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Clinical Observations

Effect of Intrathecal Baclofen on Delayed-Onset Paroxysmal Dystonia due to Compression Injury Resulting From Congenital and Progressive Spinal Bone Deformities in Chondrodysplasia Punctata

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ABSTRACT

BACKGROUND: Dystonia due to spinal lesions in adult patients is characterized by the provocation and/or amelioration of the spasm by somatosensory stimulation with a sensory trick. **PATIENT DESCRIPTION:** An infant with brachytelephalangic chondrodysplasia punctata developed flaccid tetraplegia due to cervical cord compression resulting from congenital atlantoaxial dislocation. Episodic, tonic extension of the extremities, neck, and trunk had appeared daily since age two years and was often provoked by tactile stimulation. Although decompression surgery was performed at age three years, progressive spinal deformity resulted in the aggravation of episodic dystonia thereafter, lasting for hours. Foot dorsiflexion and wearing a truncal brace for scoliosis inhibited these spasms. Intrathecal baclofen bolus injection transiently ameliorated the paroxysmal dystonia and detrusor-sphincter dyssynergia in the lower urinary tract. **CONCLUSION:** Paroxysmal dystonia is unusual in children with spinal cord lesions; however, it should be recognized for appropriate individualized clinical management.

Keywords: brachytelephalangy, spinal cord, myelopathy, atlantoaxial dislocation, paroxysmal dystonia, chondrodysplasia punctata, detrusor-sphincter dyssynergia, intrathecal baclofen

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Introduction

Chondrodysplasia punctata (CDP) is characterized by stippling in long bone epiphyses on x-ray, facial dysmorphism, and short stature. Several causative genes have been identified for the two known CDP types: brachytelephalangic CDP (BCDP) with X-linked inheritance and rhizomelic CDP with autosomal recessive inheritance. Arylsulfatase E (*ARSE*) gene mutations are found in two thirds of BCDP patients. In both CDP groups, cervical myelopathy due to spinal cord compression by abnormal bone formation can occur.[1-3]

We report a boy with BCDP who developed flaccid tetraplegia due to congenital atlantoaxial dislocation. He additionally manifested episodic extensor dystonia after tactile stimulation. Specific somatosensory manipulation alleviated

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the dystonic posturing. In addition, intrathecal baclofen bolus was effective in treating paroxysmal dystonia and lower urinary tract dysfunction.

Patient Description

This 5-year-old boy with unrelated parents presented with hypotonia, mild dyspnea, and poor sucking at birth. Neuroimaging revealed atlantoaxial dislocation with odontoid hypoplasia resulting in cervical spinal cord compression (Fig 1A). Tube feeding was initiated at age 1 month. Obstructive sleep apnea became evident at age 2 months and was managed by positive airway pressure therapy. Voluntary movements of sucking fingers, grasping toys, and limb elevation were rarely noted until age 3 months. Intermittent, brief hypertonia with extension of the neck and trunk accompanied by pronation and extension of the arms emerged by age 5 months. Cervical cord signal change was observed on magnetic resonance imaging at age 9 months (Fig 1B). Short-latency somatosensory evoked potentials did not yield brainstem or cortical responses since neonatal period. Gastrostomy and fundoplication, and tracheotomy were performed at age 10 months and 1 year, respectively, because of recurrent aspiration pneumonia.

He was diagnosed with BCDP at age nine months, based on the detection of stippling calcification in the epiphyses of the humerus and tarsal bones, facial dysmorphism with flat and anteverted nose, and

