Case Report

A Case of Split Notochord Syndrome with Left Congenital Diaphragmatic Hernia: A Rare Association

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ABSTRACT

Split notochord syndrome is a rare neural tube malformation involving the brain, spinal cord, and vertebral column. In nearly half of the cases, the malformation also involves the gastrointestinal tract in the form of a dorsal neurenteric fistula and imperforate anus. In the literature, less than 50 cases have been reported. To the best of my knowledge, this is the first reported case associated with a left congenital diaphragmatic hernia.

KEYWORDS: Congenital anomaly, neurenteric fistula, split notochord syndrome

INTRODUCTION

Split notochord syndrome (SNS) represents a rare and pleomorphic form of spinal dysraphism. It is associated with a wide spinal defect and persistent communication between endoderm and ectoderm, manifesting as an intestinal fistula opening in the dorsal area. Several variations are observed depending on the type of spinal deformity and the site of the associated anomalies. It occurs mainly in the thoracicolumbar region, but when it affects the lumbosacral area, it is usually attended by imperforate anus and meningo(myelo)cele.

CASE REPORT

A 5-day-old baby boy presented to the pediatric surgery department with a lump at the thoracic region, difficulty in breathing, and an enlarged head. He was born to a primigravida from a non-consanguineous marriage by normal vaginal delivery. The mother received no prenatal care and there was no history of fever or exposure to any teratogenic agents. The family history was unremarkable. The weight of the baby was 2.4 kg, and he was dull, hypothermic, tachypneic with a respiratory rate of 62/min, and had tachycardia of 130/min. The baby was resuscitated. On examination, he had enlarged head with bulging fontanel, dorsal midline spinal defect with prolapsing bowel loops at 5th–7th thoracic vertebrae [Figure 1]. There was no leakage of cerebrospinal fluid or visible exposure of neural tissue elements. The infant had absent breath sounds in the left lung. He was awake and responsive to stimuli. Neurological examination showed flaccid paraplegia. The anal opening was present, and the baby had normal genitalia. Chest radiograph showed vertebral anomalies in the thoracic region, extending from the 5th to 7th thoracic vertebral and left congenital diaphragmatic hernia with the shift of mediastinum toward the right side [Figure 2]. Cranial ultrasound of the patient revealed hydrocephalus with ventricle to head ratio of 60%. Echo of heart showed a single ventricle, tricuspid atresia, poorly developed right heart structures, and double outlet left ventricle. The patient could not be revived and succumbed to death.

DISCUSSION

The SNS was first described by Rembe in 1887.[1] This syndrome involves vertebral anomalies, central nervous system abnormalities, and gastrointestinal defects. The embryological origin of this anomaly is uncertain. Several hypotheses have been put forward. One is the persistence or partial obliteration of an accessory neurenteric canal that connects the yolk sac and the amniotic cavity in the 3rd week of gestation as suggested by Feller and Sternberg.[2] This theory

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was disproved because the notochord occurs in the infracoccygeal region, whereas in the SNS, the vertebral deformity is frequently proximal (cervical, thoracic, or lumbosacral). Bremer attributes this discrepancy to the different positions of Hensen’s node or to an accessory neurenteric canal. Alternately, it can occur from primary midline notochordal integration defect and paraxial mesodermal changes resulting in the split notochord, which is inadequately separated from the primitive intestine. It leads to herniation of the endoderm and underlying primitive bowel, which adheres to the dorsal ectoderm and eventually ruptures. The most widely accepted theory suggests the failure of fusion of the lateral ossification centers of the vertebral bodies (the cartilaginous hemicentrum) and interposition of disklike material with the diverse extent of attachment of the spinal cord and gastrointestinal system. The defect appears before the embryo is 63 mm. The terminal, dorsal part of the enteric fistula, forms between the endoderm and the ectoderm through a partially duplicated notochord in the development of the embryo. It remains after the obliteration of the fistula, and subsequently, the mucosa of the enteric remnant is inverted and projects through the skin of the back. Other etiological theories put forward are division or local redundancy of the notochord, endodermal–ectodermal adhesion, neural tube rupture caused by oversecretion of fluid, and failure or aberrancy of dorsal aortic distribution to the region of the neural folds, resulting in the prevention of timely neural tube closure.

Recent research revealed a complex, unified organization of molecular and genetic interactions, which receives and interprets the positional information supplied by graded Sonic hedgehog gene (Shh signaling) on chromosome 7q36. Thus, defective signaling produces a specific pattern acting as a morphogen for dorsoventral patterning of somites, anteroposterior polarity of the limb, and left–right asymmetry. Transforming growth factor, bone morphogenetic protein, and Wnt signaling also appear to influence patterning in the ventral neural tube.

SNS shows a relatively homogenous incidence in all ethnic groups, with no geographic or socioeconomic variations. No clear etiology has been acknowledged, but nearly half are related to maternal folic acid nutritional deficiency. There is a threefold higher incidence in consanguineous marriages and monozygotic twins, reflecting the importance of genetic factors. The babies
do not inevitably present with functional spinal cord defects. The location of the intestinal fistula varies and may be found either in the distal ileum or in the large intestine.

As the spectrum of SNS is broad, treatment options should be individualized according to the anomalies present that include correction of the intestinal lesion and correction of spinal dysraphism and cranial defects. The literature has described a poor prognosis for survival with only a few survivors being reported.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

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