

Are your MRI contrast agents cost-effective?
Learn more about generic Gadolinium-Based Contrast Agents.



AJNR

The split notochord syndrome with dorsal enteric fistula.

C H Hoffman, R B Dietrich, M J Pais, D S Demos and H F Pribram

AJNR Am J Neuroradiol 1993, 14 (3) 622-627
<http://www.ajnr.org/content/14/3/622>

This information is current as of April 19, 2024.

The Split Notochord Syndrome with Dorsal Enteric Fistula

C. H. Hoffman,^{1,3} R. B. Dietrich,¹ M. J. Pais,¹ D. S. Demos,² and H. F. W. Pribram¹

Summary: Split notochord syndrome with dorsal enteric fistula is an extremely rare congenital anomaly that may be associated with meningomyelocele or meningocele, and genitourinary anomalies. This case presented with an additional finding of bladder exstrophy, raising the possibility of a relationship between this syndrome and the OEIS complex.

Index terms: Spinal cord, congenital anomaly; Pediatric neuro-radiology; Fetus, growth and development; Spinal cord, magnetic resonance; Spinal cord, computed tomography

The split notochord syndrome with dorsal enteric fistula is a rare congenital anomaly, consisting primarily of division of the spinal cord and vertebral column at the lumbar level and, occasionally, the lower thoracic level. Associated findings have included meningocele or meningomyelocele and genitourinary abnormalities (1–10). We present such a case, discuss the embryology of this confusing syndrome, and raise the possibility of a relationship between the split notochord syndrome and the OEIS complex (omphalocele, exstrophy, imperforate anus, and spinal defects).

Case Report

A 3.1-kg genetically female newborn was delivered by cesarean section at 39 weeks of gestation. Pregnancy and family history were unremarkable; specifically, there was no history of maternal drug abuse or consanguinity. Initial clinical examination revealed a meningocele at the T11 level, imperforate anus, bladder exstrophy, ambiguous genitalia, bilateral equinovarus deformity, and herniation of the bowel posteriorly with fistula formation (Fig. 1). There was active movement of the lower extremities.

Plain radiographic films obtained after administration of intravenous contrast soon after birth demonstrated duplication of the vertebral bodies caudally from approximately the T11-T12 level, a bony protuberance projecting from the vertebral bifurcation, and a hemivertebra at the T4 level (Fig. 2). The anomalous bone, with an adjacent epiphysis, resembled a long bone such as a humerus or femur

(Fig. 2). There was sacral dysgenesis. The iliac bone and superior and inferior pubic rami were well defined bilaterally.

Magnetic resonance images of the body using both T1- and T2-weighted spin-echo pulse sequences demonstrated extensive abnormal anatomy including a hemivertebra at the T4 level and bifurcation of vertebral bodies at the T11-T12 level. The spinal cord was normal until the T11-T12 level, where it bifurcated into two separate divisions of the spinal cord. Nerve roots were identified lateral to the two spinal cord divisions but were not seen medially (Figs. 3A–3C). A meningocele was seen posterior to the T11 vertebral body (Fig. 3D). The liver was midline with a slight right-sided predominance. The spleen was small and posterior to the stomach. The kidneys were malrotated, malascended, and there was left hydronephrosis. The dorsal enteric fistula (Fig. 3E) and bladder exstrophy were identified.

Computed tomographic images showed well-defined bony posterior elements both above and below the bifurcation of the vertebral bodies. Inferior to the bifurcation, the bony posterior elements were more completely developed on the right (Fig. 4). Superior to the bifurcation, the spinal canal was singular and very large.

Attempt at closure of the enteric fistula failed and the patient died because of electrolyte imbalance and infection. An autopsy was refused.

Discussion

Bentley and Smith (1) were the first to note that an abnormal splitting of the notochord could cause a wide variety of malformations not only of the vertebral bodies and neural tissue but also of the enteric viscera. Others have since evaluated various stages of development of the notochord in an attempt to duplicate this abnormality (11, 12). Despite this, the split notochord syndrome remains complex and poorly understood. Of the 14 human cases previously reported (1–10), seven have been associated with meningocele or myelomeningocele (1–6, 8), four had an

Received November 12, 1991; revision requested February 7, 1992; revision received July 3 and accepted July 7.

