had no family history of congenital anomalies, or consanguinity. His mother was not medicated during pregnancy.

At birth, a meningocele and a cyst were noted on the patient's dorsal site (Fig. 1), along with anal atresia and urethral atresia of the penis. Hypoplasia of the left kidney was recognized by abdominal CT. The spine showed a complete cleft below the tenth thoracic vertebra in a chest abdominal X-ray and three-dimentional CT (Fig. 2).

The first surgery was performed at 0 days of age. A congenital ileal atresia was identified near the terminal ileum, without a distal ileum and colon (Fig. 3a). An end-ileostomy was constructed. A cystostomy was performed to rectify the urethral atresia. Furthermore, there was an isolated ceca at the midline of the retroperitoneum just above urinary bladder (Fig. 3b). There was no definitive evidence that the retroperitoneal isolated ceca was in contact with the dorsal cyst; therefore, only the dorsal cyst was resected. A postoperative histopathological examination showed the cyst to be an enteric cyst that was composed of a continuous mucosal epithelium which originated from the esophagus, stomach, and colon. After the first operation, a continuous urinous discharge was recognized from the dorsal wound that formed when the dorsal enteric cyst was removed. Dorsal fistulography and vesicography showed a dorsal enteric fistula and a vesical fistula via the retroperitoneal isolated ceca (Fig. 4a, b). The retroperitoneal isolated ceca was in contact with the dumbbell-shaped

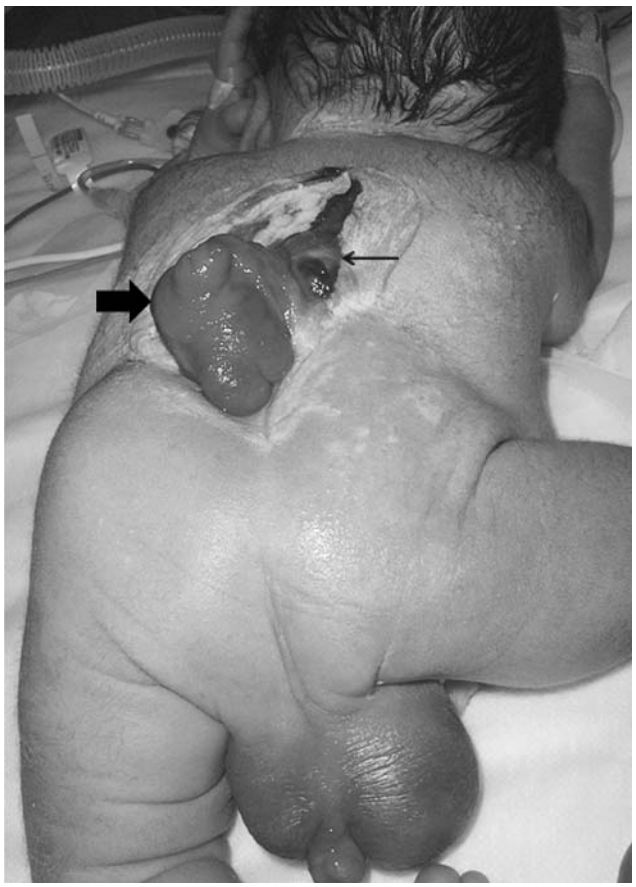

Fig. 1 Photograph of the back of patient. A meningocele (*arrow*) and enteric cyst (*bold arrow*) were noted on the patient's dorsal site, and anal atresia was recognized

