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CLINICAL CASE REPORT

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Arnold Chiari Malformation With Sponastrime (Spondylar and Nasal Changes, With Striations of the Metaphyses) Dysplasia

A Case Report

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Abstract: SPOndylar and NAsal changes, with STRIations of the Metaphyses (SPONASTRIME) dysplasia (SD) is a dwarfing autosomal recessive syndrome, characterized by a variety of clinical and radiographic features, which form the basis for diagnosis. We describe the presentation of an Arnold Chiari malformation in a patient with a clinical diagnosis of SD. The malformation was successfully treated by decompression of the foramen magnum and elevation of the cerebellum, with complete resolution of pain.

We report a rare case of Arnold Chiari malformation in a patient presenting with clinical and radiographic features strongly suggestive of SD and be successfully treated.

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Abbreviations: CSF = cerebrospinal fluid, MRI = magnetic resonance imaging, SD = SPONASTRIME dysplasia, SPONASTRIME = SPOndylar and NAsal changes, with STRIations of the Metaphyses.

INTRODUCTION

SPOndylar and NAsal changes, with STRIations of the Metaphyses (SPONASTRIME) dysplasia (SD) is an acronym first used by Fanconi et al[1] to describe the primary features of a new autosomal recessive syndrome of skeletal dysplasia. The most striking features of this rare genetic disorder are as follows: a moderate dwarfism, with shortness of the lower limbs; midfacial hypoplasia, including a saddle nose giving a somewhat ''oriental'' facial appearance; frontal bossing; vertebral malformations; generalized mild osteoporosis; striated metaphyses; and delayed ossification of the carpal bones.[2].

Less than 20 undisputed cases of SD have been reported in the clinical literature. Recently, short dental roots, hypogammaglobulinemia, and cataracts have been added as other clinical features specific of the syndrome.[3] In our review of the English literature on SD, we did not identify dysplastic malformation of the craniocervical junction as a characteristic feature of SD. Therefore, the aim of our case report was to describe the presentation of an Arnold Chiari malformation in a patient with radiological typical SD and to describe the surgical management of the malformation to decompress the foramen magnum.

CASE DESCRIPTION

A 36-year-old man was referred to our department of neurosurgery, from a local clinic, for assessment of neck and bilateral shoulder pain. The patient reported that the pain had been continuously present for about 4 years, but had significantly increased over the previous 2 months. The patient had a unique physical appearance, shown in Figure 1, which included a short stature; prominent forehead; midfacial hypoplasia, including a flat nasal bridge and short-upturned nose; and prognathism. He was 136 cm tall, which is 6 standard deviations (SDs) below the population mean height for males, and weighed 44 kg, 2 standard deviations below the population mean weight for males. Plain radiographs identified an Arnold Chiari malformation, which the patient reported had been previously diagnosed with, owing to concave vertebral bodies, posterior scalloping, and metaphyseal striation (Figure 2). Spinal magnetic resonance imaging (MRI) showed mild descent of the cerebellar tonsils into the foramen magnum, with syringomyelia from C2 to T7 (Figure 3). The short stature, facial dysmorphism, and radiographic skeletal findings were consistent with SD, and decompression of the foramen magnum was recommended to correct the tonsillar descent and alleviate the patients' pain.

Upon opening of the dura matter, adhesiolysis was performed for nonsevere adhesions of the arachnoid. This was followed by tonsillar cauterization, which led to a sudden release of cerebrospinal fluid (CSF) from the fourth ventricle (Figure 4). A spontaneous CSF leak occurred at the surgical site on postoperative day 3, which was managed clinically using continuous lumbar drain until complete resolution. The patient's neck and shoulder pain improved gradually without sequelae. Follow-up MRI, performed 2 months postsurgery, showed a marked decrease in the size of the syringomyelia (Figure 5).