¹ Department of Radiological Sciences and ² Departments of Pediatrics and Neurology, University of California, Irvine, CA.

³ Address reprint requests to Cheryl H. Hoffman, MD, University of California, Irvine Medical Center, 101 The City Drive, Orange, CA 92668.

AJNR 14:622–627, May/June 1993 0195-6108/93/1403-0622 © American Society of Neuroradiology

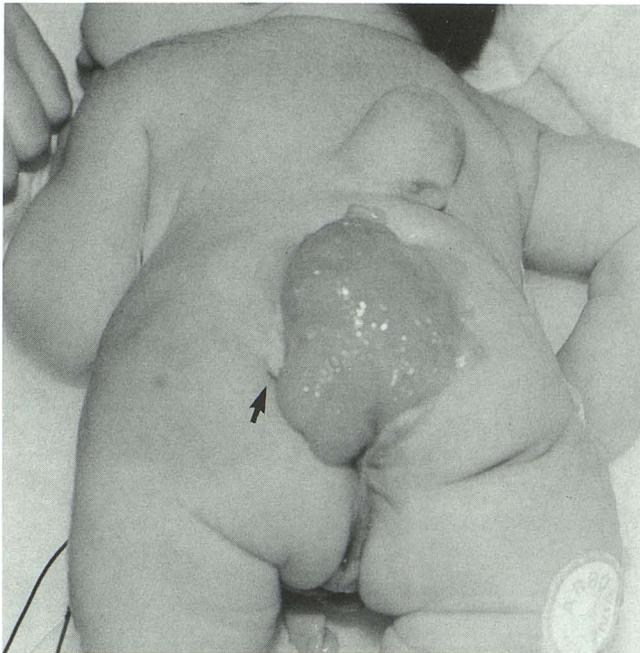
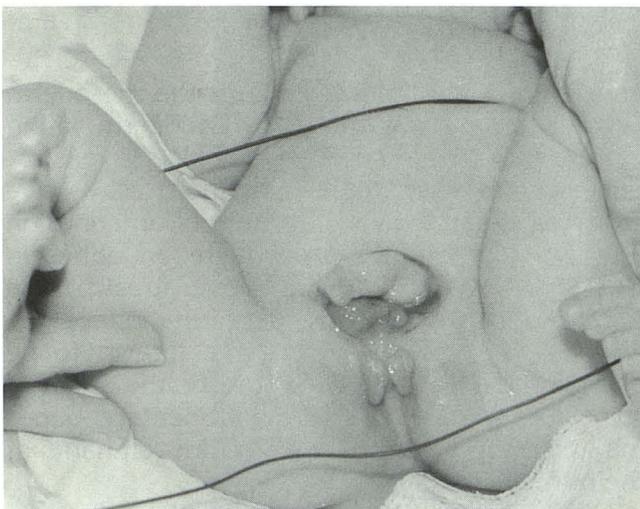
**A****B**

Fig. 1. Photographs.

A, A large dorsal enteric cyst is seen, from which meconium is being extruded (*arrow*). Two meningoceles are identified at the midthoracic level. There is no anal orifice.

B, An anterior wall defect is present that contains the exstrophied bladder and the exiting umbilical cord. No vaginal orifice is seen. Bilateral equinovarus deformities are also present.

imperforate anus (3, 5, 7, 8), and one had a rudimentary extremity, most resembling an arm, extending from the spinal cleft (3). One of these cases was associated with bladder exstrophy (10). To explain the findings seen in this case, it is necessary to review briefly the events that occur

during the second and third weeks of normal embryogenesis.

Normal Embryogenesis

By the end of the second week, the embryo is in the form of a bilaminar disk consisting of layers of endoderm and ectoderm. The ectoderm is positioned adjacent to the amnion and the endoderm abuts the yolk sac (13). Following development of the prochordal plate in the cephalic region of the disk (13), the primitive streak begins to develop on the caudal portion of the amniotic surface of the ectoderm, immediately adjacent to the area later to become the cloaca. The cephalic portion of the primitive streak then thickens to form the primitive knot. The notochordal process forms from cells that migrate cephalad from the center of the knot, invaginating between the endoderm and ectoderm, until they reach the prochordal plate. At approximately the same time, the interembryonic mesoderm is formed from cells that originated from the primitive streak and have migrated laterally again invaginating between the endoderm and ectoderm (13, 14).

