Cervical Stenosis in a Patient with Arthrogryposis: Case Report

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Abstract

Study Design Case report.

Objective Amyoplasia-type arthrogryposis is a rare congenital disease that presents as multiple contractures involving various areas of the body. To the authors' knowledge, there have been no reports of adults with amyoplasia in the current literature. We report a case of an adult patient with cervical stenosis in the setting of amyoplasia.

Patients and Methods A 48-year-old patient with amyoplasia and over 30 previous orthopedic reconstructive surgeries presented with neck pain radiating down his left shoulder and into the fingers, dysesthesia in his fingertips, and left-sided periauricular headache. A diagnosis of central spinal canal stenosis and bilateral foraminal stenosis at C3–C7 with radiculopathy was made based on computed tomography scans. Because of a prior right-side sternocleidomastoid muscle transfer, a left-side C3–C4, C5–C7 anterior cervical discectomy and fusion procedure was performed.

Results The patient experienced significant improvement in symptoms that was transient. Symptoms returned to preoperative values after 1 year, despite significant and persistent improvement in stenosis.

Conclusions Both amyoplasia and cervical stenosis can manifest in neurologic symptoms. Distinguishing the causing pathology can be challenging. The radiographic improvement of cervical stenosis in a patient with amyoplasia is not always associated with long-standing pain relief.

Keywords
- arthrogryposis
- cervical stenosis

Introduction

Arthrogryposis is a congenital disease that presents as a group of conditions with the clinical presentation of multiple contractures potentially involving various areas of the body.[1,2] Amyoplasia, the classic form of arthrogryposis, occurs sporadically, whereas distal arthrogryposis is often hereditary and has been linked to genetic mutations.[3–5] Amyoplasia is characterized by symmetrical limb contracture. Patients typically present with internally rotated shoulders, extended elbows, pronated forearms, and deformities of the wrists and fingers. In terms of the lower extremity, patients may present with externally rotated hips, flexed or extended knees, and complex clubfoot.[6–9] Amyoplasia has been reported to be associated with spinal deformity in 30 to 67% of cases.[10] It has been shown that almost one-third of cases with amyoplasia have a C-shaped thoracolumbar curve.[6,11] Congenital cervical spine deformities associated with asymmetrical amyoplasia have recently been reported in children.[12]

Amyoplasia has been associated with neurologic symptoms such as sensory ataxia.[13,14] The pathogenesis of amyoplasia is neurologic or neuromuscular in the majority of cases, with alterations in anterior horn cells, roots, peripheral nerves, and myopathy being reported.[15,16] Scoliosis, which

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Table 1 Distribution of neurologic deficits (Medical Research Council grading system)

	Right	Left
Grip	1/5	1/5
Wrist flexion and extension	0/5	0/5
Triceps	0/5	2/5
Biceps	2/5	0/5
Deltoids	1/5	2/5

can be associated with amyoplasia, can cause radiculopathy in the lower extremities.[17,18] We present a unique case of a 48-year-old male patient with the clinical presentation of neuropathy with cervical stenosis in the setting of congenital amyoplasia-type arthrogryposis.

Case Report

A 48-year-old, left-handed male patient with amyoplasia presented with a 1.5-year history of constant neck and interscapular pain radiating into his shoulders, going down to his left arm and into the hand and fingers. The pain was accompanied by dysesthesia and loss of dexterity with the same distribution. At initial office consultation, the patient presented on a wheelchair with obvious upper and lower extremity deformities. Physical examination revealed a wide neck and limited range of motion of the right shoulder due to prior fusion surgery. The patient was able to wiggle fingers and had intact sensation to light touch. The distribution of his neurologic deficits is presented in ►**Table 1**.

The patient's surgical history was positive for more than 30 orthopedic reconstructive surgeries involving the hips, knees, ankles, elbows, wrists, fingers, and shoulders, the last which was performed over 20 years prior. Furthermore, the patient received a prior right-side sternocleidomastoid muscle transfer procedure. He was obese and had high blood pressure, pyelonephritis, osteoarthritis, and sleep apnea.