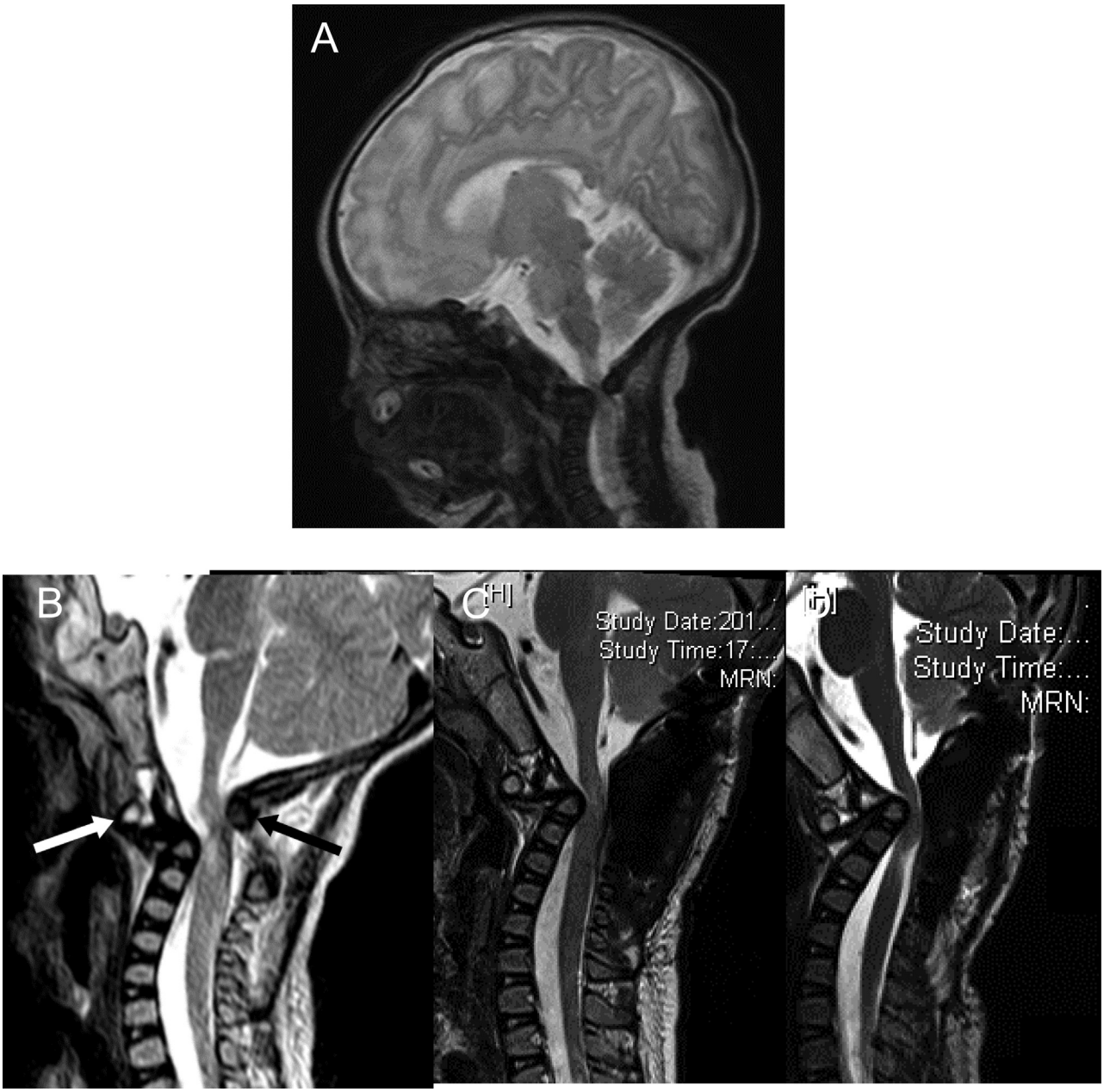

FIGURE 1.
Magnetic resonance imaging of the patient at age (A) 10 days, (B) 1 year and 3 months, (C) 3 years and 2 months, and (D): 5 years. (A) Compression of the medullospinal junction by bone structures is seen. Brain structures above the level of compression are normal. (B) Note that the C1 vertebra (white and black arrows) is anteriorly deviated, with posterior compression of the spinal cord (black arrow). (C) Compression was ameliorated after craniotomy and laminectomy. (D) The C2 vertebra and anterior edge of the foramen magnum protruding posteriorly, resulting in the aggravation of spinal compression.