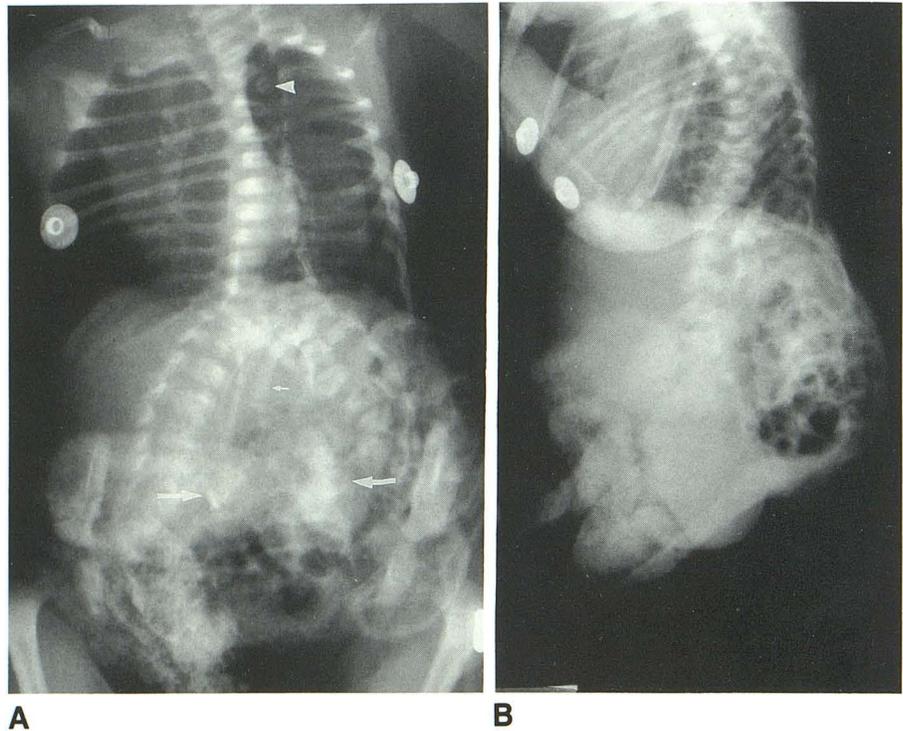
Therefore, within the third week of development, the embryo has become a trilaminar disc and the notochordal process and the mesoderm separate the endoderm from the ectoderm, except in the region cephalad to the prochordal plate and the region caudal to the primitive streak, the cloacal plate. At this time the primitive streak deepens and invaginates into the notochordal process forming the notochordal canal (the canal of Kovalevsky) (13). Multiple areas of dehiscence then occur, forming accessory neur-enteric canals, connecting the notochordal canal to the amniotic canal. The notochordal canal and these areas of dehiscence connect the amniotic cavity with the yolk sac, and are normally obliterated within a few days of their formation (3, 8). Separation of the endoderm (the eventual gut roof) from the ectoderm (the future neural tube) then begins in a rostral-to-caudal direction and the solid central core of tissue that is formed is the true notochord (14, 15).

Lateral mesodermal masses, somites, are then laid down adjacent to the notochord (14). During the fourth week of development, the medial half of the somite, the sclerome cells, migrate around the notochord and form the vertebral column. The lateral cells of the somite, myotome cells,

Fig. 2. Anteroposterior and lateral radiographs of the chest and abdomen following injection of intravenous contrast.

A, The kidneys (*large arrows*) are malascended and malrotated, and left hydronephrosis is present. A hemivertebra is present at T4 (*arrowhead*). Sacral agenesis and bifurcation of the vertebral bodies and posterior elements below the T11-T12 level are seen. There is anomalous bone formation present at the level of the bifurcation (*small arrow*).