Imaging indicated multilevel spondylosis without deformity or instability, and severe central and foraminal stenosis with ossification of the posterior longitudinal ligament at C3–C4, C5–C6, and C6–C7 levels (►**Figs. 1–3**). Preoperative work-up was conducted and was significant for hypertension and nonspecific T-wave abnormalities on electrocardiography. There were no contraindications for surgery, and laboratory values were within an acceptable range except for hyperglycemia and hyperlipidemia that were treated with medication.

For the surgical procedure, an anterior cervical discectomy and fusion (ACDF) was opted to treat the intervertebral foraminal stenosis. With the patient in supine position, general endotracheal intubation and anesthesia lines were placed without difficulty. A left-sided surgical approach was elected due to the previous right-side sternocleidomastoid muscle transfer. Three-level ACDF was performed at the C3–C4 and C5–C7 levels, augmented by iliac crest bone autograft and stabilized with plates (►**Figs. 4** and **5**). There were no intraoperative or immediate postoperative complications. The patient wore a hard collar for the first 2 postoperative weeks and was instructed to apply ice packs as needed. Radiographically, there was successful fusion and significantly improved central and foraminal stenosis at 10 months (►**Fig. 6**).

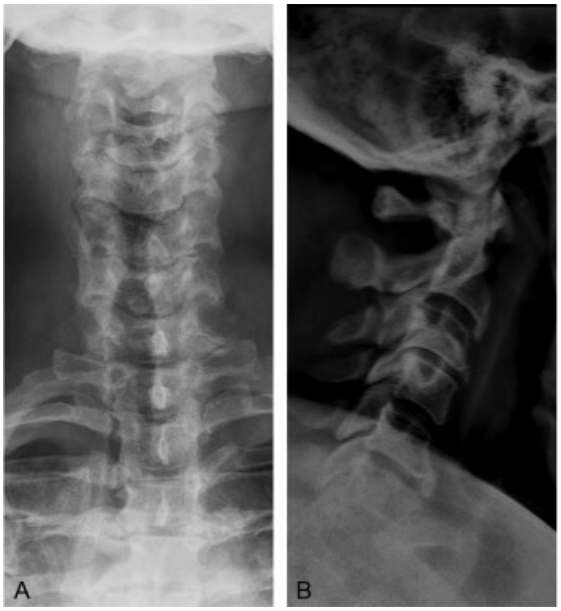

Fig. 1 (A) Anteroposterior and (B) lateral X-ray images taken 2-month pre-op, revealing severe central and foraminal stenosis at C3–C4, C5–C6, and C6–C7 levels.

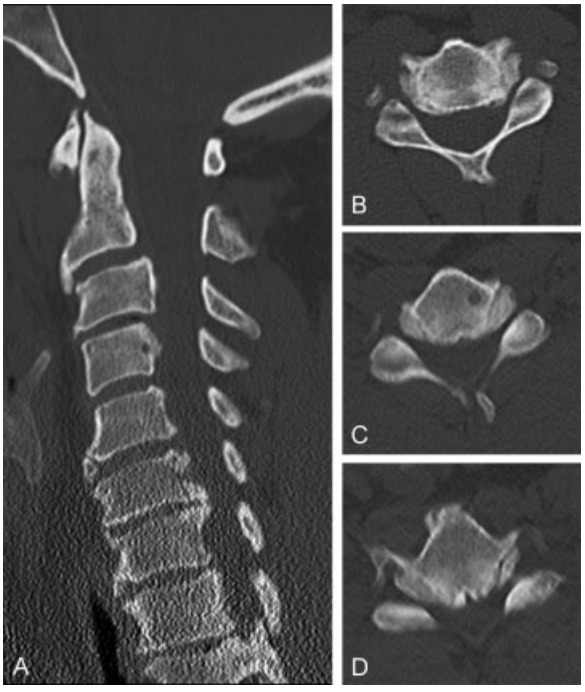

Fig. 2 Sagittal and axial computed tomography images taken 3 months pre-op. (A) Sagittal image reveals narrowing of the central foraminal space. (B) Axial image at C3–C4 reveals narrowing of the left intervertebral foraminal space. (C) Axial image at C5–C6 reveals narrowing of the left intervertebral foraminal space. (D) Axial image at C6–C7 reveals bilateral narrowing of the intervertebral foraminal space.

