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Spondylocostal Dysostosis Associated with Split Spinal Cord and Other Malformations

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Keywords
Radiology · Rib anomalies · Split spinal cord · Spondylocostal dysostosis · Vertebra malformations

Abstract
Spondylocostal dysostosis is a very rare combination of complex vertebra and rib malformations, accompanied occasionally by other disorders. A 3-year-old girl presented kyphoscoliosis, foot deformities, gate disturbance, and urinary incontinence. The CT and MRI examination revealed kyphosis and scoliosis with a double curve, some absent, broadened, bifurcating and fused ribs, hemivertebrae, butterfly and cleft vertebrae in thoracic and lumbar region, sporadic cleft or absent vertebral arches or pedicles, and hypoplastic sacrum with a cleft of the S2 vertebra. Spina bifida occulta extended from T10 to T11, and from L3 to the end of the sacrum. Two hemicords, separated by a bony septum and surrounded by their own dural tubes (type I), were present from the level of T9 to the conus medullaris. Filum terminale was thick and duplicated. Syringomyelia was present in the thoracic cord from T5 to T8. Finally, a small meningocele was seen at the T10–T11 level, and a subcutaneous lipoma in the thoracolumbar region. To our knowledge, such a combination of vertebra, rib, and cord malformations, including the mentioned additional disorders, has never been reported.

Introduction

The term spondylocostal dysostosis (SCD) is a combination of certain Greek and Latin words (Gr. *spondylos* = vertebra + Lat. *costa* = rib; Gr. *dys* = bad + Lat. *ostosis* = osteogenesis, i.e. a defect of a normal ossification of fetal cartilages). SCD, which occurs very rarely, is referred to as a combination of rib deformities and anomalies, and multiple vertebral malformations, e.g. hemivertebrae, hypoplastic, butterfly, cleft, and fused vertebrae, often with a characteristic "pebble beach" sign, which may affect any part of the spine [1, 2]. It is similar to the Jarcho-Levin syndrome in which, however, the latter sign is absent, as

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Table 1. Anomalies associated with spondylocostal dysostosis

Body systems	Associated anomalies	References
Nervous	Split spinal cord, syringomyelia, meningocele, myelomeningocele, lipomyelomeningocele, terminal ventricle, tethered spinal cord, dermal sinus, low-lying conus, Chiari malformation, congenital hemangioma, lipoma	[2, 9–19]
Cardiovascular	Double outlet right ventricle, septal defects, dextrocardia, tetralogy of Fallot, transposition of great arteries, congenital hemangioma	[3, 17, 20, 21]
Digestive	Anal atresia, bowl incontinence, abdominal wall or diaphragmatic hernias	[3, 19, 22, 23]
Respiratory	Restrictive lung disorders, pulmonary hypertension	[24]
Urogenital	Kidney or bladder agenesis, hydronephrosis, urinary incontinence, genitals agenesis	[17, 19, 25]
Skin	Polythelia, hemangioma, local hyperpigmentation, and/or hypertrichosis	[2, 15, 21]
Skeletal	Craniofacial asymmetry, micrognathia, skullbase-spine fusion, spina bifida, atlas aplasia or arch rachischisis, odontoid process agenesis, Klippel-Feil deformity, upper and lower limb anomalies, scoliosis or kyphoscoliosis, agenesis or hypoplasia of distal sacrum, osteoporosis	[10, 19, 26, 27]

are intrinsic rib anomalies, as well as to spondylothoracic dysostosis, also without intrinsic rib anomalies and with sparing of the sacrococcygeal region [1].

SCD is occasionally, albeit very rarely, associated with some other malformations and dysraphic disorders [2]; for example, with cranial and other bone anomalies, craniofacial anomalies, the Chiari malformation, spina bifida, split spinal cord, low conus medullaris, and filum terminale disorders with a consecutive tethering spinal cord, meningocele, dermal sinus, congenital hemangioma, lipoma, and polythelia, as well as with malformations or agenesis of certain thoracic and abdominal organs [3] (Table 1). One or more of these anomalies can be associated with SCD in the same patient.

Some patients with SCD are asymptomatic, whilst most others complain of back pain, weakness in the lower limbs, or urinary incontinence [1, 4]. These patients are candidates for surgical interventions, e.g. corrective surgery for scoliosis or kyphoscoliosis, chest wall reconstruction, and eventually surgery related to certain accompanying malformations [4].