B, The lateral view demonstrates the posteriorly herniating bowel.



develop into the supporting musculature (16). As the notochord is formed, and most likely under its inductive influence (13, 14), the ectoderm overlying the notochord gives rise to the central nervous system. Subsequently, the notochord regresses and only persists as a portion of the nucleus pulposus and, rarely, as a remnant within the vertebral body (17).

Theories of Embryogenesis of the Split Notochord Syndrome

Several interesting theories of embryogenesis have been proposed for the split notochord syndrome.

An endodermal-ectodermal adhesion occurring within the primitive streak because of incomplete separation of the two layers may be the primary abnormal event that is responsible for producing this anomaly (18–20). As the notochord developed, it would have to migrate and bifurcate around the adhesion. Not only would this theory explain the spinal anomalies seen, it would also allow for the possibility of either a duplication or a division of the spinal cord. This theory may also explain the associated bladder exstrophy and the rudimentary bone formation. The presence of an endodermal-ectodermal adhesion within the primitive streak would not only have prevented invagination by the notochord but would also

have interrupted the migration of the mesoderm between the ectoderm and the endoderm within the cloacal plate. Therefore, the bladder exstrophy and the split of the notochord may have occurred simultaneously because of an adhesion (Fig. 5). Moreover, if there were an adhesion caused by injury to the primitive streak, there may also be anomalous bone formation, as the cells of the primitive streak are known to be pluripotential and are thought to be the origin of spinal teratomas (21). The rudimentary bones may therefore have developed from remnants of the primitive streak.

Other studies suggest that an initial early teratogenic or spontaneous mutation of the developing notochord occurs leading to a split of its inferior portion (3). Under the stimulus of the notochordal tissue, normal vertebral bodies could then develop around both divisions of the spinal cord. The enteric yolk sac would herniate between both divisions of the spinal cord and adhere to the dorsal ectoderm or rupture into the amniotic cavity (22). This theory, on its own, would not explain the findings of imperforate anus, exstrophy of the bladder, and the rudimentary bone anomaly. However, if a caudal spontaneous or teratogenic mutation that split the notochord and also ruptured the cloacal membrane occurred, the findings seen in this case might be produced (23, 24).

