Clinical considerations in the chiropractic management of the patient with Marfan syndrome.

Ruling JR; Crowther ET; McCord P

Applied Health Sciences Research Unit, University of Waterloo, BMH 1602, Waterloo, Ontario N2L 3G1, Canada

Journal of Manipulative & Physiological Therapeutics (J MANIPULATIVE PHYSIOL THER), 2000 Sep; 23(7): 498-502 (40 ref)

OBJECTIVE: To describe the chiropractic management of a patient with whiplash-associated disorder and a covert, concomitant dissecting aneurysm of the thoracic aorta caused by Marfan syndrome or a related variant. Clinical Features: A 25-year-old man was referred by his family physician for chiropractic assessment and treatment of neck injuries received in a motor vehicle accident. After history, physical examination, and plain film radiographic investigation, a diagnosis of whiplash-associated disorder grade I was generated. Intervention and Outcome: The whiplash-associated disorder grade I was treated conservatively. Therapeutic management involved soft-tissue therapy to the suspensory and paraspinal musculature of the upper back and neck. Rotary, manual-style manipulative therapy of the cervical and compressive manipulative therapy of the thoracic spinal column were...
implemented to maintain range of motion and decrease pain. The patient achieved full recovery within a 3-week treatment period and was discharged from care. One week after discharge, he underwent a routine evaluation by his family physician, where an aortic murmur was identified. Diagnostic ultrasound revealed a dissecting aneurysm measuring 78 mm at the aortic root. Immediate surgical correction was initiated with a polyethylene terephthalate fiber graft. The pathologic report indicated that aortic features were consistent with an old (healed) aortic dissection. There was no evidence of acute dissection. Six month follow-up revealed that surgical repair was successful in arresting further aortic dissection. CONCLUSION: The patient had an old aortic dissection that pre-dated the chiropractic treatment (which included manipulative therapy) for the whiplash-associated disorder. Manipulative therapy, long considered an absolute contraindication for abdominal and aortic aneurysms, did not provoke the progression of the aortic dissection or other negative sequelae. The cause, histology, clinical features, and management considerations in the treatment of this patient's condition(s) are discussed.
INTRODUCTION

Marfan syndrome was first reported in 1896 by French pediatrician Antoine Marfan, who described a 5-year-old child with physical features including scoliosis and long thin extremities.[1] This unique syndrome is a pleiotropic condition that affects multiple tissues and organs.[2] Marfan syndrome is an autosomal-dominant, connective-tissue disorder that arises from a mutation of the fibrillin FBN1 gene, resulting in the generation of weak fibrillin protein fibers.[3–5] Because many tissues within the body incorporate fibrillin as a constructive substrate, various organ systems are susceptible to structural variations.[5,6] These anomalies lead to structural weaknesses and/or phenotypic anomalies of many areas of the body, such as the skeletal, cardiac, ocular, and pulmonary systems and skin and dura.[2,7] Because manipulative therapy generates considerable forces within the holding elements of articular structures, the chiropractor must be aware of the clinical presentation and physical implications when encountering a patient with this genetic condition. With knowledge of the various characteristics associated with Marfan syndrome, the chiropractor can then manage more safely and effectively patients with this genetic anomaly.

We report the case of a 25-year-old man who underwent successful chiropractic management for cervical spine injuries sustained in a motor vehicle accident (MVA). This patient was subsequently discovered to have a dissecting thoracic aneurysm (related to Marfan syndrome or a closely related variant), and he underwent successful surgical intervention.

We discuss the incidence, cause, and pathophysiologic features of this condition. The clinical presentation involving the most common manifestations of Marfan syndrome, with an emphasis on those that can help identify the patient with this syndrome, is reviewed.

CASE REPORT

A 25-year-old man had general neck and upper back stiffness after an MVA. This man was the driver of a van that collided with the rear of another vehicle. Although he denied contact with any of the interior components of the vehicle, objects within the van subsequently struck him in the head. He was wearing a standard 3-point seatbelt at the time of impact; his van was fitted with head restraints. There was a suspicion of loss of consciousness after the collision because he was unable to recall all details of the MVA.

He was taken to a local hospital where he was clinically and radiographically assessed. There was no evidence of fracture or dislocation. He was discharged with a diagnosis of soft-tissue injuries and instructed to see his physician the next day. He was examined by his family physician and subsequently referred to a chiropractor for further...
The patient was a relatively healthy man in his third decade of life. His height was 74 inches, and he weighed 226 pounds, with endomorphic body habitus. Postural assessment from the posterior and lateral aspects failed to demonstrate clinically significant findings. Palpation of the associated musculature demonstrated hypertonic and tender cervical paraspinal and suspensory muscles. Range of motion of the cervical spine was full but painful in all directions. On orthopedic examination, cervical distraction/compression tests were normal. Neurologic tests for the upper and lower limbs were unremarkable. Deep tendon reflexes were graded 2++; muscle strength testing was 5/5 throughout. There were no sensory deficits to light touch or pinprick. Plantar responses were downgoing bilaterally. Chiropractic motion palpation revealed painful intersegmental restrictions in the cervical and thoracic spine. No further radiographic evaluation was indicated.

A diagnosis of type I whiplash-associated disorder was made. A therapeutic plan of management, including soft-tissue therapy to the cervical paraspinal and suspensory musculature and spinal manipulative therapy to the cervical and thoracic spinal column for symptomatic relief, was implemented. Cervical manipulation was characterized as rotary-style, manual intervention; thoracic manipulation consisted of anterior-posterior (anterior thoracic) compressive procedures. The patient was treated 4 times and achieved full recovery within 3 weeks.

