J Orthop Sci
DOI 10.1007/s00776-013-0520-0

CASE REPORT

Two siblings with neuropathic scoliosis caused by Chiari malformation type I with syringomyelia

Hiroshi Kuroki · Naoki Inomata · Hideaki Hamanaka · Kiyoshi Higa · Etsuo Chosa · Naoya Tajima

Received: 5 December 2013 / Accepted: 11 December 2013

Introduction

Chiari malformation type I (CMI) has been considered a sporadic condition without a heritable etiology; however, there have been a number of case reports identifying familial aggregation and clustering of CMI, suggesting a genetic basis [1]. We report two siblings with neuropathic scoliosis (NS) caused by CMI with syringomyelia. The patients and their families were informed that data from the cases would be submitted for publication and gave their consent.

Case presentation

Sibling 1 The patient was a 17-year 1-month-old boy who had a left convex thoracic curve with a Cobb angle of 70° between T7 and L1 (Fig. 1). He did not have any symptoms except spinal deformity. However, because of laterality of the abdominal skin reflex and a left thoracic curve pattern, MRI was performed. We diagnosed him as having NS caused by CMI with syringomyelia. The syringomyelia was first treated by foramen magnum decompression (FMD) (Fig. 2). After confirming the diminished size of the syrinx, posterior instrumentation and fusion were performed for scoliosis, and the Cobb angle improved to 45° postoperatively (Fig. 3). The patient was engaged in standing work without any inconvenience at 4 years after scoliosis surgery.

Sibling 2 The patient was a 15-year 8-month-old girl who had right convex thoracic and left convex lumbar curves with Cobb angles of 80° between T5 and T11 and 90° between T11 and L4 at the first visit to our clinic (Fig. 4). Her neurological findings, including the abdominal skin reflex, were normal. However, MRI was performed because not only had her elder brother been previously diagnosed with NS caused by CMI with syringomyelia, but also she had large major double curves. The MRI showed CMI with syringomyelia. Her Cobb angles were 90° and 90° before surgical intervention. The syringomyelia was first treated by FMD (Fig. 5). After confirming the diminished size of the syrinx, posterior instrumentation and fusion were performed for scoliosis, and the Cobb angle improved to 58° and 60° postoperatively (Fig. 6). She was employed as a care giver at 6 years after scoliosis surgery.

Discussion

Chiari malformations, originally described by Hans Chiari in the late nineteenth century, are regarded as a pathological continuum of hindbrain maldevelopments characterized by downward herniation of the cerebellar tonsils [2]. CMI has been traditionally defined as tonsillar herniation at least 3–5 mm below the foramen magnum [3].

The most common symptom of this disorder is pain, which is usually occipital or posterior cervical. Scoliosis is another important and common finding [4] in children with syringomyelia associated with CMI, with progressive scoliosis found in approximately 30 % of such children [5].

H. Kuroki (✉) · N. Inomata · H. Hamanaka · K. Higa · E. Chosa
Department of Orthopaedic Surgery, Faculty of Medicine, University of Miyazaki, 5200 Kihara Kiyotake, 889-1692 Miyazaki, Japan
e-mail: hiroshik@med.miyazaki-u.ac.jp

N. Tajima
Department of Orthopaedic Surgery, Nozaki Higashi Hospital, Miyazaki, Japan

Published online: 01 January 2014

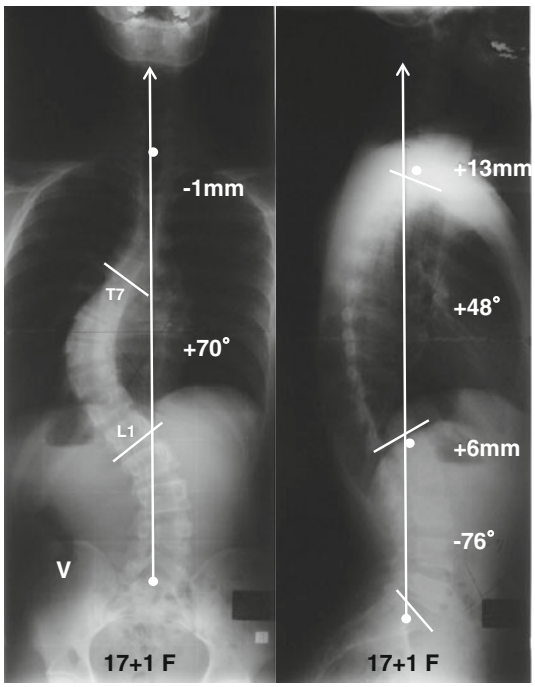

Fig. 1 Case 1. Preoperative X-rays. The patient had left convex thoracic scoliosis with a Cobb angle of 70° between T7 and L1