As regards the mentioned split spinal cord (SSC) or diastematomyelia, it was associated with SCD in less than 10 patients [2]. Two main types of SSC were distinguished. Type I is characterized by two hemicords separated by a bony median septum (spur), each of them in its own dural tube. In type II, the two hemicords, separated by a fibrous septum, share a common dural tube. SCD is most often accompanied by type I [2], as was the case in our patient.

Case Study

A 3-year-old girl of married, healthy, and nonconsanguineous parents was a full-term baby delivered by caesarian section. There was no history of bony or central nervous system malformations in any member of the family, including the first child of the couple. She was admitted to the Clinic of Neurosurgery because of a previously diagnosed split spinal cord type I associated with kyphoscoliosis and a small lipomatous swelling in the thoracolumbar region. Her foot deformities were surgically corrected a few months before admission. A neurological examination revealed some gate disturbance, slightly diminished tendon reflexes and a slight weakness of the lower limb muscles, as well as urinary incontinence. Her parents refused a surgical intervention. The patient underwent a computed tomography (CT) and magnetic resonance (MR) examination.

Radiologic Features

The CT scans showed a broadened 4th right rib, a missing proximal part of the 5th rib, agenesis of the 6th and 7th ribs, and bifurcation of the 8th rib, as well as a fusion of the 8th and 9th left ribs (Fig. 1). As regards the vertebral

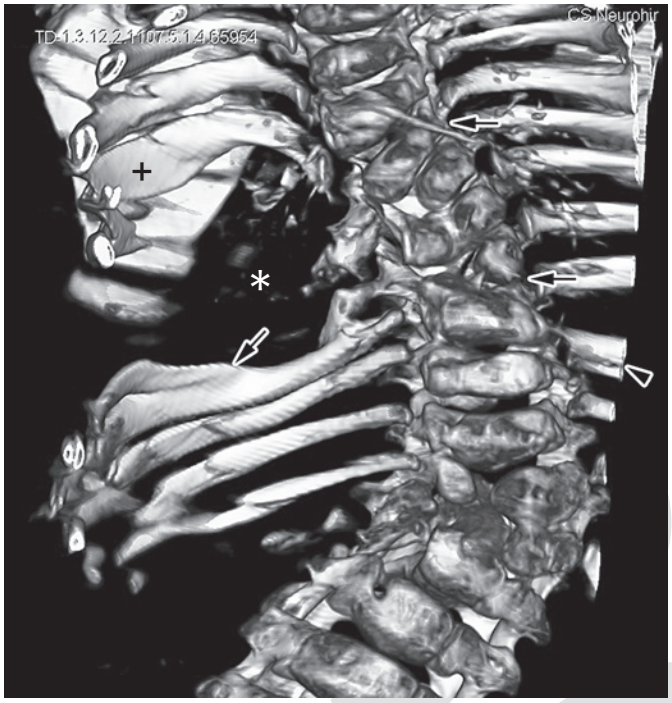

Fig. 1. Anterior view of the thoracic cage on a 3D CT image. Note the missing ribs (*), a broadened (+) and a bifurcated rib (arrow) on the right side, fused ribs (arrowhead) on the left side, and "pebble beach" thoracic vertebrae malformations (between the two arrows on the left side).

