Familial cervical spondylosis

Case report

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Cervical spondylosis is a disease that is often attributed to aging and considered the result of degenerative changes in the spine. The idea that there is a genetic predisposition to develop diseases of the skeletal elements of the spine has been discussed previously, but has never been proven conclusively. The authors report three cases of severe cervical spondylosis in patients who are first-degree relatives: a mother and her two sons. All three individuals had cervical disc herniations and stenosis at C3–4, C4–5, C5–6, and C6–7, and all three required decompressive procedures. The location and degree of cervical spondylosis were as similar among these three patients as they have been in identical twins reported in other studies. Such familial inheritance of cervical spondylosis has been reported only once. The existence of familial cervical spondylosis is not an unrealistic proposal because other studies have shown that genetics determines the shape of one’s spine and that similar spines tend to degenerate in similar ways. Therefore, genetic counseling for a family such as the one reported here may prove to be of great benefit to warn siblings that they are at high risk for cervical spine injury. However rare it might be, familial cervical spondylosis may be a phenomenon that any spine surgeon should suspect in a family with cervical spine abnormalities found in several members.

KEY WORDS • cervical spondylosis • familial occurrence • genetics

Case Reports

Case 1: The Mother

This 56-year-old woman had suffered pain and weakness in the upper extremities for many years, which required long-term physical therapy, rehabilitation, and medications. She was involved in a motor vehicle accident and was reported at the scene of the accident to have numbness and weakness in the arms and hands bilaterally.

Examination. On presentation to our service, the patient exhibited signs of central cord injury including absence of motor function in the triceps, the wrist extensors and flexors, and the intrinsic muscles of the hands with decreased sensation in a C-7 to T-1 distribution. She also exhibited decreased sensations in the lower extremities with some hypertonicity, but otherwise had normal strength. Plain x-ray films and computerized tomography scans of the cervical spine showed that the patient had a stenotic cervical canal from C-3 to T-1 with no evidence of fractures, but with large osteophytes at C4–5, C5–6, and C6–7 and significant disc herniations at C5–6 and C6–7 (Fig. 1 left).

Medical Treatment. The patient was admitted to the hospital and given methylprednisolone therapy for spinal cord injury. She then underwent rehabilitation and regained much of the lost motor functions. She continued to have pain and weakness in the upper and lower extremities, and a decompressive procedure was recommended as prophylaxis against future injury.

Operation. Extended anterior cervical discectomy with interbody fusion was performed at C4–5, C5–6, and C6–7, along with microsurgical decompression of the spinal cord.

Case 2: The First Son

Approximately 1 year later, a 34-year-old man, a son of the woman described in Case 1, presented with pins and needles in his hands and Lhermitte’s sign, which consisted of electrical shocks that ran down his arms and legs when he coughed or moved his neck.

Examination. The only significant finding on physical examination was decreased sensation to pinprick in both hands. Magnetic resonance imaging of his cervical spine...
showed significant disc herniations at C4–5, C5–6, and C6–7 (Fig. 1 center). The disc herniation at C5–6 was the most pronounced; the spinal cord at that level had increased signal attenuation.


Case 3: The Second Son

One year after the first son presented, a second son, who was also 34 years old at the time of presentation, came to our attention after having fallen out of bed and striking his head. He developed similar shooting electrical shocks in the lower back and numbness in the right hand.

Examination. On physical examination, clonus in the left ankle was the only significant finding. Magnetic resonance imaging in this patient revealed cervical spondylosis with disc herniations at the C3–4 and C4–5 levels (Fig. 1 right).


Discussion

There are certain conditions of the spine that are known to show some traits of inheritance such as ankylosing spondylitis, idiopathic scoliosis, ossification of the posterior longitudinal ligament, spondylothoracic dysplasia, spondylolisthesis, and even herniated discs in certain populations. The common disease of cervical spondylitis has also been proposed by some to be genetically passed from one member of a family to another. However, cervical spondylosis is more often than not considered to consist of degenerative changes in the cervical spine that result from aging and/or repeated episodes of trauma. In contrast to that notion, cases of elderly individuals who have an absolutely pristine cervical spine with no radiological evidence of wear and tear are frequently seen. Moreover, there are many cases of young people who have no history of trauma but who do have radiological features of cervical spondylosis. On the basis of these anecdotal phenomena, one can postulate that there may well be a genetic basis for cervical spondylosis.