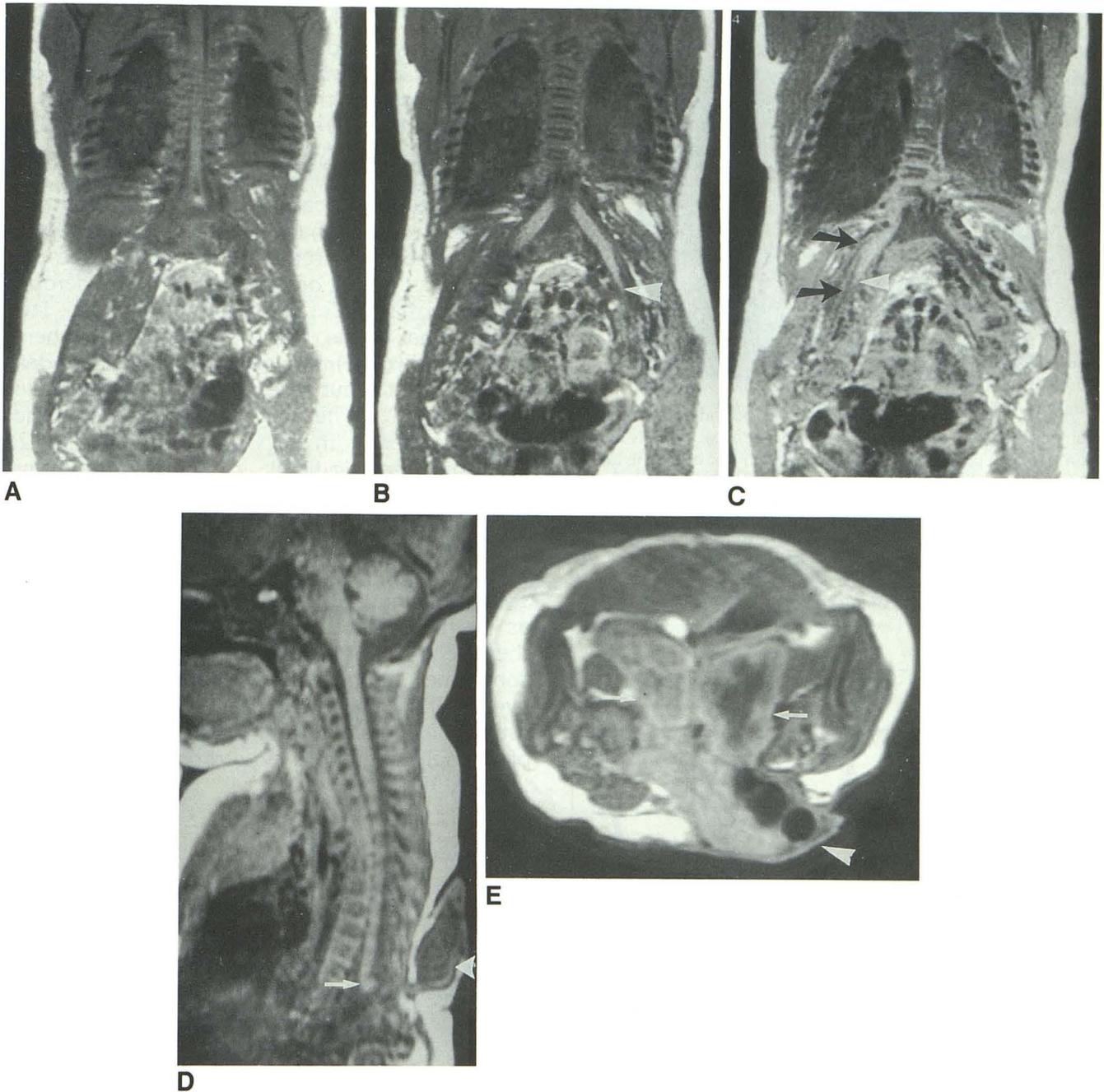


Fig. 3. Magnetic resonance imaging.

A–C, Coronal spin-echo image (600/20) (TR/TE). Section thickness was 3 mm with a 1-mm gap. There is bifurcation of the spinal cord at T11–T12 with continuation to a conus bilaterally (*arrowheads*). Nerve roots are identified lateral to the spinal cords bilaterally (*arrows*). A hemivertebra is seen at T4.

D, Sagittal spin-echo (300/20). A meningocele (*arrowhead*) and abnormal “kinking” of the cord (*arrow*) are seen.

E, Axial spin-echo (800/20). At the level of the iliac crest, the malascended and malrotated kidneys (*arrows*) with left-sided hydronephrosis are demonstrated. The enteric fistula is seen exiting posteriorly (*arrowhead*).

Alternatively, the syndrome may occur because of persistent or only partial obliteration of an accessory neurenteric canal or the notochordal canal. This theory might also explain both the presence of a spinal anomaly and a dorsal enteric

fistula (25, 26) and would imply that the cord was split rather than duplicated. Furthermore, a teratogenic event, occurring as the notochordal canal or an accessory neurenteric canal is closing, may produce not only persistence of a canal but

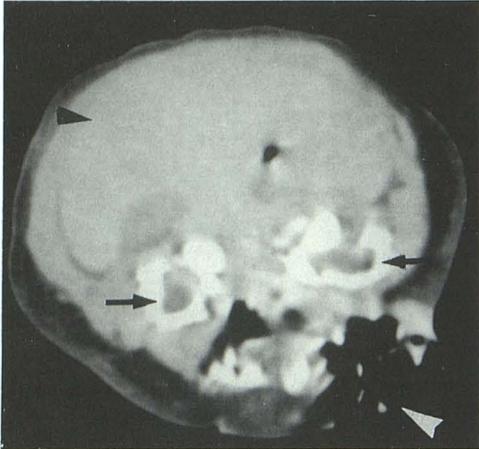


Fig. 4. Computed tomography. The posterior and anterior bony elements are delineated bilaterally (arrows). There is herniation of bowel posteriorly (white arrowhead) and a midline liver is identified (black arrowhead).