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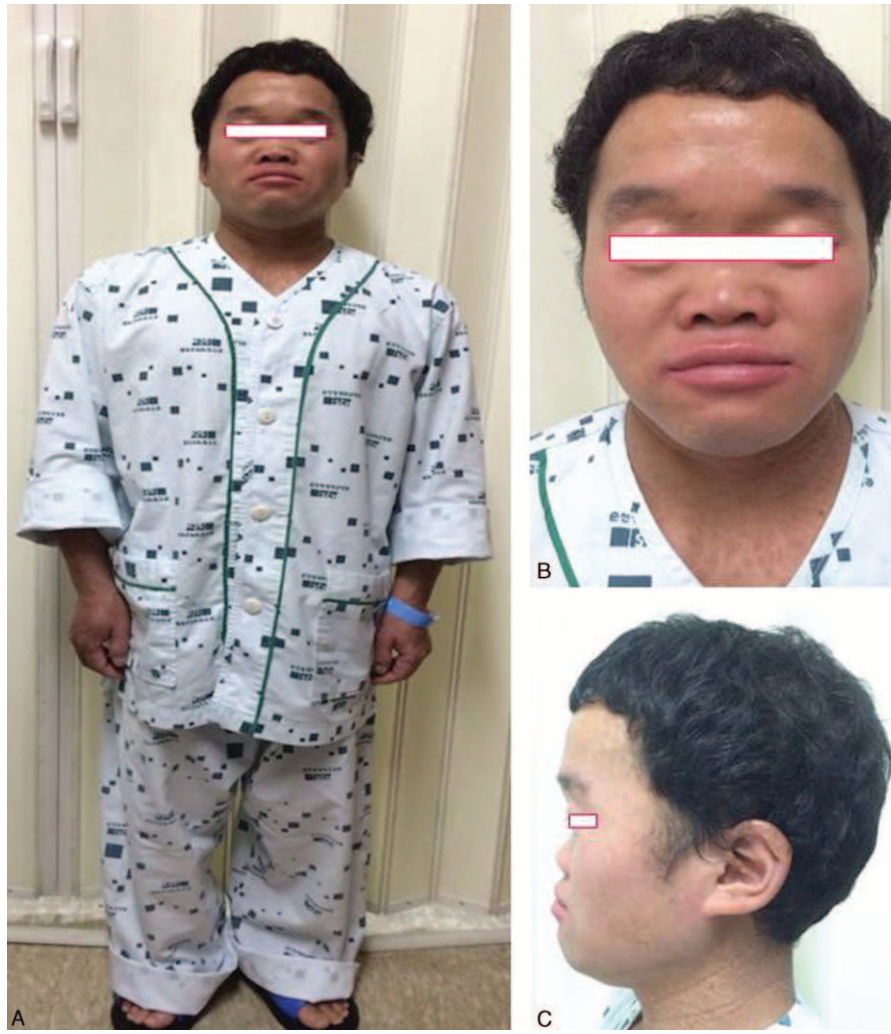

FIGURE 1. Facial (A) and full-body view (B) of the 36-year-old male patient reported in our case. The facial view shows midface hypoplasia, a short-upturned nose, and a prominent forehead.

DISCUSSION

Variants of the SD syndrome have recently been reported, indicative of a possible heterogeneity of this genetic syndrome.[4–6] In our review of the English literature, we identified 13 reported cases of SD,[1,4,7] with description of an associated Arnold Chiari malformation in only 1 case.[2] With regard to the genetics of SD, both autosomal-recessive and X-linked inheritance of the syndrome have been suggested.[1,2,4,7] However, an X-linked inheritance is unlikely has both males and females can be severely affected; meanwhile, the radiographic skeletal changes tend to be more severe in males than females, suggests that SD is an X-linked inheritance of the syndrome.[2]

The patient in our case presented with a number of clinical features of SD, including short-limbed dwarfism, relative macrocephaly, frontal bossing, midface hypoplasia, a saddle nose, and normal intelligence.[8] Radiographic findings of marked skeletal dysplasia included marked delay of epiphyseal ossification, evidence of metaphyseal dysplasia, and metaphyseal striation of the long bones.[8] The radiographic finding most specific to SD was the characteristic ovoid configuration of the vertebral bodies, identified on plain, sagittal plane, radiographs, with relatively small vertebral bodies of the lumbar spine, with concomitant moderate narrowing of interpediculate distances.[2,9] Therefore, the clinical manifestations of the 36-year-old man described in our case report are strongly suggestive of a diagnosis of SD.

Only a few investigators have reported craniocervical dysplasia as one component of the SD syndrome. Two plausible pathways could lead to the development of an Arnold Chiari malformation in patients with SD. From an embryological perspective, the foramen magnum consists of cells of 2 different origins[10]; the chondrocranium, which forms the bones at the base of the cranium through endochondral ossification; and 2 masses of parachordal cartilage, derived from the sclerotomal regions of the occipital somites and located around the cranial end of the notochord, which grow extensively and fuse into a cartilaginous mass forming the base of the cranium and the boundaries of the foramen magnum.[11] After birth, the base of the cranium continues to grow at the sphenooccipital synchondrosis until adolescence. A disturbance of the formation of cartilage tissue and/or a defect in the synthesis of collagen and proteoglycans have been described as a component of SD,[7]