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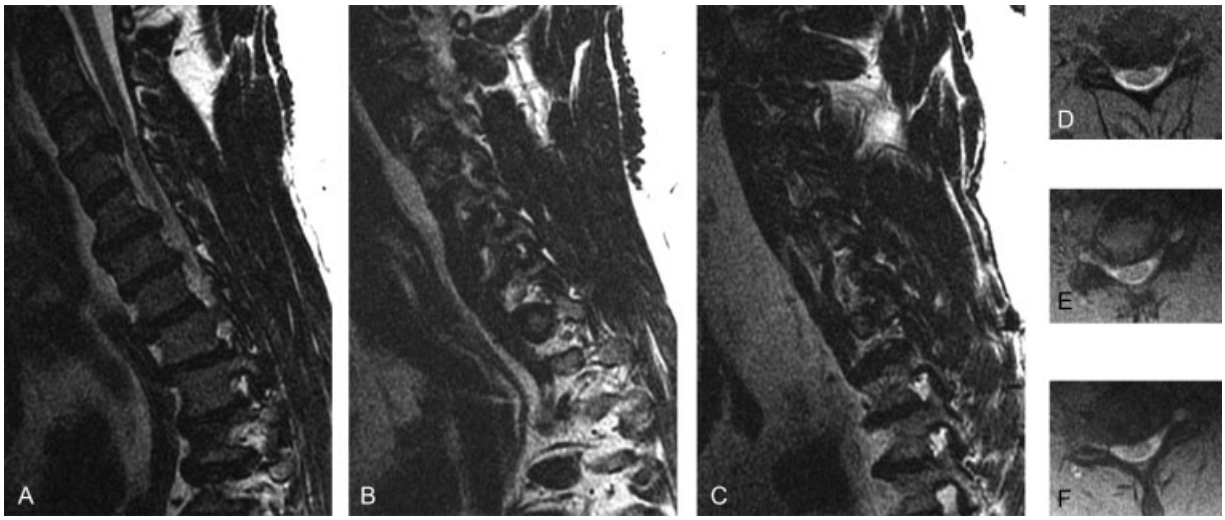

Fig. 3 Sagittal and axial T2-weighted magnetic resonance images taken 2 months pre-op. (A) Sagittal image reveals central foraminal stenosis at C3–C4, C5–C6, and C6–C7. (B) Sagittal image reveals right-side intervertebral foraminal stenosis. (C) Sagittal image reveals left-side intervertebral foraminal stenosis. (D) Axial image at C3–C4 reveals compression of nerve left nerve root. (E) Axial image at C5–C6 reveals compression of the left nerve root. (F) Axial image at C6–C7 reveals bilateral compression of nerve roots.

At 1-month follow-up, the patient reported almost complete improvement of his preoperative complaint of upper-left extremity pain. At 3-month follow-up, he reported difficulty in tapering off medication, and was referred to pain management. At 6-month follow-up, he began to experience recurrent pain and dysesthesia, with a reported 50% improvement over preoperative symptoms. Physical therapy was prescribed with focus on strengthening upper extremities and also increasing range of motion of shoulders. After completing two rounds of physical therapy, he reported worsening of pain and dysesthesia despite significant improvement of central and foraminal stenosis on plain films. At 1-year follow-up, the patient received two epidural steroid injections to the C6–C7 level. His symptoms of pain and dysesthesia were reported to be back to preoperative levels.

