Original Article

Coexistence of multiple rare spinal abnormalities in type 1 neurofibromatosis: a case report and literature review

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Abstracts: Orthopaedic involvement is the most common clinical presentation of Neurofibromatosis type 1 (NF-1) patients with the spinal abnormalities more frequently affected. In the spinal deformities of NF-1 patients, despite the scoliosis is the most frequent finding, several distinctive radiographic features, such as dural ectasia, defective pedicles, and spondylolisthesis, are relatively less common. Here, we reported a 16-year-old boy diagnosed with NF-1 who presented with dural ectasia, defective pedicles, and spondylolisthesis concomitantly, described the surgical treatment and provided a literature review. The boy complained of low back and leg pain for two months. On clinical examination, the patient showed multiple café au lait spots on his back and no neurological deficit. He had a family history of neurofibromatosis as his father suffering from NF-1. Imaging results demonstrated mild scoliosis, posterior scalloping of the lumber spine, L5 spondylolisthesis on plain radiographs, and marked dural ectasia of L3-L5 on MRI. Furthermore, the CT scan showed presence of thin pedicles at L3, bilateral symmetrical pedicle clefts at L4, and pars interarticularis fractures at L5. The patient received a long level posterior fusion from L1 to S1 with pedicle screws. Iliac crest autogenous graft mixed with artificial bone were used to achieve solid arthrodesis. At nine-month follow-up, the patient was asymptomatic and able to live a normal life. Our observation demonstrated that familiarity with those distinctive features in NF-1 patients could be contributed to making an early diagnosis and optimizing treatment.

Keywords: Neurofibromatosis, dural ectasia, spondylolisthesis

Introduction

Neurofibromatosis type 1 (NF-1), or Von Recklinghausen disease, is one of the most common autosomal dominant disorders with a prevalence of 1 in 3000 [11]. This disorder is caused by the defects of the NF-1 gene on the long arm of chromosome 17 whose product is neurofibromin. Approximately 50% of NF-1 cases are attributable to new mutations [15, 23].

This single-gene disorder may yet multiple and varied clinical manifestations. The clinical spectrum can range from mild to severe deformities. In 1987, The Consensus Development Conference of the National Institutes of Health has established diagnostic criteria for NF-1. The diagnosis can be made if two or more of the described criteria are present [1].

Orthopaedic involvement is the most common clinical presentation of NF-1 patients with the spinal abnormalities more frequently affected. The incidence of spinal deformities in NF-1 patients varies from 2% to 64% [1, 3, 19]. These deformities are traditionally classified into non-dystrophic and dystrophic types based on the radiographic evaluation [8]. The non-dystrophic type has the presentations similar to the idiopathic scoliosis, however, the dystrophic type has its distinctive features, including vertebral scalloping, rib penciling, transverse process spindling, vertebral wedging, paravertebral soft-tissue mass, short curve with severe apical rotation, intervertebral foraminal enlargement,
Despite scoliosis is the most frequent skeletal manifestation in affected NF-1 patients with the incidence between 10% and 71% [4, 14], these distinctive radiographic features, such as dural ectasia, spondylolisthesis, and defective pedicles, are relatively less common in NF-1 patients. Familiarity with these deformities is, therefore, contributed to making an early diagnosis and optimizing treatment [22]. Here, we reported a 16-year-old boy diagnosed with NF-1 who presented with dural ectasia, defective pedicles, and spondylolisthesis concomitantly.

Case report

A 16-year-old boy was admitted to our center with low back and leg pain for two months. The patient was treated with medications in the local hospital, but his symptom did not be relieved. On examination, the patient showed multiple café au lait spots on his back and no neurological deficit. He had a family history of neurofibromatosis, and his father had NF-1.

Plain radiographs and magnetic resonance imaging (MRI) were obtained. The anterior-posterior and lateral radiographs showed mild scoliosis, posterior scalloping of the lumber spine, and L5 spondylolisthesis (Figure 1). MRI revealed marked dural ectasia with vertebral scalloping of L3-L5 (Figure 2). Axial image showed a lateral meningocele extending through the L3-L4 foramen (Figure 3).

Initially, the patient was treated conservatively with a Boston brace. One month later, he was referred to our hospital again complaining of increasingly severe back and leg pain. To prevent the progressive deterioration of this symptom, a surgical stabilization was advised. Preoperative CT scan was performed to understand the anatomy of pedicle. The CT scan demonstrated the presence of thin pedicles at L3 (Figure 4), bilateral symmetrical pedicle clefts at L4 (Figure 5), and pars interarticularis...
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Fractures at L5 (Figure 6). Sagittal CT image showed L3-L5 vertebral scalloping with spondylolisthesis of L5 on S1 (Figure 7).

Under general anesthesia, a long level posterior fusion from L1 to S1 using pedicle screws was performed. During the procedure, the pedicle screws were not possible to set at the left side of L3, L4 pedicle and the two sides of L5 pedicles. Iliac crest autogenous graft mixed with artificial bone was used to enhance a solid arthrodesis (Figure 8).
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James [20] reported a case of a child who had neurofibromatosis, in whom the dural ectasia produced erosion of the middle column and induced a spontaneous, complete dislocation in the upper part of the thoracic spine development without neurological compromise. The patient received an internal fixation and fusion surgery to stabilize the spine. In our case, although the neurological function was intact, the symptom could not be resolved by conservative measures, thus a posterior fixation and fusion procedure was performed.