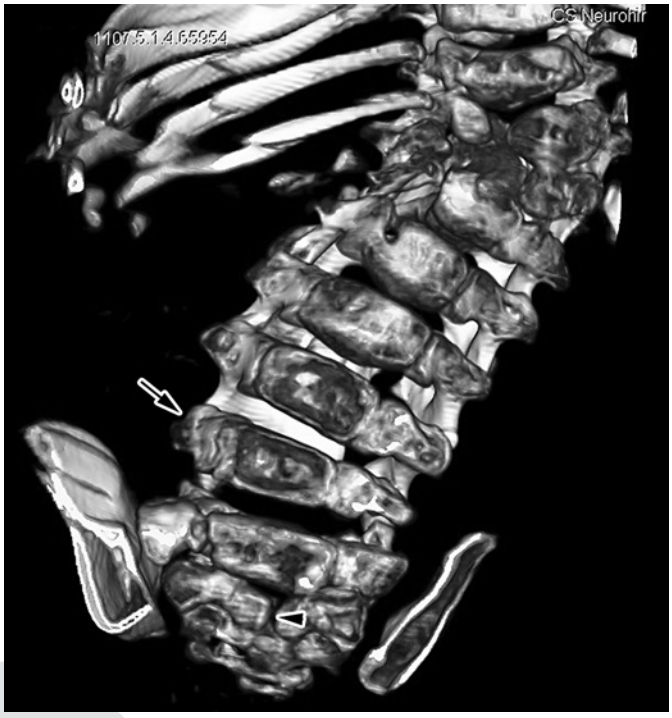

Fig. 2. Anterior view of the lumbosacral spine on a 3D CT image. The arrow points to the L5 vertebra. Note a hypoplastic sacrum with a cleft (arrowhead) of the S2 vertebra.

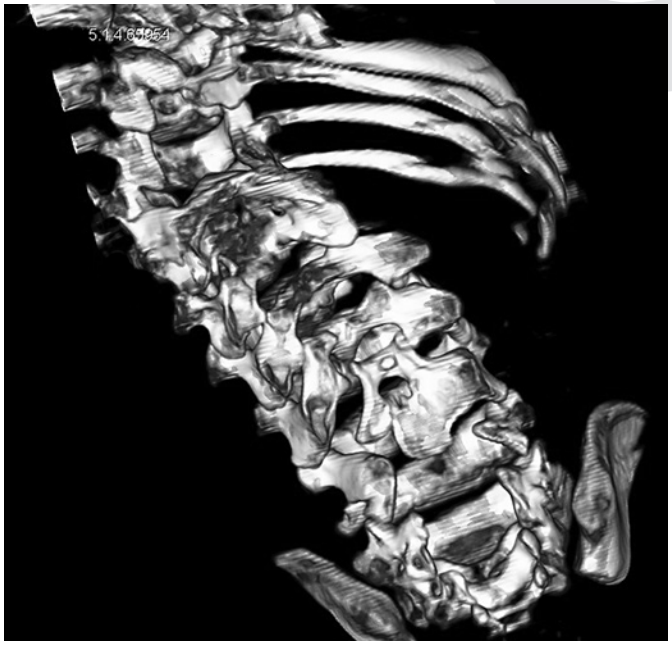

Fig. 3. Posterior view of the lower thoracic and entire lumbosacral spine on a 3D CT image. Note a long spina bifida.

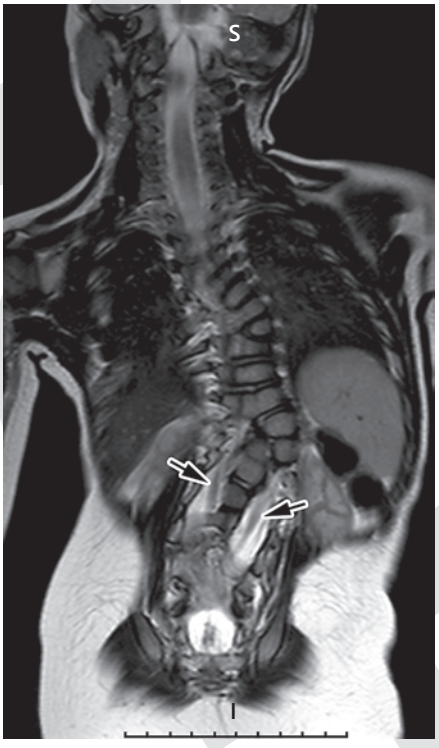

Fig. 4. Split spinal cord (arrows) in the lumbar region on a T2-weighted coronal MR image.