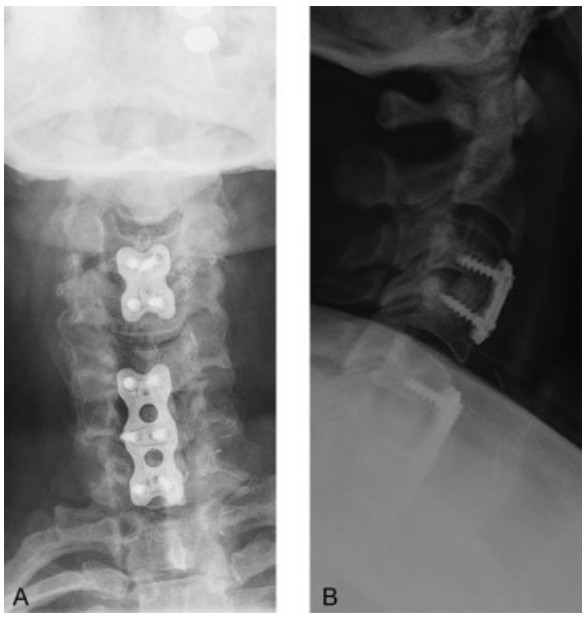

Fig. 4 (A) Anteroposterior and (B) lateral X-ray images taken 6 months post-op, revealing anterior cervical discectomy and fusion with anterior plating at C3–C4, C5–C6, and C6–C7 levels.

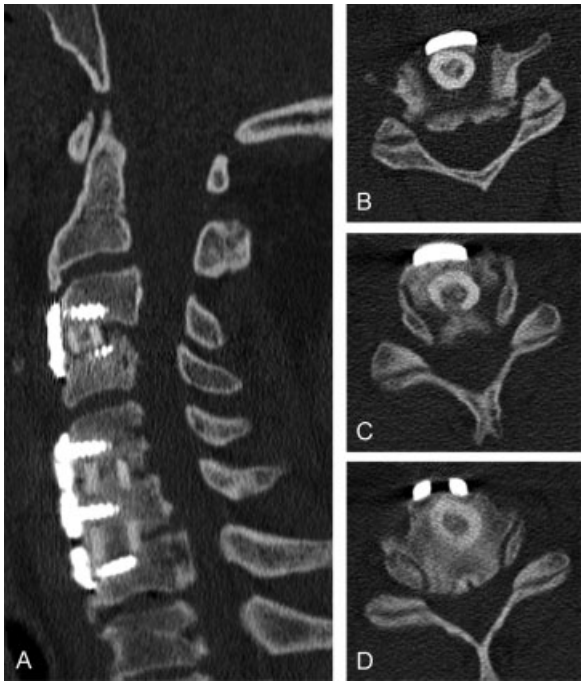

Fig. 5 Sagittal and axial computed tomography images taken 6 months post-op. (A) Sagittal image reveals anterior cervical discectomy and fusion with anterior plating. Successful fusion of intervertebral spaces is evident, with improvement in central foraminal stenosis at C3–C4, C5–C6, and C6–C7. (B) Axial image at C3–C4 reveals bilateral improvement in intervertebral foraminal space. (C) Axial image at C5–C6 reveals bilateral improvement in intervertebral foraminal space. (D) Axial image at C6–C7 reveals bilateral improvement in intervertebral foraminal space.

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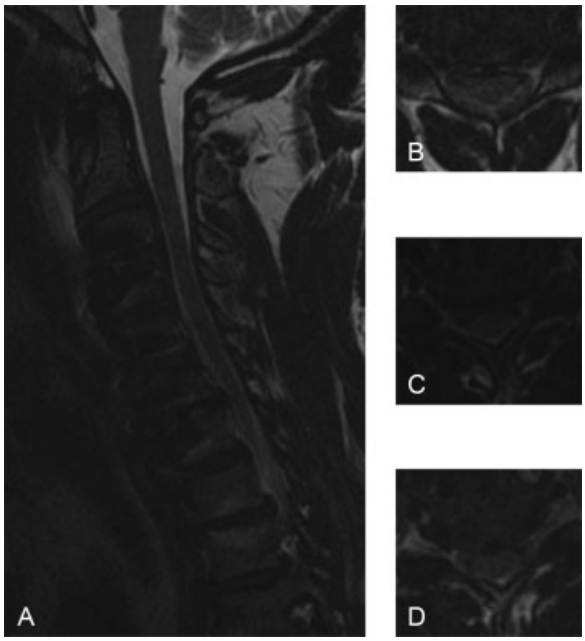

