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Case Report

Surgical correction of kyphotic deformity in a patient with Proteus syndrome

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Abstract

BACKGROUND CONTEXT: Proteus syndrome (PS) is an extremely rare congenital disorder causing asymmetric overgrowth of different tissues. The etiology remains unclear. Limb deformities are common and often necessitate amputations. Only a few cases associated with spinal deformities have been described.

PURPOSE: The aim was to report a rare case of PS associated with spinal deformity and its surgical management.

STUDY DESIGN: A case of young boy with PS causing vertebral hypertrophy and kyphoscoliotic deformity, which was surgically corrected, is presented.

METHODS: The patient was assessed clinically and with whole spine plain radiographs, computed tomography, and magnetic resonance imaging. Surgical correction was performed.

RESULTS: Satisfactory correction of the deformity was achieved by posterior spinal fusion with instrumentation from T4–L5, five Ponte osteotomies T8–L1, and an L2 pedicle subtraction osteotomy. The kyphosis was corrected from 87° to 55°; there was improvement in all spinopelvic parameters. One year after surgery, there was maintenance of the deformity correction with no deterioration of the sagittal balance, and the patient was free of pain and had no loss of neurologic function.

CONCLUSIONS: Proteus syndrome can be associated with spinal stenosis and deformity. Although the syndrome can be progressive in nature, the symptomatic spinal pathology should be treated appropriately.

Keywords: Proteus syndrome; Kyphosis; Spinal instrumentation; Spinal deformity; Overgrowth; Spinal osteotomy

Introduction

A patient with the signs of Proteus syndrome (PS) was first described in 1979 [1]. The syndrome was assigned its name several years later [2] after the Greek god of the sea, Proteus, who could change his form at will to avoid capture. The etiology remains unclear. However, there is evidence that it occurs as a result of mutation of the oncogene *AKT1*. This mutation is typically lethal, except in the setting of mosaicism. This gene encodes the AKT1 kinase, an enzyme known to mediate processes such as cell proliferation and apoptosis [3]. The syndrome is extremely rare, and its diagnosis is still controversial because of high interpatient variation and overlap with other asymmetric overgrowth syndromes.

Clinical presentation is typically asymmetric partial gigantism and hemihypertrophy, which can affect any body tissue; however, bone connective tissue and fat are the most commonly involved. Clinical findings include macrodactyly, subcutaneous tumors, plantar or palmar hyperplasia, cranial exostosis, kyphoscoliosis secondary to vertebral abnormalities, and other skeletal anomalies [4]. The clinical

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course is usually rapidly progressive in childhood in which the affected bones can become unrecognizably distorted. The progression may, however, slow or stabilize during early adolescence. The patients are prothrombotic, often leading to premature death due to deep vein thrombosis and pulmonary embolism.

We present a young male patient with PS causing vertebral hypertrophy and resultant kyphoscoliosis, which was surgically corrected.

Case report

A 12-year-old boy attended the spinal outpatient clinic with back pain and progressive kyphotic deformity. He was diagnosed with the PS at the age of 5 and a half years. There was no family history of the condition. His visual analog scale pain score was 6 of 10. He stood with a flexed left knee and hip and a plantigrade foot. This posture compensated for his left leg hemihypertrophy and balanced his pelvis. His shoulders were level. He had a right rib prominence and thoracic kyphosis, which were not correctable. He had mild waist asymmetry, trunk shifting, and listing to the right. He had a flexion contracture of the left hip and knee (Fig. 1). There was no neurologic deficit, and his abdominal reflexes were symmetrical. There was no clinical evidence of a deep vein thrombosis in either leg.

Whole-spine radiographs demonstrated a global kyphotic deformity of 63° (T4–L3) and a long C-shaped scoliosis measuring 25° (T2–L3). A computed tomography scan of the spine revealed severe dystrophic changes: apart from C1–C4, all the vertebral bodies showed marked hyperplasia of the posterior elements. Incomplete segmentation of the vertebral bodies and posterior arches was particularly marked around the cervicothoracic junction. There was also severe canal stenosis in the thoracic and lumbar region owing to the bony overgrowth of the posterior elements, especially in T11–L1 and L4–S1 levels. The computed tomography scan also showed long narrow thorax (Figs. 2–4). A magnetic resonance image of the spine revealed syringomyelia at C6–C7 level (Figs. 5 and 6). He was followed up on a regular basis with a radiograph of his whole spine.

The surgical correction was performed 2 years after the initial presentation (at the age of 14 years) when the kyphotic deformity had progressed to 87° (Fig. 7) and the scoliosis to 36°. There was now a clinical imbalance of shoulders and pelvis, his visual analog scale pain score had increased to 7 to 9 of 10, and he was complaining of right L5 radiculopathy. On examination, there was no objective deterioration in his neurologic function compared with the initial presentation 2 years previously. The preoperative lumbar lordosis was 23°, and the spinopelvic parameters were sacral slope, 27°; pelvic incidence, 56°; pelvic tilt, 29°; and sagittal vertical axis, 140 mm. He was now Risser Grade 3 to 4. His weight was 61.6 kg and he was 172.5 cm in height, equating to a body mass index of 20.5.

Procedure

A posterior correction of the deformity was performed under general anesthesia with dual-modality spinal cord monitoring. Intermittent calf pneumatic compression devices were used for perioperative thromboembolic prophylaxis. The patient was positioned prone on Montreal mattress ensuring that the abdomen was hanging free. A subperiosteal exposure of the spine was performed up to the tips of the transverse processes bilaterally. After meticulous hemostasis, five Ponte osteotomies were carried out from T8–L1 [5], and at L4–L5, a discectomy and decompression was performed. The posterior spinal architecture was significantly hypertrophic, but the bone was soft. Bilateral segmental pedicle screws were placed (T4–L5) using a freehand technique [6,7]. A pedicle subtraction osteotomy (PSO) was performed [5] at L2. Temporary rods prevented sudden sagittal translation during the PSO. Definitive contoured rods were first secured to the lumbar region and then to the thoracic spine to close down the osteotomies, correcting and stabilizing the deformity. Morcellized local bone mixed with demineralized bone matrix was used as bone graft after decortication. The total blood loss was 1,170 mL. There was no perioperative bleeding or thromboembolic complications. Chemical thromboembolic prophylaxis was started postoperatively on the day of surgery, with a regime of 20-mg enoxaparin twice daily. Anti–factor Xa assays were performed 4 hours after the third dose, and subsequently, the dose of enoxaparin was increased to 30 mg twice daily, which resulted in factor Xa levels falling within the therapeutic range. He continued on this regime for 3 months after surgery. He was discharged after 18 days, when he was able to move from bed to chair, and he wore a molded thoracolumbosacral orthosis (TLSO) for 3 months.

Outcome, follow-up

The patient was seen at 3 and 12 months after surgery. Radiographs were taken at 3 months; the thoracic kyphosis had been corrected to 55°, the lumbar lordosis to 36°, the scoliosis to 13°, sacral slope to 19°, pelvic tilt to 26°, and sagittal vertical axis to 30 mm (Fig. 8). At 12 months, he was able to walk on his right leg using walking sticks. He is currently on the waiting list for a midthigh amputation of his left leg due to the intractable flexion deformity of his left knee. The surgery resulted in a significant improvement in his back pain, which at 12 months was 1 to 2 of 10, and there remained no neurologic deterioration after surgery (Fig. 9). The whole-spine radiographs, at 12 months, showed a satisfactory correction of the deformity, with maintenance of the correction over time.