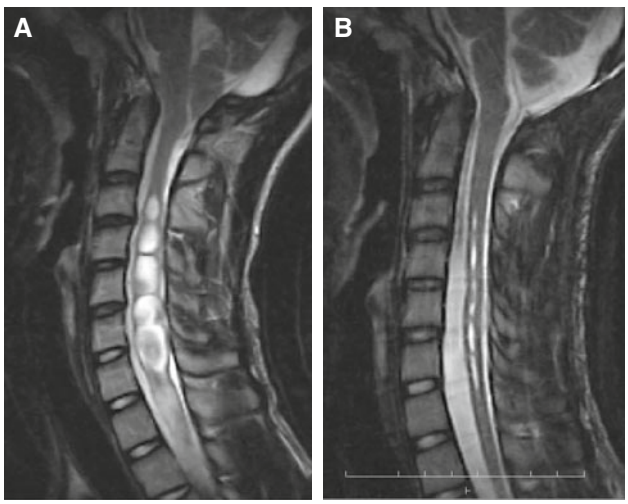

Fig. 2 Case 1. Preoperative and postoperative cervical MRI (T2WI sagittal). **a** Preoperative MRI CMI with syringomyelia was found by screening MRI. **b** MRI after foramen magnum decompression. Postoperative MRI revealed a decrease in the size of the syrinx cavity

The pathogenesis of most CMIs is unclear. Although Tubbs et al. [6] have shown that not all patients with a CMI will have a reduced cranial posterior fossa volume, the authors of most studies have concluded that CMI results from an underdeveloped paraxial mesoderm leading to posterior fossa hypoplasia [3]. Overcrowding in the posterior cranial fossa due to a normal-sized hindbrain induces a downward herniation of the brain as well as occlusion of

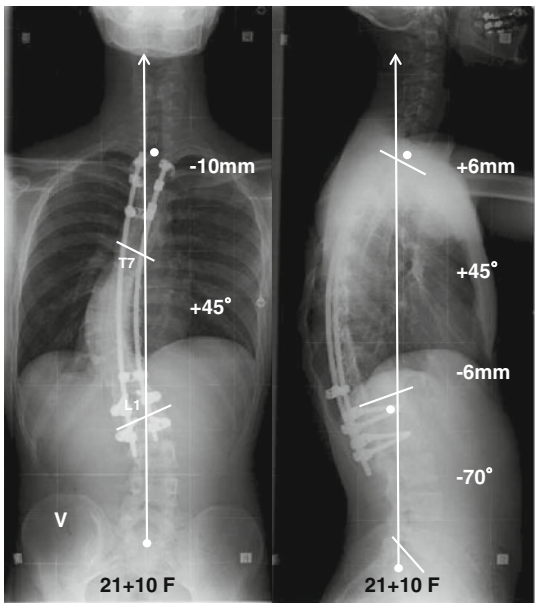

Fig. 3 Case 1. Postoperative X-rays at 4 years 3 months after surgery. Posterior instrumentation and fusion were performed, and the Cobb angle improved to 45°

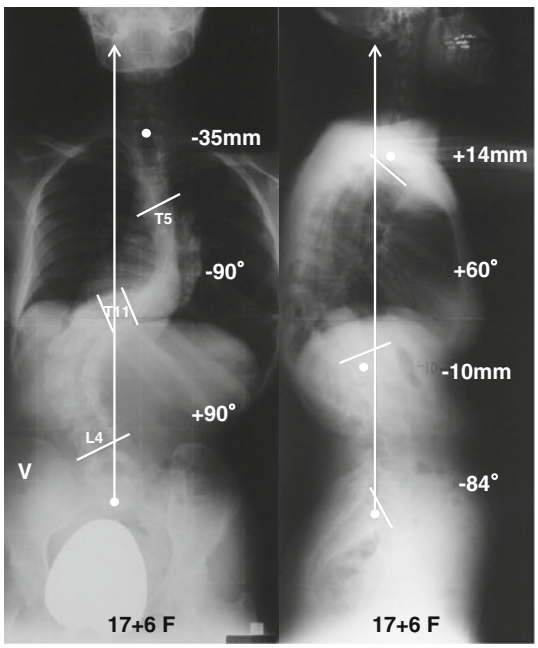

Fig. 4 Case 2. Preoperative X-rays. The patient had double major scoliosis with Cobb angles of 90° between T5 and T11 and 90° between T11 and L4, respectively

cerebrospinal fluid (CSF) flow across the foramen magnum [7]. This obstructs the natural flow of CSF and may be responsible for the origin and maintenance of syringomyelia by the pulsatile pressure waves forcing CSF into the spinal cord through the perivascular and interstitial spaces