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Spinal Deformity in Bethlem Myopathy

Ozgur Dede, MD[a,*], Hoda Z. Abdel-Hamid, MD[b], Vincent F. Deeney, MD[a]

[a]*Department of Orthopaedic Surgery, Children's Hospital of Pittsburgh of University of Pittsburgh Medical Center, 4401 Penn Avenue, Pittsburgh, PA 15224, USA*

[b]*Department of Pediatrics, Division of Child Neurology, Children's Hospital of Pittsburgh of University of Pittsburgh Medical Center, 4401 Penn Avenue, Pittsburgh, PA 15224, USA*

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Abstract

Study Design: Retrospective review.
Objectives: To report the characteristics of spinal deformity in a series of 3 patients with Bethlem myopathy.
Summary of Background Data: Bethlem myopathy presents with mild muscular weakness and typically has a benign course. Severe scoliosis in patients affected with Bethlem myopathy has not been previously reported.
Methods: Clinical records of 3 brothers with Bethlem myopathy were reviewed. Clinical and radiographic features of the spinal deformity are presented.
Results: All 3 patients had progressive scoliosis with coronal and sagittal imbalance. At a minimum of 26 months of follow-up after posterior instrumented fusion, there were no complications and deformity correction was maintained. Posterior instrumentation and fusion did not negatively affect the pulmonary function in this group of patients with Bethlem myopathy.
Conclusions: Bethlem myopathy may present with severe scoliosis along with proximal muscle weakness. This condition should be included in the differential diagnosis of adolescent patients with progressive spinal deformity.

Keywords: Scoliosis; Syndromic; Muscular dystrophy

Introduction

Collagen VI–related disorders are a group of muscle diseases of variable severity, classified under muscular dystrophies [1]. The milder end of the spectrum is Bethlem myopathy, which is characterized by proximal muscle weakness, joint contractures, and hyperlaxity. The other end of the spectrum, Ullrich myopathy, causes weakness early in life; the affected children may never walk [1,2]. Although one of the clinical features of Bethlem myopathy has been reported to be a rigid spine [3], the authors of the current study were not able to retrieve a description of scoliosis or severity of the spinal deformity in Bethlem myopathy from the extant medical literature.

The aim of this study was to report the association of severe scoliosis with Bethlem myopathy in a series of 3 patients.

Materials and Methods

Since 2005, the authors have seen 3 brothers with scoliosis and a diagnosis of Bethlem myopathy at their pediatric orthopaedic surgery clinic. A diagnosis of Bethlem myopathy was confirmed by a pediatric neurologist (HZD). All patients underwent posterior instrumentation and fusion by the senior author (VD). One patient with more severe deformity was operated on using an all–pedicle screw construct; the other 2 patients with less severe curves were operated on using hybrid constructs consisting of hooks, sublaminar wires, and lumbar pedicle screws. Surgical technique involved facetectomies on every level; however, no other extensile anterior or posterior release was done and no preoperative or intra-operative traction was used. Deformity correction was attempted with rod derotation maneuver and *in situ* rod bending. Stainless-steel

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*Corresponding author. Department of Orthopaedic Surgery, Children's Hospital of Pittsburgh of University of Pittsburgh Medical Center, 4401 Penn Avenue, Pittsburgh, PA 15224, USA. Tel.: (412) 692-9888; fax: (412) 692-6088.

E-mail addresses: ozgurdede@gmail.com or ozgur.dede@chp.edu (O. Dede).

