CLINICAL REPORT

Split cervical spinal cord malformation and vertebral dysgenesis

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Summary We report a case of vertebral malformation associated with diplomyelia believed to be a type II split cord malformation. Cervicothoracic level cases are exceptional. This article reports the case of an 11-year-old boy with no neurological symptoms who had not undergone surgery. The diagnosis was made during pregnancy by prenatal screening with ultrasound and MRI. Several embryological theories have been offered to provide an explanation for this syndrome. Close follow-up is mandatory. Surgery must only be considered if neurological deterioration occurs.

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Introduction

We report an exceptional case of split cord malformation (SCM), associating diplomyelia and cervicothoracic vertebral malformation. These abnormalities are more frequently observed in the thoracolumbar region [1]. To date, only about 30 cases with cervical segment involvement have been described and each presented morphological specificity [2–5]. After the presentation of our patient, the embryological hypotheses and the anatomical and clinical characteristics of this remarkable case will be discussed. Finally, we will discuss the main therapies and the course to follow.

Observation

An 11-year-old boy was born at term by vaginal birth. He was the second child of two children. There was no family history and no evidence of consanguinity. At 28 weeks amenorrhea of pregnancy, the ultrasound examination revealed a malformation associating intrauterine growth delay syndrome, type A omphalocele, and a cervicothoracic spine malformation.

These abnormalities called for prenatal amniocentesis karyotyping, which was normal (46XY).

The prenatal MRI showed small lateral evaginations of the spine, but did not provide a precise etiological diagnosis for this malformation.

At birth, the neurological exam was normal and no other associated locomotor system or visceral malformation was found. The integument, particularly the median cleft, was normal. The first radiographs taken at birth showed a spinal
malformation extending from C3 to T7, with a split spine aspect.

The CT exam demonstrated that the cervico-occipital hinge was normal. In the spine, only the first and second cervical vertebrae were normal. The other cervical and thoracic vertebrae, down to T7, were split in two. The vertebral bodies and the posterior arcs were not closed at the maximum gap of the two hemicords, resulting in an anterior and posterior bone defect (Figs. 1 and 2).

This abnormality was associated with rib malformations, responsible for a deformity in the thoracic cage.

The medullary canal MRI showed a dural sheath that followed the spinal deformity. At the cervicothoracic junction, there was a division of the medulla forming diplomyelia pulling laterally to the right and left, with the nerve roots of the upper limbs emerging from this area. An osseocartilaginous septum could not be demonstrated between the two hemicords (Fig. 3). There was no encephalic abnormality.

The child was 11 years old at the time this article was written. He presented a balanced growth delay of $-2$ standard deviations, his psychomotor development was normal, as was the neurological exam. He was 129 cm tall. He attended middle school and took part in recreational sports. From a morphological point of view, he presented a short neck, reduced cervical range of motion, and low posterior hair implantation (Fig. 4). He underwent respiratory physical therapy for restrictive pulmonary syndrome, which was responsible for frequent bronchopulmonary superinfections. Until now, the child has received simple medical and surgical monitoring. No surgical treatment has been undertaken.

Discussion

This type of malformation is exceptional. To date, few cases of cervicothoracic vertebral-spinal column malformations have been described in the literature.

Clinically speaking, this case suggests Klippel-Feil syndrome. This syndrome associates the classical clinical triad of low posterior hair implantation, a short neck, and limited range of motion in the neck found in this child. From an anatomical point of view, this syndrome is characterized by congenital fusion of the cervical vertebrae. In the present case, there was no vertebral fusion, but rather a split spine aspect, eliminating Klippel-Feil syndrome. However, David et al. [2] reported seven cases of Klippel-Feil...
The patient presents as with Klippel-Feil syndrome: a short neck and low posterior hair implantation.

Definition of split cord malformation (according to Pang et al. [6])

Notochordal-dysraphic disorders are rare congenital vertebral-spinal malformations, including SCM. The varied radiological and clinical forms of this syndrome partially explain the controversy on its exact definition. Several authors have established definitions. According to Pang et al. [6], there are two types of SCM. Type I, or diastematomyelia, consists of two hemicords in two individual dural tubes, separated by a rigid osseous or cartilaginous septum. Type II, or diplomyelia, corresponds to two hemicords housed within a single dural sac, separated by a fibrous septum. The case reported herein is similar to type II SCM. These spinal malformations are often associated with vertebral abnormalities: hemivertebrae, butterfly vertebrae, vertebral fusions, or spina bifida. However, a complete division of the spinal cord as in our case is exceptional.