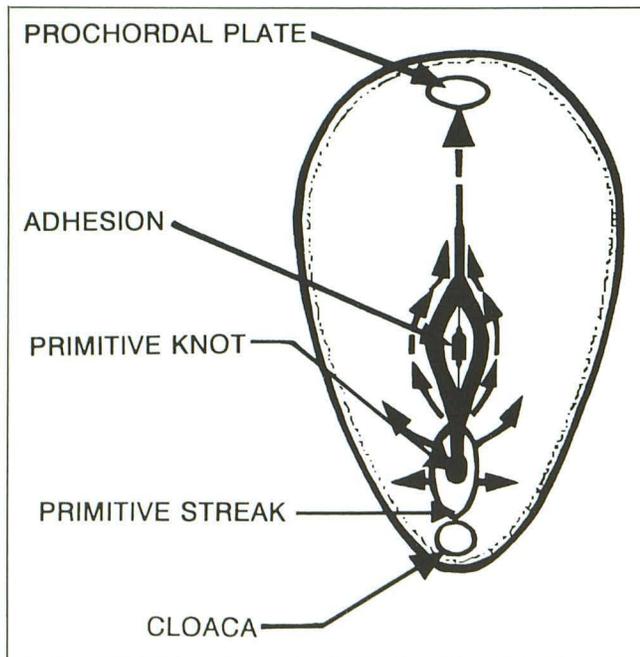


Fig. 5. Possible embryogenesis of the split notochord syndrome. An adhesion in the primitive streak or in the pathway of migration of the notochordal process would produce a cleft in the notochord.

also a cloacal abnormality such as bladder exstrophy.

More recent theories for the split notochord are based upon the presumed occurrence of vascular compromise of the inferior neural structure, preventing neural tube closure (27). Alternatively, incomplete or attempted twinning may be responsible for the findings seen in our case (28, 29).

Of the 14 reported cases of the split notochord syndrome with dorsal enteric fistula, only four were associated with imperforate anus and one was associated with bladder exstrophy. Both of these findings were present in this case. This raises the possibility of an association of the split notochord syndrome with the OEIS complex, also referred to as cloacal exstrophy. This case and the OEIS complex have several features in common; namely, exstrophy of the bladder, imperforate anus, spinal defects, equinovarus deformity, abnormalities of the external genitalia, lack of a vaginal orifice, and the presence of a hemivertebra (30, 31). An omphalocele is not definitely present in our case; however, omphaloceles are reported to be only "commonly" but not always present with the OEIS complex (19, 32). The dorsal enteric fistula in this case may have decreased the intraabdominal pressure that is required to produce an anterior omphalocele.

In summary, one can only speculate as to the relationship of the embryogenesis of both the split notochord and the OEIS complex. However, as we have discussed above, it is possible that the spectrum of anomalies seen in both may sometimes be caused by the same or similar events. It is likely that cases presenting in the future will give still further insight into the etiology of the split notochord syndrome with dorsal enteric fistula.

Acknowledgments

Malcolm Jones, MD, Kenneth Lyons Jones, MD, and Kurt Benirschke, MD, provided invaluable insight and guidance. We would also like to thank Judge Marilyn Hoffman for editing the manuscript, Betty Trent for typing the manuscript, and Dana Glidden for helping with the illustrations.

References

1. Bentley JFR, Smith JR. Developmental posterior enteric remnants and spinal malformation: the split notochord syndrome. *Arch Dis Child* 1960;35:76-86
2. Burrows FGO, Sutcliffe J. The split notochord syndrome. *Br J Radiol* 1968;41:844-847
3. Faris JC, Crowe JE. The split notochord syndrome. *J Pediatr Surg* 1975;10:467-472
4. Singh A, Singh R. Split notochord syndrome with dorsal enteric fistula. *J Pediatr Surg* 1982;17:412-413
5. 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;38:186-187
6. Gupta DK, Deodhar MC. Split notochord syndrome presenting with meningomyelocele and dorsal enteric fistula. *J Pediatr Surg* 1987;22:382-383