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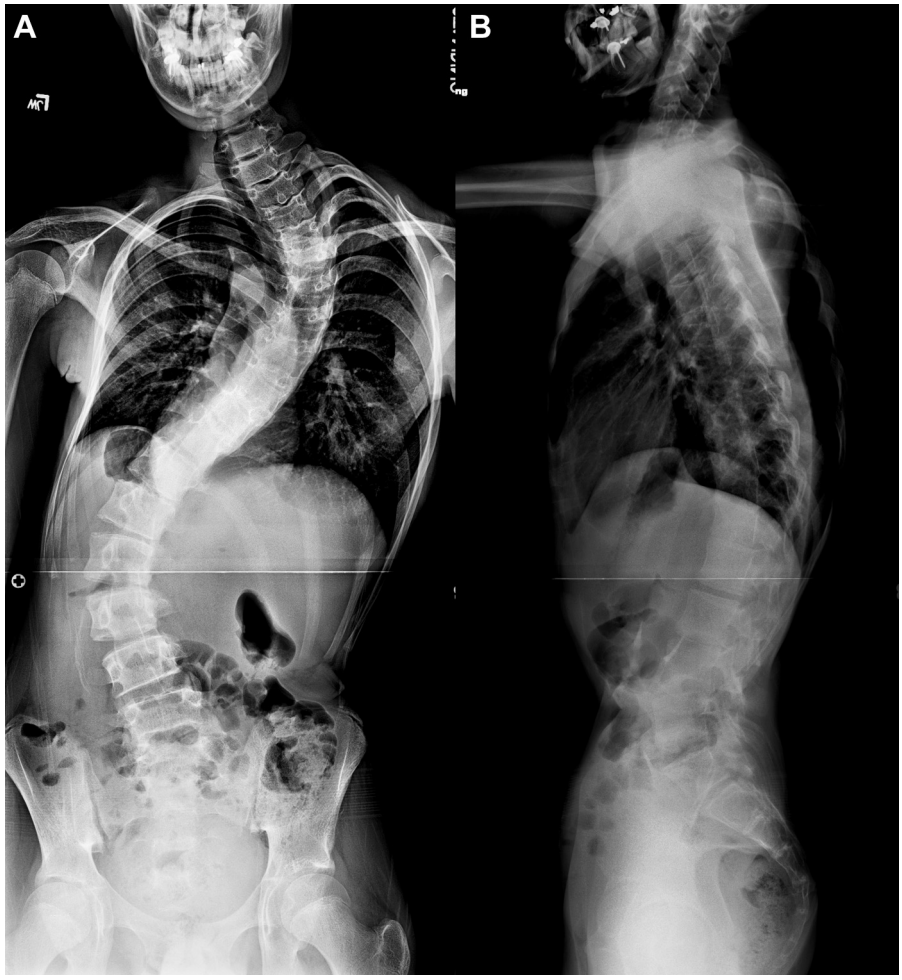

Fig. 1. Preoperative radiographs of Case 1, demonstrating the spinal deformity.

implants were used in all patients. Intra-operative neuromonitoring consisted of somatosensory evoked potentials and there were no changes in intra-operative signals. Shoulder balance (the angle formed by a line subtending the highest points of the clavicles and the horizontal), pelvic obliquity (the angle formed by a line tangential to both iliac crests and the horizontal), and coronal and sagittal spinal deformity measurements (using the sagittal vertical axis and central sacral vertical line as reference lines) were done on preoperative, postoperative, and follow-up radiographs. Preoperative and postoperative pulmonary function studies were assessed to evaluate the effect of posterior spinal fusion on pulmonary parameters.

Case 1

A 16-year-old boy with a prior history of progressive scoliosis was transferred to the authors' hospital with a diagnosis of respiratory failure. The patient had no other systemic signs or symptoms. History revealed that he had been an active adolescent, played basketball, and had had no symptoms (other than scoliosis) until recently. His mother noticed that his breathing had been labored during the previous day and he was difficult to arouse on the morning of hospital admission. When he was taken to the hospital, he was unresponsive and was subsequently intubated. His condition improved during the hospital course, with ventilator support. He was discharged to home with part-time bilevel positive airway pressure (BIPAP) support. During the evaluation, the patient was noted to have contractures of the elbows and the joints of both hands, and proximal muscle weakness. He had no previously known family history of muscle weakness. However, on further questioning, it was noted that his younger brother also had scoliosis and the same pattern of muscle weakness. The patient was examined for a variety of muscular disorders with muscle biopsy, electromyelogram, and genetics testing, and a diagnosis of Bethlem myopathy was established.

Assessment of the spine showed a double major curve pattern with an 86° right thoracic curve between T2 and T11 and an 85° left lumbar curve between T11 and L4 (Fig. 1). Iliac apophyses were graded as Risser 4. On side-bending X-rays, thoracic curve corrected to 47° and lumbar