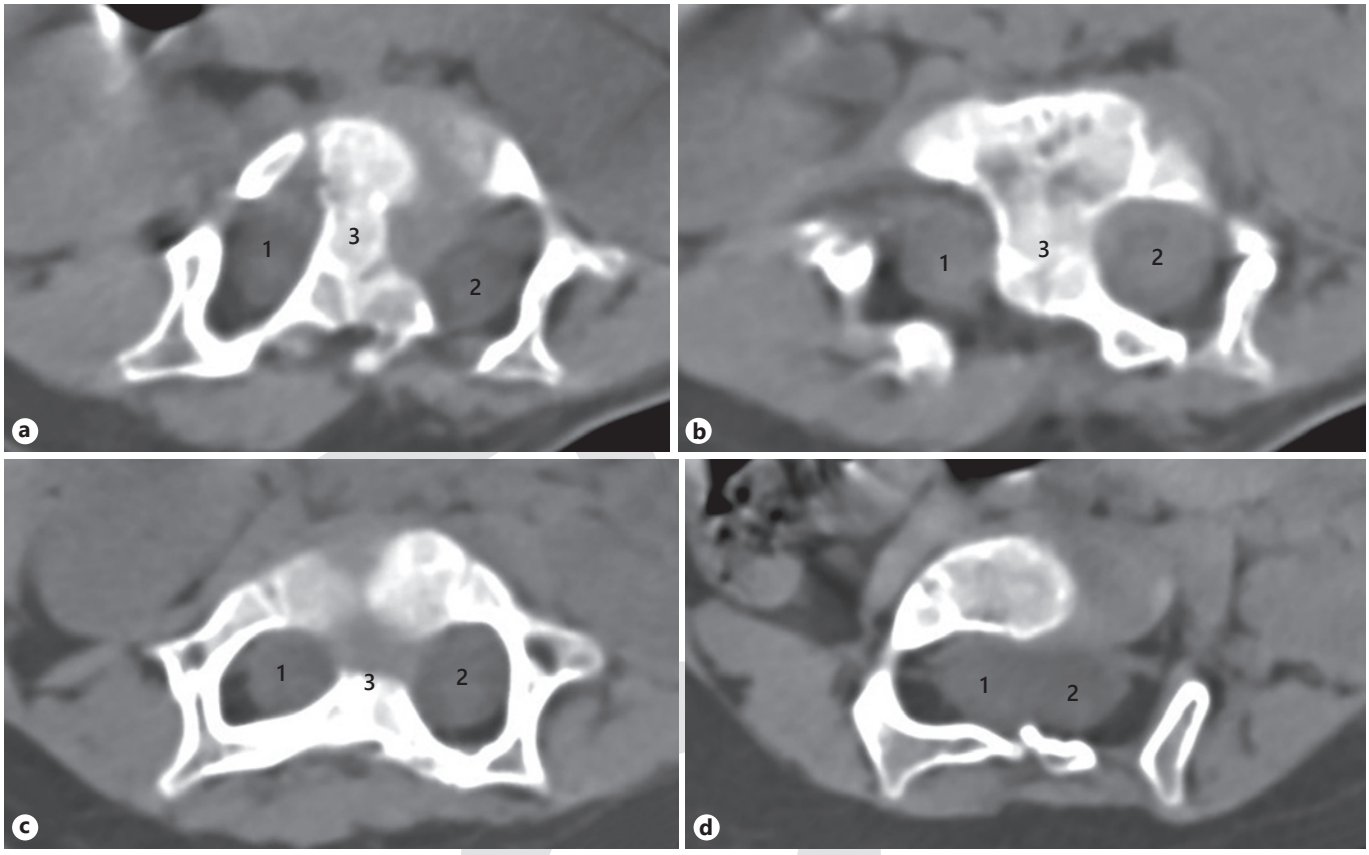

Fig. 5. Split spinal cord (1 and 2) with a large bony septum (3) on axial CT scans (**a–d**).

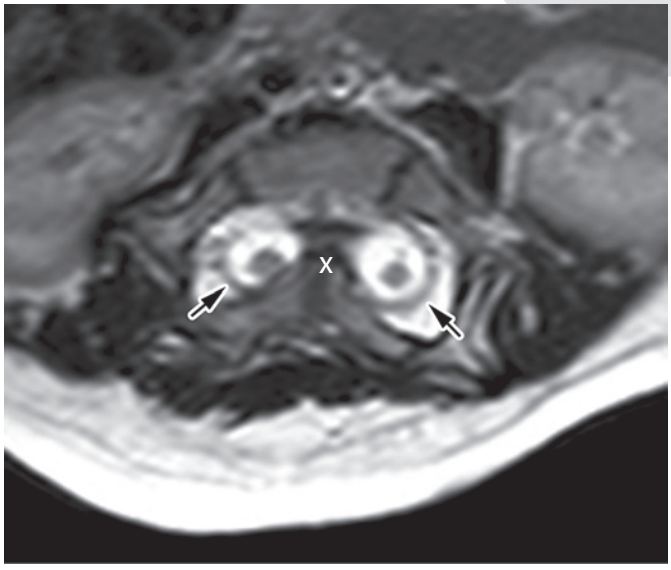

Fig. 6. The "owl eyes sign" on a T2-weighted axial MR image. Note the double dural tube (arrows) and a large septum (X) between the two hemicords.