This syndrome's clinical and radiological presentation is highly variable, with, however, a predominant involvement of the thoracolumbar segment and a higher frequency in girls. Diagnosis is most often made during childhood, at a mean four years after the appearance of the first symptoms. However, the diagnosis can also be late because when there is no clinical manifestation [5]. Currently, prenatal imaging techniques (ultrasound and MRI) can detect this type of abnormality during pregnancy.

Normal and pathological embryology

The etiopathogenesis of SCM syndrome has not been clearly established to date (Fig. 5). Several embryological hypotheses have been developed: abnormalities in the neural tube, doubling of the cord, persistence of an abnormal neurenteric canal.
The development of the spinal column and cord begins in the third week of embryonic development with the differentiation of the germ layers (ectoblast, mesoblast, and endoblast) [8]. The key structure in this development is the notochord, which emerges from the endoderm. The notochord constitutes the axis around which all the other structures later develop. During primary neurulation, the notochord induces the formation of the neural plate at the ectoblast level. This will then invaginate to form the neural tube, the source of the spinal cord. The formation of the vertebral column occurs toward the 17th day of embryonic life: mesmerization of the para-axial mesoblast forms pairs of somites from the action of the signals emitted for the most part by the notochord and the neural tube. The cells of the medial and ventral walls are deposited around the cord to form sclerotomes, which fuse to give birth to future vertebral bodies. The cells of the somites’ dorsal walls migrate around the neural tube and form the posterior arches (Fig. 5A, B).

Cervical SCM occurs during primary neurulation (Fig. 5C). According to Pang et al. [6], a lesion on the medial line from adherence between the endoderm and the ectoderm creates an accessory neurenteric canal. This canal induces the division of both the notochord and the neural plate at one segment of the embryo. The two resulting hemi-notochords behave like two autonomous cords, which each induce a neural tube and somites. At the end of development, the spinal cord that is separated into two, frequently associated with vertebral abnormalities in one segment of the vertebral column.

In type I SCM (diasstomatomyelia), the existence of two individual dural tubes, separated by a rigid septum, could be explained by the incorporation between the two hemitubes of primitive cells to the sclerogenic potential. They form a median dural wall and an osseocartilaginous septum between the two hemicords [6].

At the cervical level, a fibrous septum is described most frequently (type II SCM or diplomyelia), as in the case reported herein.

In addition, the vertebral column of the child in this report presented an aspect of separation into two hemi- cords, which is explained by the simultaneous presence of an anterior and posterior bony defect. In 1996, David et al. [2] reported seven cases of cervical SCM associated with vertebral abnormalities. They classified them into two groups: those having a defect in the posterior arch and those presenting an anterior osseous defect. This report provides an embryological theory for each case. The posterior defect may be related to a neural tube-closing abnormality, whereas the anterior defect may be secondary to invagination of the endoderm, preventing fusion of the sclerotomes. Our case is even more remarkable in that the two defects occur simultaneously.

Clinical considerations

The indications for surgical treatment in cervical SCM remain highly controversial. The appearance of medullary pain conditions the course to follow.

The cases of cervical SCM reported in the literature are mostly asymptomatic. One explanation would be that in the cervical region there is less often a rigid septum, and like the difference in growth speed between the spinal cord and the vertebral cord, it would be less substantial in this region[4,9], spinal shearing and adherence phenomena (tethered spinal cord) may be less frequent.

Finally, microinjuries related to flexion and extension movements of the vertebral column may be limited because of cervical stiffness.

Most authors do not recommend prophylactic surgical treatment. Close clinical monitoring is recommended for these patients. The appearance of neurological signs should open the discussion of surgical management, particularly if they are evolving.

Conclusion

SCM syndrome is rare and its varied anatomical and clinical forms make it difficult to study and develop standardized management.

The development of prenatal imaging with ultrasound and MRI allowed us to make a very early diagnosis for this case and follow up the patient since birth. Management should be multidisciplinary and individualized since each case is unique.

When present, the neurological deficit is often at the forefront, but it should not overshadow the pulmonary repercussions and the deformation of the rib cage.

Recourse to surgery should be envisaged only if there is clinical deterioration.

Conflicts of interest

None.

References