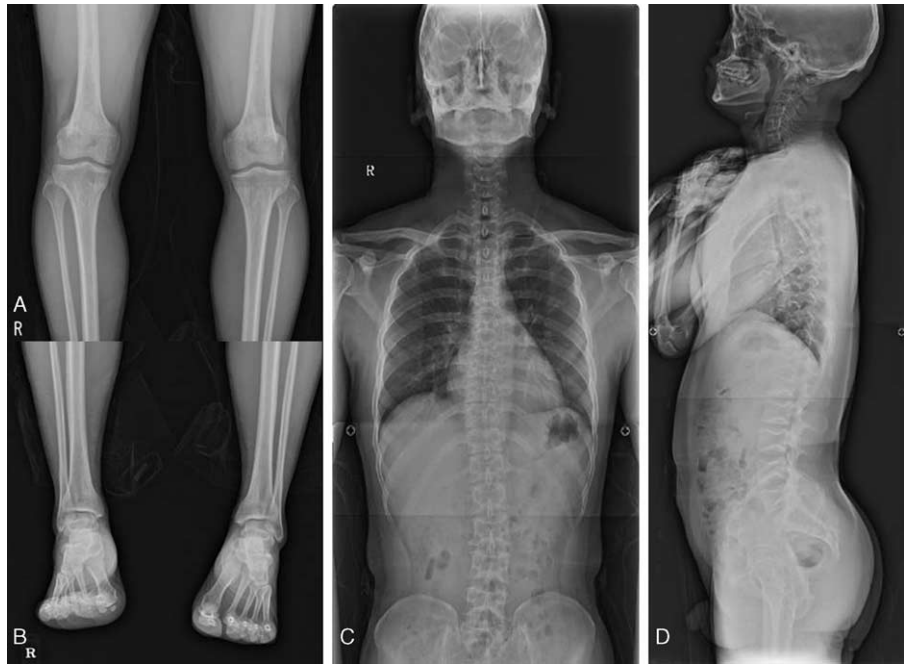

FIGURE 2. A, Computed tomography imaging of the cervical spine in the sagittal plane. B, Plain radiographs of the whole spine in anterior-posterior view, showing multiple pear-shaped abnormal vertebral bodies and thoracolumbar scoliosis.

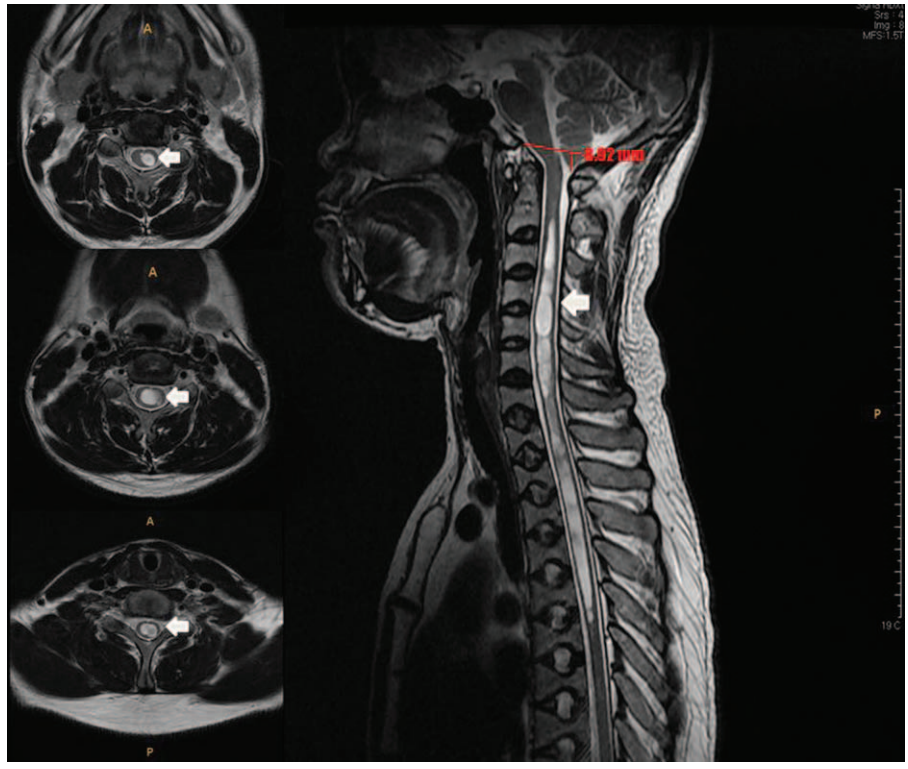

FIGURE 3. Magnetic resonance imaging showing extensive syringomyelia in the cervical and upper thoracic spinal cord, and mild downward herniation of the cerebellar tonsils. The pear- and biconcave-shape of the bodies of the vertebrae is evident.