bodies, multiple hemivertebrae, the butterfly, hypoplastic, and cleft vertebrae were seen in the thoracic (T) spine from the T5 to the T7 vertebra, and at the level of the T12 and the first lumbar (L1) vertebra, with a local formation of "pebble beach" vertebrae (Fig. 1). The sacrum consisted of only three vertebrae, the middle of which (S2) showed a median cleft of its body (Fig. 2). Further, an incomplete cleft of the left vertebral peduncle was noticed in T1 and T2, and a missing right peduncle in T4. Rachischisis of the vertebral arches was observed as well, extending from T3 to T9. Agenesis of the arches (spina bifida occulta) was seen at the level of T10 and T11, as well as between L3 and the terminal part of the sacrum (Fig. 3). Due to the mentioned malformations, congenital kyphosis and scoliosis with a double curve were expressed (Fig. 1 and 4).

The CT and MR also showed a bony spur along the vertebral canal, or an osteocartilaginous septum at a lower level, with a complete or partial division of the canal. At the same time, a split spinal cord was observed as well, with laterally positioned roots of the spinal nerves (Fig. 4 and 5). The two hemicords extended from the level of the

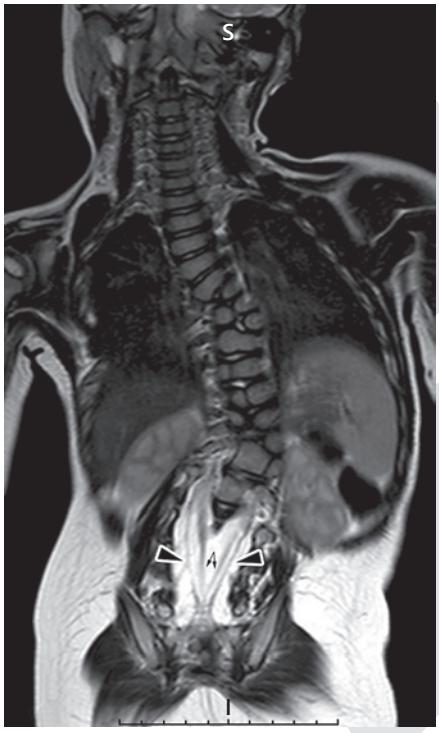

Fig. 7. Duplicated and thick filum terminale (small arrows), and a singular dural sack (arrowheads) on a T2-weighted coronal MR image.

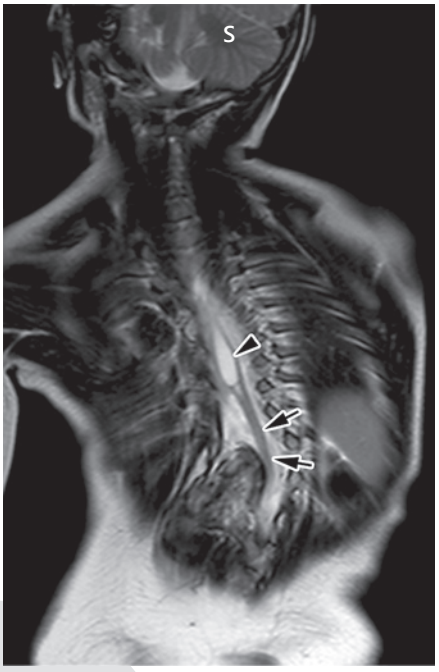

Fig. 8. Syringomyelia (arrowhead) as well as the thoracic spinal cord (arrows) just above the split cord on a T2-weighted coronal MR image.