Fig. 6 Sagittal and Axial T2-weighted magnetic resonance images taken 10 months post-op. (A) Sagittal image reveals improvement of central foraminal stenosis at C3–C4, C5–C6, and C6–C7. (B) Axial image at C3–C4 reveals residual stenosis of the intervertebral foraminal space. (C) Axial image at C5–C6 reveals bilateral improvement of intervertebral stenosis. (D) Axial image at C6–C7 reveals significant bilateral improvement of intervertebral stenosis.

Discussion

Congenital arthrogryposis is a rare condition affecting less than 0.03% of live births.[1] The disease presents as a group of conditions involving multiple contractures in various areas of the body.[1,2] Generally, multiple surgical procedures are performed on patients at an early age to correct these deformities.[19]

Spinal deformity in the form of scoliosis is frequently reported in cases of amyoplasia, with reported incidence ranging from 30 to 67%.[10] The severity of the condition depends on the degree of muscle weakness.[11] Scoliosis associated with amyoplasia has been reported to cause thoracic insufficiency syndrome from reduction in hemithoracic height as well as intrinsic lung diseases, such as bronchiectasis.[20] Surgical treatment for scoliosis associated with amyoplasia involving growing rods has been reported in young children.[21] Congenital cervical scoliosis associated with asymmetrical amyoplasia has recently been reported.[12] However, congenital spinal stenosis has not been associated with amyoplasia.

Scoliosis has been associated with radiculopathy, and amyoplasia has been associated with neurologic symptoms.[13,17,18] The combinations of neuropathic and myopathic factors in amyoplasia remain poorly defined.[22] It can therefore be difficult to identify the underlying cause of radiculopathy in a patient with amyoplasia due to the diffuse nature of the disease. In our patient, multilevel cervical stenosis was treated to address the pain and dysesthesia of the patient. Treatment with ACDF initially significantly improved the patient's symptoms through treatment of central and intervertebral stenosis. However, the patient's symptoms returned within a year, despite physical therapy and epidural steroid injections. It appears that neuropathy due to amyoplasia was the underlying cause of the patient's neurologic symptoms with minimal contribution from stenosis. Thorough nerve conduction studies may have helped determine the specific affected nerves and whether neurological symptoms were due to radiculopathy from central or foraminal stenosis or more distal neuropathy due to amyoplasia.

There have been very few reports of arthrogryposis in patients past skeletal maturity.[23,24] Of these reports, patients were followed until young adulthood. Another case on two older patients with distal-type arthrogryposis-related ptosis and ophthalmoplegia has been reported.[25] To our knowledge, the patient in this report is the first adult patient with the amyoplasia-type of arthrogryposis to present with neurologic symptoms in a setting of cervical stenosis. Whether cervical stenosis is associated with amyoplasia is still unclear. The patient in this case received 30 prior orthopedic surgeries to correct for deformities. A previous right-side sternocleidomastoid muscle transfer procedure and his prior body habitus limited the surgeon to a left-side approach with access surgery performed by a head and neck specialist.

In conclusion, patients with amyoplasia can be challenging cases for surgeons to treat. Surgical considerations should be made following a thorough review of prior surgical history to treat patients with amyoplasia. Radiographic improvement of cervical stenosis is not always associated with long-standing pain relief.