7. Pathak VB, Singh S. Double split of notochord with massive prolapse of the gut. *J Pediatr Surg* 1988;23:1030-1040
8. Kramer EL, Giacoia GP. Split notochord syndrome with dorsal enteric fistula and sacral agenesis. *Teratology* 1988;38:1-5
9. Meller JL, Loeff DS. A variant of the split notochord syndrome. *J Pediatr Surg* 1989;24:733-735
10. Taybi H, Lachman RS. *Radiology of syndromes, metabolic disorders, and skeletal dysplasias*. 3rd ed. Chicago: Year Book, 1990
11. Muller F, O'Rahilly R. The early origin of vertebral anomalies, as illustrated by a "butterfly vertebra." *J Anat* 1986;149:157-169
12. Yasuda Y, Konishi H. Discontinuity of primary and secondary neural tube in spina bifida induced by retinoic acid in mice. *Teratology* 1990;41:257-274
13. Naidich TP, Zimmerman RA, McLone DG. Congenital anomalies of the spine and spinal cord. In: Atlas SW, ed. *Magnetic resonance imaging of the brain and spine*. New York: Raven Press, 1991
14. Langman J. *Medical Embryology*. 4th ed. Baltimore: Williams & Wilkins, 1981
15. Kalousek DK, Fitch N, Paradise BA. *Pathology of the human embryo and previsible fetus*. New York: Springer Verlag, 1990.
16. Muecke EC. The role of the cloacal membrane in exstrophy: the first successful experimental study. *J Urol* 1964;92:659-667
17. Peacock A. Observations on the prenatal development of the intervertebral disc in man. *J Anat* 1951;85:260-274
18. Beardmore HE, Wiglesworth FW. Vertebral anomalies and alimentary duplications. *Pediatr Clin North Am* 1958;5:457-471
19. Prop N, Frensdorf EL. A postvertebral endodermal cyst associated with axial deformities: a case showing the "endodermal-ectodermal adhesion syndrome." *Pediatrics* 1967;39:555-562
20. Naidich TP, McLone DG, Harwood-Nash DC. Spinal dysraphism. In: *Computed tomography of the spine and spinal cord*. Newborn TH, Potts DG, eds. San Anselmo, CA: Clavadel Press, 1983
21. Moore KL. *The developing human*. Philadelphia: W. B. Saunders, 1982
22. Sanders RL de CH. Combined anterior and posterior spina bifida in a living neonatal human female. *Anat Rec* 1943;87:255-278
23. Zarabi C, Rupani M. Cloacal exstrophy: a hypothesis on the allantoic origin of the distal midgut. *Pediatr Pathol* 1985;4:117-124
24. Thomalla J, Rudolph R, Rink R, Mitchell M. Induction of cloacal exstrophy in the chick embryo using the CO₂ laser. *J Urol* 1985;134:991-995
25. Bremer JL. Dorsal intestinal fistula, accessory neurenteric canal, diastematomyelia. *AMA Arch Pathol* 1952;54:132-138
26. Gaskill S, Kagen-Hallett K, Marlin A. Diastematomyelia associated with ectopic renal tissue. *Pediatr Neurosci* 1988;14:108-111
27. Stevenson RE, Kelley JC, Aylsworth AS, Phelan MC. Vascular basis for neural tube defects: a hypothesis. *Pediatrics* 1987;80:102-106
28. Arey LB. *Developmental anatomy*. Philadelphia: W. B. Saunders, 1974
29. Benirschke K, Kaufman P. Multiple pregnancy. In: *Pathology of the human placenta*. New York: Springer-Verlag, 1990
30. Carey J, Greenbaum B, Hall B. The OEIS complex (omphalocele, exstrophy, imperforate anus, spinal defects). *Birth Defects Orig Art Ser* 1978;14:253-263
31. Kelalis P, King L, Belman A. *Clinical pediatric urology*. 2nd ed. Philadelphia: W. B. Saunders, 1985
32. Hurwitz R, Antonio G, Manzoni M, Ransley P, Stephens F. Cloacal exstrophy: a report of 34 cases. *J Urol* 1987;138:1060-1068