T9 to the end of the conus medullaris (Fig. 4). A partial reunion of the cord was seen in one segment (Fig. 5d), but below that level the two hemicords were separated again (Fig. 4). As regards the dural tubes, they were observed to surround each hemicord separately (Fig. 6) to the level of the L4 vertebra, where they fused into a singular sack (Fig. 7). The filum terminale was seen to be thick and duplicated (Fig. 7). In addition, syringomyelia (Fig. 8) was noticed in the thoracic cord, extending from the T5 to the T8 vertebrae. It was 30 mm in length and 7 mm in width. A lipomatous swelling was observed beneath the skin of the thoracolumbar region, and finally a small meningocele at the level between T10 and T11.

Based on the mentioned clinical and radiologic examinations, a diagnosis of SCD and the associated anomalies was made.

Embryologic Basis

Certain genes are important for a proper embryonal development of the vertebral column and ribs, but also of the neural tube [5]. Their mutations result in a disorder of the formation and segmentation process of the vertebrae and ribs, as well as the occurrence of certain dysraphic malformations, especially a split spinal cord and spina bifida [6].

Normally, during the first 2 weeks of development (Fig. 9a), the primordial disc is formed which consists of the epiblast and hypoblast cell layers [6]. In the 3rd week of gestation (Fig. 9b and c), some cells of the epiblast displace the hypoblast to create the endoderm, others form the mesoderm (fig. 9B), whilst the remaining cells create the ectoderm (Fig. 9b and c). At the same time, the longitudinal notochordal plate is formed (Fig. 9c) which soon becomes the cylindrical notochord (Fig. 9d and e).

The notochord induces the formation of the primordial nervous system [6]. Namely, the median part of the ectoderm transforms into the neural plate (Fig. 9d) that soon invaginates to form the neural groove and then the neural tube (Fig. 9e and f), whose caudal part gives rise to most of the spinal cord. Soon after, the tip of the conus medullaris and filum terminale develop, as well as most of the sacrum and coccyx [6].

Meanwhile, the mesoderm close to the neural tube forms the somites (Fig. 9e and f), each of which differentiates into