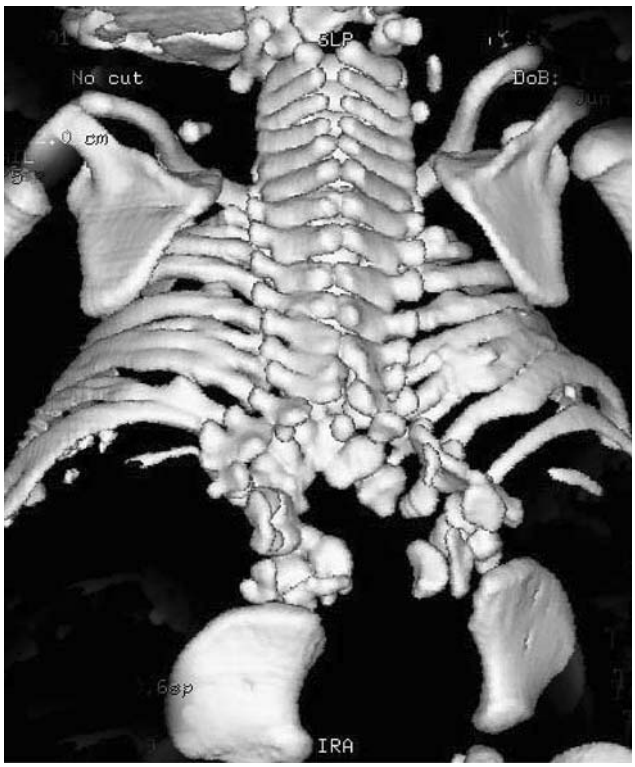

Fig. 2 The spine showed a complete cleft below the 10th thoracic vertebra in 3-dimentional CT

dorsal cyst (Fig. 5). The second surgery was performed at 7 days in order to repair a meningocele.

The third surgery was performed at 10 months. After the isolated ceca was incised, the vesical fistula and the dorsal

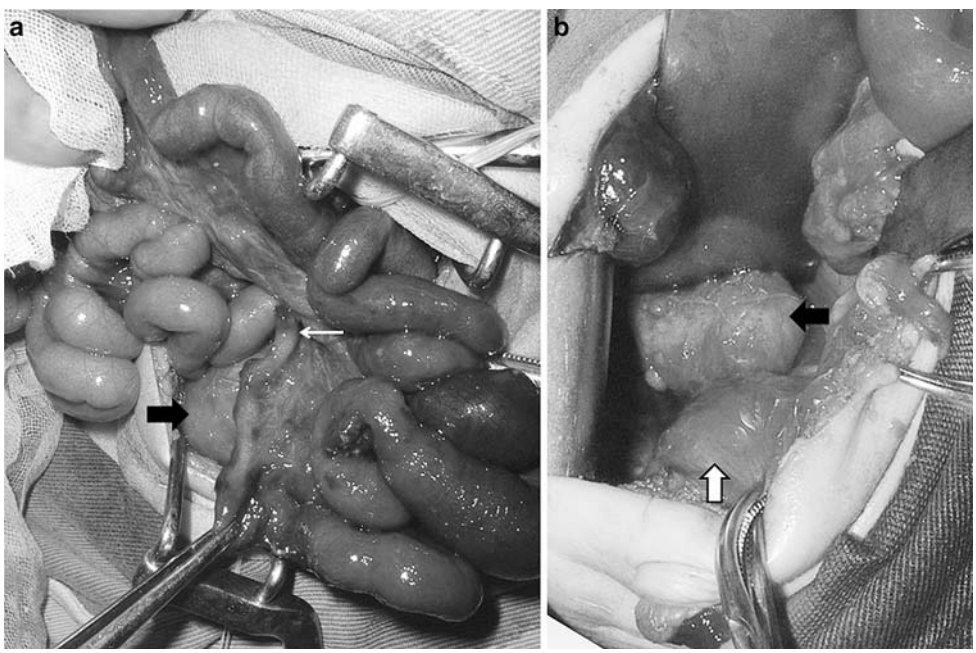

Fig. 3 Operative finding (first operation): **a**. Congenital ileal atresia was shown near the terminal ileum without a distal ileum and colon. The end point of the ileum (*white arrow*) and the isolated ceca (*black bold arrow*) were shown. **b**. There was an isolated ceca (*black bold arrow*) at the midline of the retroperitoneum. The *white bold arrow* shows the urinary bladder