References

1 Kalampokas E, Kalampokas T, Sofoudis C, Deligeoroglou E, Botsis D. Diagnosing arthrogryposis multiplex congenita: a review. ISRN Obstet Gynecol 2012;2012:264918
2 Rink BD. Arthrogryposis: a review and approach to prenatal diagnosis. Obstet Gynecol Surv 2011;66(6):369–377
3 McMillin MJ, Below JE, Shively KM, et al; University of Washington Center for Mendelian Genomics. Mutations in ECEL1 cause distal arthrogryposis type 5D. Am J Hum Genet 2013;92(1):150–156
4 Kimber E, Tajsharghi H, Kroksmark AK, Oldfors A, Tulinius M. Distal arthrogryposis: clinical and genetic findings. Acta Paediatr 2012; 101(8):877–887
5 Hageman G, Willemse J. Arthrogryposis multiplex congenita. Review with comment. Neuropediatrics 1983;14(1):6–11
6 Bevan WP, Hall JG, Bamshad M, Staheli LT, Jaffe KM, Song K. Arthrogryposis multiplex congenita (amyoplasia): an orthopaedic perspective. J Pediatr Orthop 2007;27(5):594–600
7 Stilli S, Antonioli D, Lampasi M, Donzelli O. Management of hip contractures and dislocations in arthrogryposis. Musculoskelet Surg 2012;96(1):17–21
8 Zlotolow DA, Kozin SH. Posterior elbow release and humeral osteotomy for patients with arthrogryposis. J Hand Surg Am 2012;37(5):1078–1082
9 Bamshad M, Jorde LB, Carey JC. A revised and extended classification of the distal arthrogryposes. Am J Med Genet 1996;65(4):277–281
10 Greggi T, Martikos K, Pipitone E, et al. Surgical treatment of scoliosis in a rare disease: arthrogryposis. Scoliosis 2010;5:24
11 Goldberg M. The Dysmorphic Child: An Orthopedic Perspective. New York, NY: Raven Press; 1987
12 Fletcher ND, Rathjen KE, Bush P, Ezaki M. Asymmetrical arthrogryposis of the upper extremity associated with congenital spine anomalies. J Pediatr Orthop 2010;30(8):936–941

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13 Aroojis AJ, King MM, Donohoe M, Riddle EC, Kumar SJ. Congenital vertical talus in arthrogryposis and other contractural syndromes. Clin Orthop Relat Res 2005;(434):26–32
14 Shibasaki H, Hitomi T, Mezaki T, et al. A new form of congenital proprioceptive sensory neuropathy associated with arthrogryposis multiplex. J Neurol 2004;251(11):1340–1344
15 Vuopala K, Leisti J, Herva R. Lethal arthrogryposis in Finland—a clinico-pathological study of 83 cases during thirteen years. Neuropediatrics 1994;25(6):308–315
16 Banker BQ. Arthrogryposis multiplex congenita: spectrum of pathologic changes. Hum Pathol 1986;17(7):656–672
17 Hamilton DK, Smith JS, Sansur CA, et al; Scoliosis Research Society Morbidity and Mortality Committee. Rates of new neurological deficit associated with spine surgery based on 108,419 procedures: a report of the scoliosis research society morbidity and mortality committee. Spine (Phila Pa 1976) 2011;36(15):1218–1228
18 Ploumis A, Transfeldt EE, Gilbert TJ, Mehbod AA, Pinto MR, Denis F. Radiculopathy in degenerative lumbar scoliosis: correlation of stenosis with relief from selective nerve root steroid injections. Pain Med 2011;12(1):45–50
19 Obeidat MM, Audat Z, Khriesat W. Short-term functional outcome in children with arthrogryposis multiplex congenita after multiple surgeries at an early age. J Multidiscip Healthc 2012;5:195–200
20 Campbell RM Jr. Spine deformities in rare congenital syndromes: clinical issues. Spine (Phila Pa 1976) 2009;34(17): 1815–1827
21 Greggi T, Lolli F, Maredi E, et al. Surgical treatment for scoliosis associated with rare disease. Stud Health Technol Inform 2012; 176:326–329
22 Herring JA. Orthopaedic Syndromes. Philadelphia, PA: Saunders Elsevier Press; 2008
23 Yau PW, Chow W, Li YH, Leong JC. Twenty-year follow-up of hip problems in arthrogryposis multiplex congenita. J Pediatr Orthop 2002;22(3):359–363
24 Fassier A, Wicart P, Dubousset J, Seringe R. Arthrogryposis multiplex congenita. Long-term follow-up from birth until skeletal maturity. J Child Orthop 2009;3(5):383–390
25 Friedman BD, Heidenreich RA. Distal arthrogryposis type IIB: further clinical delineation and 54-year follow-up of an index case. Am J Med Genet 1995;58(2):125–127