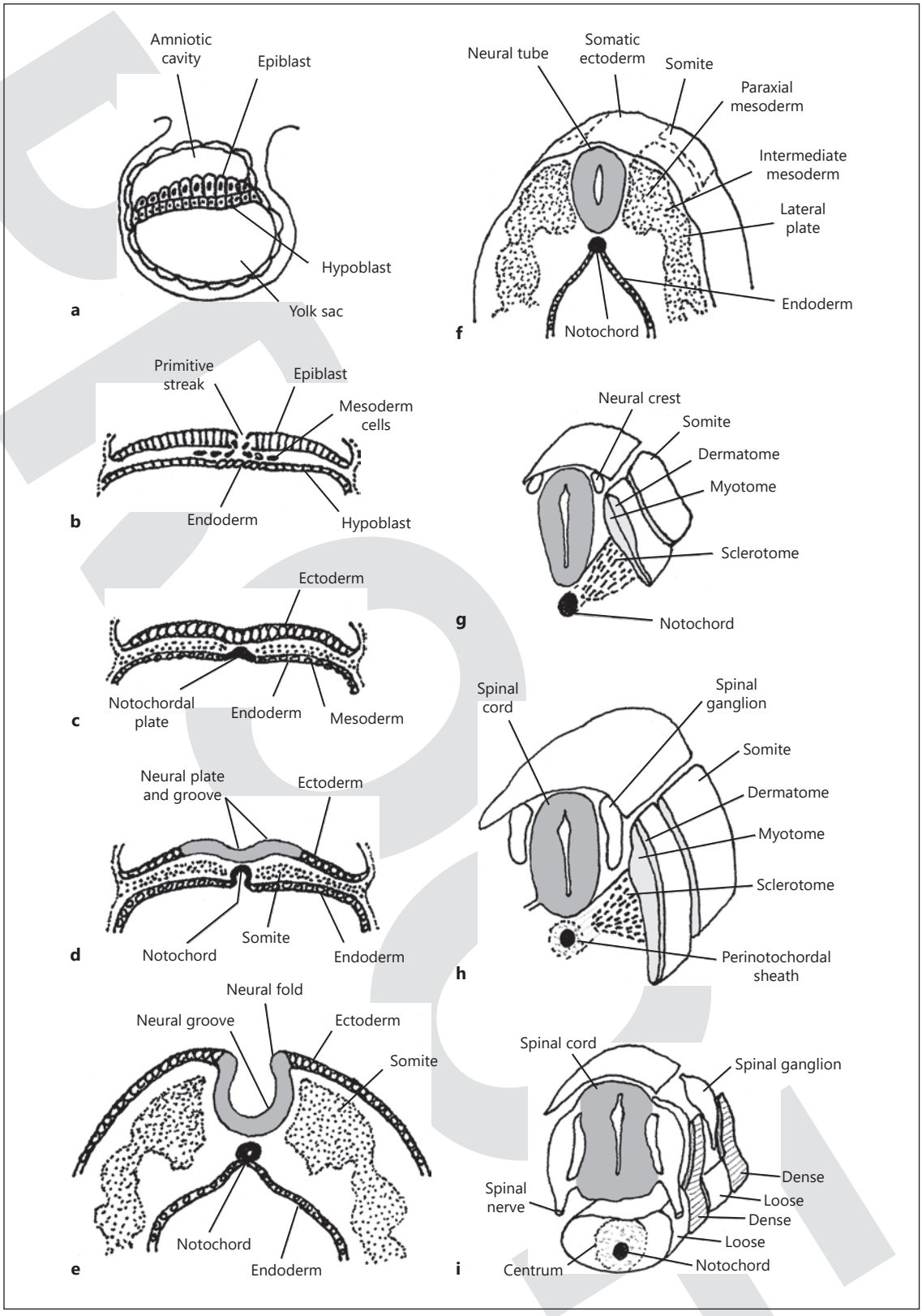

Fig. 9. a–i Early stages of the embryonal development of the vertebrae and the spinal cord. Modified after O'Rahilly and Müller [6].

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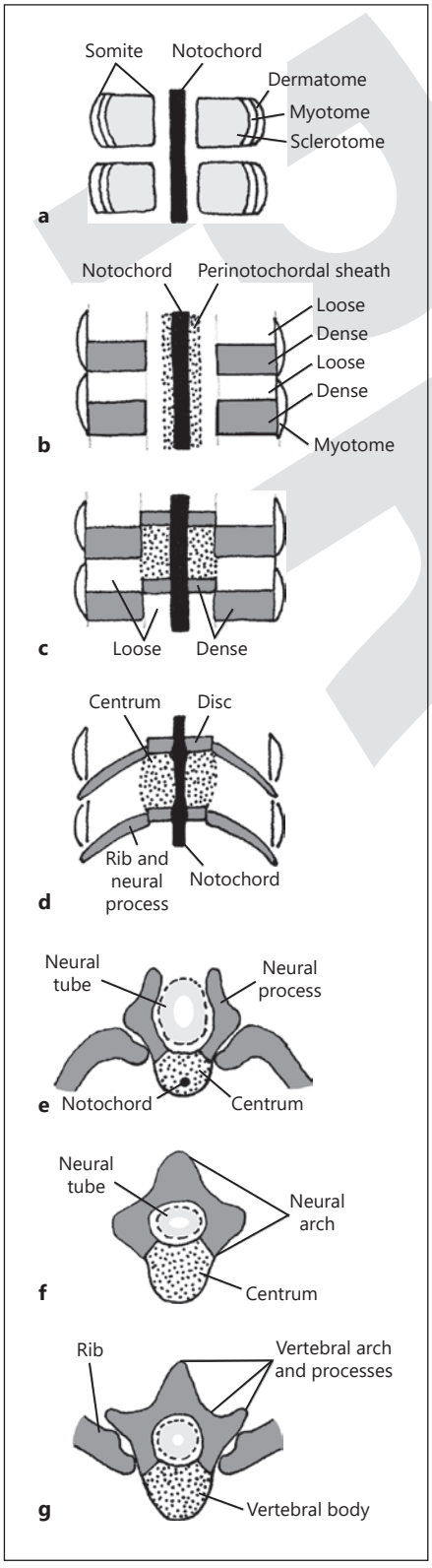

Fig. 10. a–g Embryonal development of the vertebrae and ribs (ribs are not presented in **f**). **a–d** Anterior view. **e–g** Superior view. Modified after O'Rahilly and Müller [6].