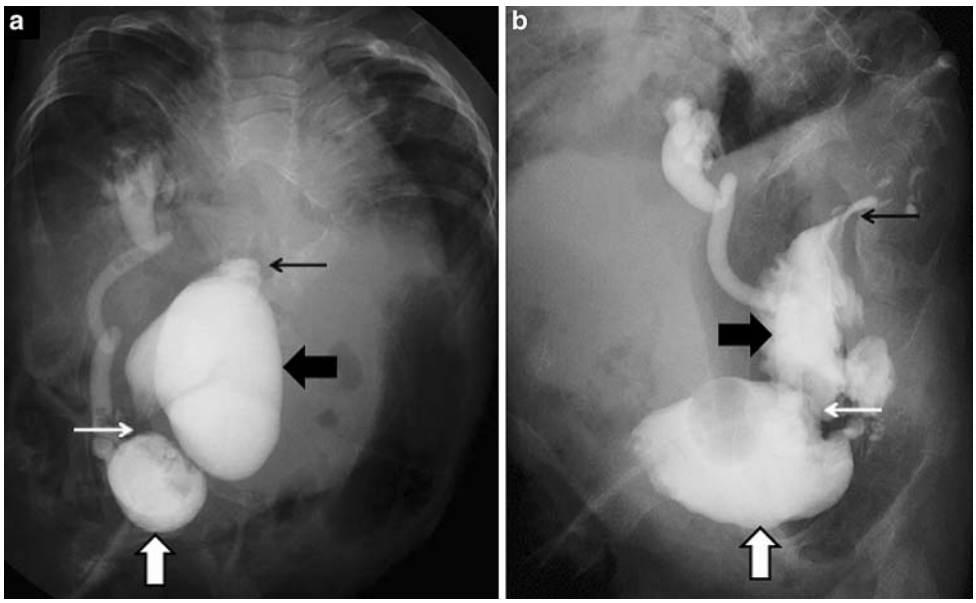

Fig. 4 **a** Vesicography and **b** Dorsal fistulography: The vesical fistula (*white arrow*), urinary bladder (*white bold arrow*), the dorsal enteric fistula (*black arrow*) and the isolated ceca (*black bold arrow*) were identified respectively

enteric fistula were identified. A vesical fistulectomy and a dorsal enteric fistulectomy were performed, and the isolated ceca was completely resected. After the third operation, the patient died from DIC syndrome.

Discussion

SNS was first described by Rembe in 1887 [20]. This syndrome is associated with vertebral anomalies, central nervous system anomalies, and intestinal anomalies. There are two theories to explain the etiology of SNS that have been described by Bremer and Sanders. Bremer [21] proposed that the dorsal intestinal fistula may result from the persistence or partial obliteration of a primitive neurentiric canal. In contrast, Sanders [2] proposed that a split or localized duplication of the notochord may cause this anomaly, and the primitive gut or endoderm herniates through the opening and adheres to the dorsal ectoderm. The latter theory is widely accepted.

Several reports have addressed SNS, but SNS with a dorsal enteric cyst (fistula) has been reported in only 19 cases.

A review of the literature of the 19 cases of SNS with a dorsal enteric fistula revealed that the spinal cleft in most of the patients reached to the end of the sacrum (Table 1). There was partial spinal defect in three cases, and a double split in one case [16]. Meningocele or meningomyelocele were present in 15 cases, and a teratoma occurred in 2 cases around the dorsal site. The intestinal fistula was located in the ileum in two cases, the cecum in four, the colon in four, the rectosigmoid in seven, a duplication of the gastrointestinal tract in three, and they were obscure in two cases. Intestinal anomalies, including an imperforated anus were reported in ten cases, a short colon in two, a malrotation in two, and the absence of a transverse colon in one. The current case may be the first report where the dorsal enteric cyst is in contact with the retroperitoneal

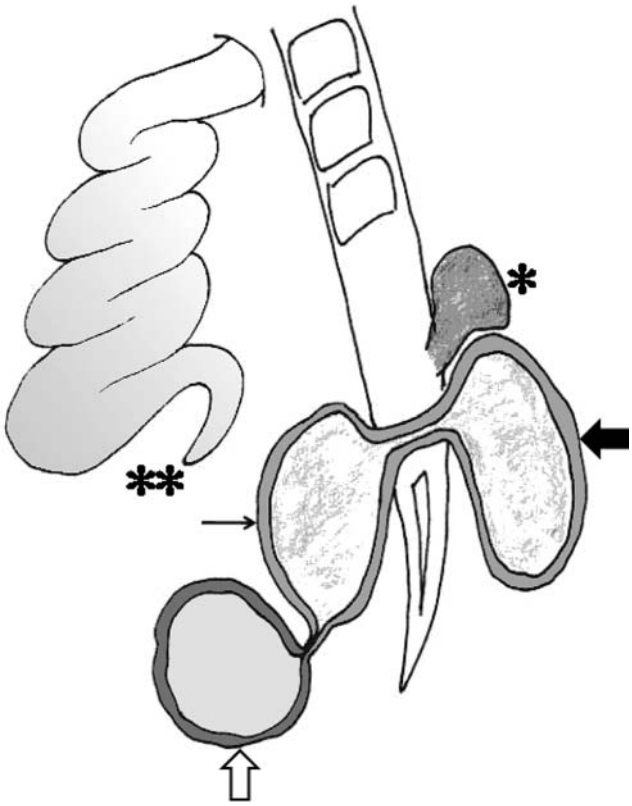

Fig. 5 Schema of this case: the detail topology of the isolated ceca (*black arrow*), the dorsal enteric cyst (*black bold arrow*), urinary bladder (*white bold arrow*), a meningocele (*asterisk*) and the end point of ileum (*double asterisk*) are indicated