The familial pattern of spondylitic changes in the spine was first studied in twins. Virtually identical radiographic similarities were first reported by King. He performed a radiographic survey of 11 pairs of identical twins and found striking similarities in many bones of both twins including the cervical, thoracic, and lumbar spine. Moreover, he found either the absence or presence of degenerative changes in matched twins at identical sites in the spine at the same levels. Although King studied a small series of twins and did not perform any statistical analysis of his data, he suggested that there might be an element of genetic determination in the degeneration of the spine.

There are two reports in the literature in which the hereditary tendency of cervical spondylosis has been studied. In the first study, Bull, et al., looked at the plain cervical radiographs obtained in monozygous twins, dizygous...
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gous twins, and siblings of individuals with cervical spondylosis. These authors found that the radiological anatomy of the cervical spine was virtually identical in both monozygous and dizygous twins, as opposed to matched pairs in the general population. Furthermore, twins of either type were more likely to have the same degree of spondylosis than any pair of persons of the same age taken at random from the general population. Therefore, it appeared that in twins the cervical spine developed in an almost identical fashion and, subsequently, these persons presented with cervical spondylosis in similar locations and patterns. The idea that genetically identical or similar individuals have virtually identical bone architecture and develop bone diseases in similar fashion appeared to lend support to the genetic basis for cervical spondylosis.

In the case of siblings of patients with cervical spondylosis, Bull, et al.,1 studied 11 families and concluded that the siblings of individuals with cervical spondylosis did not have a statistically significant incidence of cervical spondylitic changes on their cervical spine films. However, there was one large family of five siblings, aged 41 to 61 years, in whom severe cervical spondylosis was consistently present in each sibling. Although cervical spondylosis is not a familial disease in the general population, there may be families such as the one reported by Bull, et al., and the one reported in the present manuscript in which cervical spondylosis is, undoubtedly, an inherited trait.

In the second study on familial pattern of cervical spondylosis, Palmer and colleagues6 analyzed lateral cervical spine radiographs obtained in 23 pairs of twins to look for matching degenerative changes. In agreement with Bull, et al.,1 Palmer and colleagues also found striking similarities in the bone anatomy of the cervical spine in matched twins and, particularly, in monozygous twins. These authors also noted that in twins degenerative changes in the cervical spine followed a similar course. Twelve of 23 pairs of twins had matching anterior degenerative changes and 19 of 23 pairs of twins had statistically significant matching posterior changes. Palmer and colleagues suggested that the particular form of the bony elements is one of the most important determinants of what degenerative changes would develop. Furthermore, the authors surmised that because the anatomy of the cervical spine is so alike in genetically similar individuals, changes in the spines of family members should naturally follow a similar degenerative pattern. This concept could explain the distribution of cervical spondylosis found in certain families such as the one reported in this manuscript. Palmer’s group, however, also concluded that because greater matching of degenerative changes should have been found if cervical spondylosis was solely genetically determined, it would be wrong to state that all spondylosis was genetic in origin.

Conclusions

The three patients presented in this paper were not genetically identical, as would be the case in monozygous twins, but were only first-degree relatives. The location and degree of cervical spondylosis, however, were as similar as the degenerative changes found in identical twins studied by investigators who analyzed the bone structures and diseases of monozygotic twins. To our knowledge, such familial inheritance of cervical spondylosis has been discussed in the medical community, but only one instance has been reported by Bull, et al.1 To state that there is such a disease as familial cervical spondylosis is a presumptive conclusion on our part. Nevertheless, such a diagnosis can unarguably be made for the family described in this report and for the family described by Bull, et al. In addition, the existence of familial cervical spondylosis is not an unrealistic proposal based on the papers referenced in this manuscript that all seem to reach the conclusion that genetics determines the shape of one’s spine and that similar spines tend to degenerate in similar ways. Most importantly, genetic counseling for a family such as the one reported on here may have proven to be of great benefit to warn siblings that they are at risk for cervical spine injury. However rare it might be, familial cervical spondylosis may be a phenomenon that a spine surgeon should suspect in a family with cervical spine abnormalities found in several members.

References


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