Commentary

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Patients with multiple diagnoses that can contribute to clinical symptomatology often pose diagnostic and therapeutic challenges to the surgeon. For example, patients with multiple sclerosis and long-tract signs may be symptomatic from the underlying disease or from compressive cervical stenosis.[1,2] The decision to offer surgery in this instance may be difficult, as there is a chance that intervention may not be helpful and could possibly cause harm.

The authors of this case report faced a similar clinical conundrum. A 48-year-old male patient with arthrogryposis and 30 previous surgeries presented with what appeared to be symptoms of cervical radiculopathy but could also have been consistent with the underlying disease. A magnetic resonance image revealed findings consistent with nerve root compression, and the patient underwent a three-level anterior cervical diskectomy and fusion. His symptoms resolved for a short period of time, only to return a year after surgery.

It may seem logical, in hindsight, to criticize the authors for offering surgery in a patient who ultimately had no improvement. There is no way to know this when the patient presents, particularly when confronted with a very rare underlying disease, as was the case here. The surgeons made a judgment call, and the patient did experience symptomatic relief for a period of time. In the end, surgeons have their experience and their technical expertise to offer patients; if there is any chance that the patient's course will improve with intervention, the most prudent course to take may be the most invasive.

Although surgery appears to be an appropriate course of action in this case, the actual procedure performed may be debatable. The authors performed a nonconsecutive three-level diskectomy, fusing C3–C4 and C5–C7, leaving the C4–C5 level untouched. Because this level was not fused, it is very likely there will be degenerative changes during the patient's lifetime.[3] Perhaps a better choice would have been to fuse this level as well, or perform a posterior procedure, which could have achieved the same surgical goals. At the very least, the patient will require close follow-up to evaluate the uninstrumented disk space.

Nevertheless, the lesson offered in this case should be reemphasized. In patients harboring more than one disease that could explain their symptoms, surgery should be considered and frequently offered, as patients will often benefit.

References

1 Arnold PM, Warren RK, Anderson KK, Vaccaro AR. Surgical treatment of patients with cervical myeloradiculopathy and coexistent multiple sclerosis: report of 15 patients with long-term follow-up. J Spinal Disord Tech 2011;24(3):177–182
2 Ronthal M. On the coincidence of cervical spondylosis and multiple sclerosis. Clin Neurol Neurosurg 2006;108(3):275–277
3 Helgeson MD, Bevevino AJ, Hilibrand AS. Update on the evidence for adjacent segment degeneration and disease. Spine J 2013; 13(3):342–351

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Editorial Perspective

The authors deserve our recognition for being willing to present a complex case without positive outcome for our review and collective learning. Case reports are rightly and frequently looked down upon for their lack of methodological and substantive foundations. That said, rare conditions or constellations of disorders elude more formal investigations, and as this case illustrates very well, every patient, every case, is different, and we can learn from them in many ways. What can we deduct from this case?

Commentator Dr. Arnold concurred with the general decision-making process here, assuming that full disclosure with the patient and family relative to the uncertainty of outcome was undertaken prior to the intervention. As this case shows, technical points of how to perform surgery in unusual circumstances remains a very compelling topic: Do we make compromises in application of our principles (such as leaving a single motion segment unfused between other fusion levels) or should we stick to dogma under all circumstances, even if it means a more invasive surgery? Finding a middle ground might seem appealing but harbors the question of what to do if complications arise. In this case, a more extensive decompression and fusion such as a multilevel anterior decompression and fusion may have deterred the patient and the family away from surgery; a greater perioperative morbidity would also have to be taken into consideration. We invite the comments of the EBSJ readership around the world to share their opinions on the topic of decision-making in equivocal circumstances such as presented here and to also provide any observations on outcomes of spine surgery in adults with arthrogryposis.

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