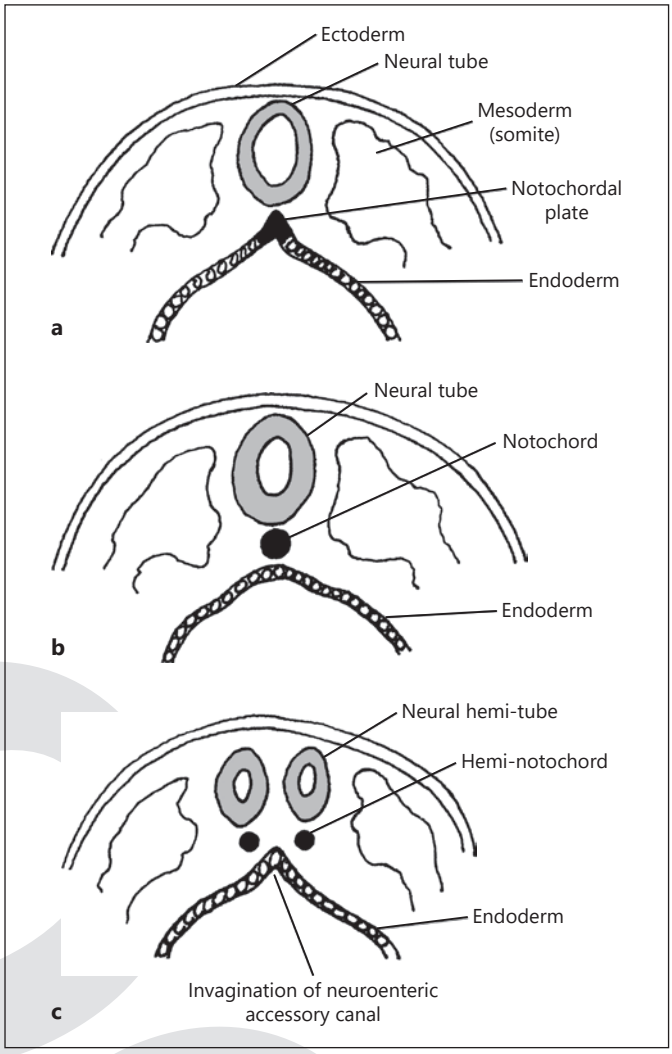

Fig. 11. A possible mechanism in split cord development. Note a normal (**a** and **b**) and anomalous (**c**) development. Modified after Pang et al. [7] and Andro et al. [8].

the dermatome (later forming dermis), myotome (forming muscles), and sclerotome (Fig. 9g) which create the vertebral column and ribs [6]. Namely, the cells of the sclerotome (Fig. 9g and h, and 10a) create the perinotochordal sheath (Fig. 9h and 10b) which differentiates soon into the cephalic loose areas and the caudal dense zones (Fig. 9i and 10b). The medial loose zone at each level will form the vertebral centrum, i.e. the future vertebral body (Fig. 9i and 10d through g), whilst the medial dense region at the same level, together with the notochord (the future nucleus pulposus), will become the intervertebral disc (Fig. 10d). The paired lateral dense areas (Fig. 10c) will form the neural process (arch) on each side (Fig. 10e), which soon fuse with each other (Fig. 10f) to form the future arch, pedicles, and

processes of each vertebra (Fig. 10g) [6]. At the same time, they will also make up ribs at this level (Fig. 10d, e, and g).

As regards the split cord, it is probably caused by the persistence of an accessory neuroenteric canal in the midline of the embryo (Fig. 11c) [7, 8]. This canal induces a division of the notochord, which may later result in various vertebral malformations. At the same time, the formed hemi-notochords induce in turn a separation of the neural tube into two halves at this level (Fig. 11c).

Our patient obviously had multiple embryologic disorders of the ribs, vertebrae, and sacrum, including spina bifida, which were associated with split spinal cord type I, syringomyelia, a small meningocele, a dorsal lipomatous swelling, and foot deformities. To our knowledge, such a combination of SCD and associated malformations has never been reported.

Statement of Ethics

Parents have given their written informed consent to publish the case of the little patient, including publication of images.

Disclosure Statement

There is no conflict of interests.

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