Table 1 SNS cases with a dorsal enteric fistula

	Authors	Sex	Cleft	Around dorsal anomaly	Enteric opening site	Anus	Associated anomalies	Result
1	Keen and Coplin [1]	F	L3–sacrum	Teratoma	Rectum	Normal		Alive
2	Saunders [2]	F	L1–S2	None	Cecum and rectum	Normal	Absent T-colon, uro anomalies	Died
3	Rosselet [3]	M	T12–sacrum	Meningomyelocele	S-colon	Normal	Ambiguous genitalia	Died
4	Bentley and Smith [4]	F	L2–sacrum	None	Rectum	Atretic	None	Died
5	Faris and Crowe [5]	M	L9–sacrum	Meningomyelocele	Colon	Atretic	Bifid scrotum, short colon	Died
6	Singh and Singh [6]	M	T10–sacrum	Meningomyelocele	Colon	Normal	Absent ear lobule, depressed nose	Died
7	Gupta and Deodhar [7]	M	L5–sacrum	Meningomyelocele	Rectum	Normal		Alive
8	Burrows and Sutcliffe [8]	M	L2–sacrum	Meningomyelocele	Colonic duplication	Normal	Paralized left leg	Alive
9	Kherapir and Ameri [9]	M	T11-sacrum	Meningomyelocele		Atretic		Died
10	Kramer et al. [10]	M	T10–sacrum	Meningomyelocele	Cecum	Atretic	None	Died
11	Meller et al. [11]	M	L1–sacrum	Meningomyelocele	Cecum duplication	Ectopic	Malrotation	Alive
12	Hoffman et al. [12]	F	T12–sacrum	Meningocele		Atretic	Cloacal extrophy	Died
13	Razack and Page [13]	F	L2–sacrum	Meningocele, teratoma	Rectosigmoid	Normal	Encephalacele	Alive
14	Akgur et al. [14]	M	T10–L5	Meningomyelocele	Ileocecal duplication	Normal		Died
15	Kiristioglu et al. [15]	M	L1–sacrum	Meningocele	Rectum	Atretic	None	Alive
16	Pathak et al. [16]	F	C1–C4, T12–sacrum	None	Stomach and small intestine, intestine	Atretic	Short colon	Died
17	Kanmaz et al. [17]	M	T11–L4	Meningocele	D-colon	Normal	Malrotation	Died
18	Jesus and Franca [18]	M	T11–sacrum	Meningocele	Small intestine	Atretic		Died
19	Agangi et al. [19]	F	L4–sacrum	Meningocele	Rectum	Atretic	Rectourethral fistula	Alive
20	Our case	M	T10–sacrum	Meningocele	Isolated intestine duplication?	Atretic	Vesical fistula, intestinal atresia, absent total colon	Died

isolated ceca. Intestinal anomalies, congenital intestinal atresia and the total absence of a colon were only recognized in the present case. The survival rate of this anomaly is poor, only seven non-fatal cases were reported.

The pathological examination of the dorsal cyst revealed it to be an enteric cyst because of the continuous mucosal epithelium which originated from the esophagus, stomach, and colon. It was similar to the etiology of a duplication cyst. A duplication cyst is a disease resulting from disorders of notochord formation, so there is a strong possibility of this occurring as a complication of SNS. This patient had an imperforated anus as an additional complication. The association of an imperforated anus with a vesical fistula in the isolated ceca is not surprising. However, the cause of the total absence of a colon is unknown.

The present case was not diagnosed prenatally; however, prenatal diagnosis of SNS with a dorsal enteric fistula is very difficult. Only one previous case was successfully prenatally diagnosed with echosonography [19]. The management of SNS must be tailored on the different anomalies present in each case basically. However, some authors have proposed a two-stage approach with the correction of the intestinal lesion and the subsequent correction of the spinal neurological lesion [15, 18, 19]. In the present case, three separate operations were performed, because of the unique associated anomalies of the congenital ileal atresia, the total absence of a colon, and the dorsal cyst in contact with the retroperitoneal isolated ceca with vesical fistula. Cystography, including dorsal fistulography and vesicography are therefore considered to be useful and essential for the detailed study of the topology.

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