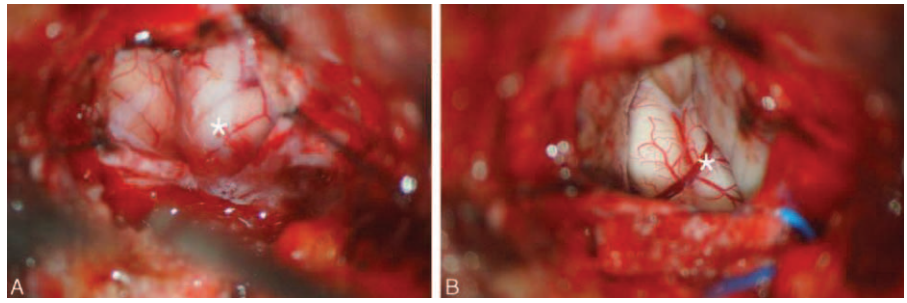

FIGURE 4. Intraoperative photographs showing (A) mild arachnoid adhesion at the level of cerebellar tonsils, bilaterally, and (B) the fourth ventricle after adhesiolysis and tonsillar cauterization.

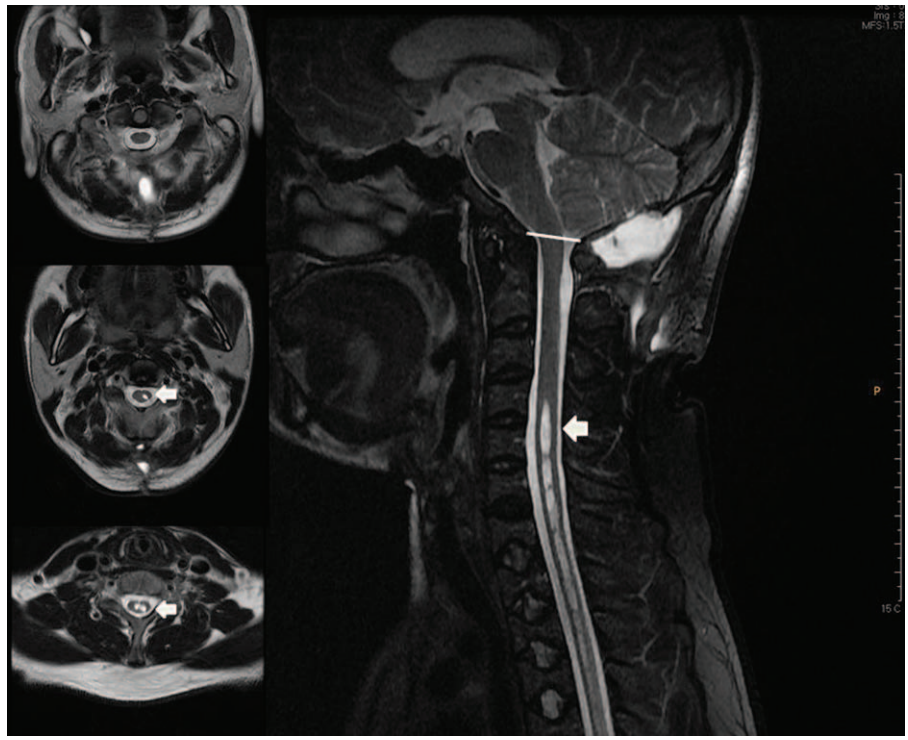

FIGURE 5. Follow-up magnetic resonance imaging, 2 months postdecompression of the foramen magnum, showing partial improvement of the extensive syringomyelia in cervicothoracic spinal cord.

which could result in craniocervical dysplasia at the level of the foramen magnum. It is also possible that the craniocervical dysplasia may be an overlapping feature of different groups of skeletal dysplasia, rather than a unique feature of SD. The 2010 revision of the Nosology and Classification of Genetic Skeletal Disorders[12] describes >450, well-characterized types of skeletal dysplasia, with clinical classification based primarily on clinical, radiographic, and molecular criteria, with widely overlapping features across the different types. Although craniocervical dysplasia is not a commonly reported feature of SD, Arnold Chiari malformation and syringomyelia, associated to malformations of the base of the skull, such as platybasia, a tight foramen magnum or hyperostosis of the base of the cranium, has been described in other syndromes of skeletal dysplasia, including achondroplasia, metaphyseal chondroplasia, and osteopetrosis.[12–15] From our case, we propose that craniocervical dysplasia may be a relatively rare finding of SD, with specific alterations in chondro-osseous morphology likely contributing to the involvement of the craniocervical junction in patients with SD.

CONCLUSIONS

We report a rare case of Arnold Chiari malformation in a patient presenting with clinical and radiographic features strongly suggestive of SD. The patient was successfully treated with decompression of the foramen magnum.

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