Pedicle deficiencies

Pedicle irregularities are quite rare in NF-1. A spectrum of pedicle deficiencies has been illustrated in the literature. Mandell [16] reported three cases of neurofibromatosis, one showed agenesis of a pedicle, another had multiple hypoplastic pedicles, and a third presented bilateral hypoplastic pedicles producing a spondylolisthesis. The author explained these pedicular modifications by congenital abnormalities, probably due to mesodermal dysplasia and suggested that hypoplasia or agenesis of a pedicle should be deemed as the possibility of underlying neurofibromatosis. Eichhorn [10] reported a case of neurofibromatosis treated with internal fixation, in whom fractures of lumbosacral spine.

Discussion

NF-1 patients may present with varied orthopaedic manifestations with the spine being most commonly affected. In the literature, various spinal deformities have been depicted in NF-1 patients. To our best knowledge, coexistence of dural ectasia, pedicle deficiencies and spondylolisthesis in one case has never been specially reported.

Dural ectasia

Dural ectasia is defined as a circumferential dilatation of the dural sac which most frequently occurs in the lumbosacral spine [2]. Dural ectasia is most commonly associated with Marfan syndrome but is also reported in patients with scoliosis, neurofibromatosis, ankylosing spondylitis, and Ehlers-Danlos syndrome, as well as following trauma [21]. The prevalence of dural ectasia in patients with Marfan syndrome is approximately 90%, conversely, that in NF-1 patients is about 10% [9]. The mechanism is not well understood. Collagen weakness or fibroblast dysfunction was ever thought to be the possible cause. However, Heard et al [12] argued that it was the result of increased dural pulsation.

After surgery, the boy wore the Boston brace for three months. At nine-month follow-up, the patient was asymptomatic and able to live a normal life.

Figure 8. Postoperative anterior-posterior (Left) and lateral radiographs (Right) show a long posterior fusion using the pedicle screw from L1 to S1 levels (pedicle screw on the left side of L3, L4 and two sides of L5 was not possible to set).
bar pedicles were founded from L1 to L4 with dural ectasia penetrating the body of L2. Bensaid [5] presented two cases of bilateral, symmetrical pedicular clefts associated with dural ectasia in neurofibromatosis. In one case the pedicular cleft was at the T12 level, while in the other it was at L4, and was responsible for spondylolisthesis. He established two hypotheses to explain this unusual change and suggested the second hypothesis to be the most probable. One was that the pedicular defect was a congenital abnormality; the other was that bilateral stress fractures related to thinning of the pedicle was caused by dural ectasia. In the described case, the presence of thin pedicles, bilateral symmetrical pedicle cleft, and pars interarticularis fractures suggested a role of mechanical effect. Thus, we also advocated the second hypothesis.

Spondylolisthesis

Spondylolisthesis is also a very rare finding in NF-1 patients. It is usually secondary to increased anteroposterior diameter of the spinal canal, with elongation and thinning of the pedicles. To our knowledge, only 13 cases have been reported so far and 4 cases underwent surgical treatment [6, 13, 17, 18, 21, 24, 25]. Winter and Edwards [24] reported the surgical treatment strategy of spondylolisthesis associated with dural ectasia in a 5-year-old boy with neurofibromatosis. The patient had spondylolisthesis of L5 on S1 of 65%. They first used halo-femoral traction to reduce the spondylolisthesis, and then carried out a posterolateral fusion from L3 to S1 with Harrington rod stabilization. Wong-Chung and Gillespie treated an 11-year-old girl with lumbosacral spondylolisthesis of Grade 5 surgically by in situ posterolateral fusion and a pantaloon body cast [25]. Toyoda et al [21] treated a case of grade 4 spondylolisthesis in a 15-year-old girl by posterior long level fusion with the Galveston method, and posterior lumbar interbody fusion in L5/S1 and posterolateral fusion in T10-S1. In 2009, Modi [18] et al performed a short segment fusion with pedicle screws to treat a 15-year-old boy with neurofibromatosis, who showed grade 4 spondylolisthesis concomitant with dural ectasia. Four months after surgery, the patient had S1 screw broken and had to receive a second surgery. It was inferred that short segment fixation and complete reduction were the two possible reasons for the implant failure.

Our patient was the fifth reported case of spondylolisthesis with neurofibromatosis and dural ectasia to receive surgical stabilization. As the primary goal is to stabilize the spine and halt further progression, we chose a posterior long level fusion and did not attempted to obtain a complete reduction. At nine-month follow-up, the result was satisfied. However, a long-term follow-up should be required, as even if the spine was stabilized, the dural ectasia may progress [7].

Conclusion

In summary, dural ectasia, spondylolisthesis, and defective pedicles are distinctive features in NF-1 patients. Although dural ectasia concomitant with defective pedicles or spondylolisthesis in NF-1 patients have been depicted in the literature, concurrence of these deformities in one case has not been specially described. Familiarity with these deformities is contributed to making an early diagnosis and optimizing treatment.

Disclosure of conflict of interest

None.

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