One month after initiation of chiropractic treatment, he was assessed by his family physician. A previously undetected aortic heart murmur was evident. All other aspects of the physical examination were unremarkable. The patient underwent additional investigations including radiographic chest examination (posterior-anterior and lateral), suggesting a possible dilatation at the aortic root. Diagnostic ultrasound revealed significant aortic valve regurgitation and mitral valve prolapse. In addition, a dissecting aneuryism at the aortic root measuring 78 mm was noted (Fig 1 and 2). Possible causes of the dissecting aneurysm included Marfan syndrome, atherosclerosis, familial aortic aneurysm, syphilis, and bacterial endocarditis.

He was referred to a cardiologist and underwent surgical correction of the dissecting aneurysm with repair incorporating a Dacron graft (DuPont, Wilmington, Del). The pathology report revealed features consistent with an old, chronic (healed) aortic dissection with no recognizable evidence of acute aortic dissection. Further surgical repair involving dehiscence of the proximal suture line and Dacron graft was undertaken 1 month after the initial surgery. A follow-up examination at 6 months revealed that surgical repair was successful in stopping further aortic dissection.

To identify the cause of the aortic aneurysm and confirm the diagnosis, a postsurgical analysis was performed. The patient exhibited evidence of joint hypermobility, high arched palate with crowding of teeth, and malar hypoplasia. Additional questioning revealed that there was a physical altercation with a family member 1 year before the MVA, leading to a 1-minute unexplained loss of consciousness. Several months after this loss of consciousness the patient reported an incident of “crushing” chest pain on hearing the events of the sudden death of his sister. This chest pain was assessed and diagnosed as pneumonia, and a course of antibiotics was prescribed. The chest pain resolved without further complication. It was believed that the patient had a dissection of the aorta during the altercation and/or the incident of chest pain. The family history revealed that several relations of the family had dissecting aneurysms, with 1 relative diagnosed with probable Marfan syndrome or a similar, related variant.

**DISCUSSION Incidence and Cause**

Within the general population, the incidence of Marfan syndrome is 0.01%[8] and prevalence is estimated at 1 to 10 per 10,000 people.[9, 10] This syndrome does not differentiate according to race, sex, or ethnicity.[10]

Although Marfan syndrome is an inherited autosomal dominant pattern, approximately 25% of the cases arise because of sporadic mutations.[9, 10] Greater than 80 different mutations of the FBN1 gene causing Marfan syndrome have been identified to date.[5] The FBN1 gene displays a high level of penetrance; having this gene mutation may induce the appearance of some dramatic physical features,[5] although there can be considerable variation and subtlety in presentation. It is this variation and subtlety in features that make clinical identification of the syndrome difficult, as in this case.[5] Our patient did not have the typical Marfanoid presentation but did fulfill other aspects of the diagnostic guidelines.[2]

DNA screening, a typically useful screening tool for other genetic conditions, is difficult to devise for Marfan syndrome because many families present with distinct genetic mutations.[11] As a consequence, the diagnosis may often be based solely on the clinical features involving the various organ systems[2, 7] and/or the inheritance of an identified mutated FBN1 gene known to cause Marfan syndrome.[7] Manifestations of the skeletal, cardiac, ocular, and pulmonary systems and skin and dura are also frequent and can aid in the diagnosis of Marfan syndrome.[2, 7] The revised diagnostic criteria for Marfan syndrome[7] are the current standard used to diagnose...
this condition and its related variants.

**Phenotypic Appearance**

Most patients exhibit varied musculoskeletal abnormalities, hyperextendable joints, and skin changes.[12] Musculoskeletal difficulties can arise because of ligament and joint laxity and excessive longitudinal growth of the tubular bones. The growth of the long tubular bones results in the body habitus of an extremely tall, thin individual, which is classically associated with Marfan syndrome. However, not all people with Marfan syndrome have tall, thin statures.


**Spinal Column Involvement**

Spinal column involvement is common in Marfan syndrome. There are higher incidences of cervical,[8] thoracic,[17] and lumbar[17] spinal column abnormalities compared with the general population. Hobbs et al.[8] found that clinical problems such as neck pain are rare, despite finding that 35% of subjects exhibited abnormal cervical lordosis. In addition, neurologic compromise was found to be rare in Marfan syndrome, despite these structural abnormalities; however, the risk for neurologic damage is believed to be higher.[8] Low back pain is more common than in the general population[15] and should be managed conservatively. Scoliosis is considered the most frequent skeletal manifestation of the lumbar spine, with incidences ranging from 40% to 70%.[17, 18] Scoliosis has been shown to increase the risk of pain in the area of curve development in adults with Marfan syndrome.[17]

A common and often under-recognized manifestation of the spinal column is dural ectasia, which can demonstrate signs and symptoms of nerve root compression and low back pain.[15, 20, 21] Dural ectasia commonly develops in the lumbosacral region where the expanded dural sac widens the spinal canal. This development can cause interpedicular widening, erosion of the vertebral bodies leading to scalloping, narrowing of the pedicles, and dilation of the neural foramina.[21] As pedicles are attenuated because of dural ectasia, possible fracture of the pedicles should be considered as a cause of low back pain in a patient with Marfan syndrome.[22] Because dural ectasia is sometimes a common feature of Marfan syndrome leading to low back pain and radiculopathy, it is suggested that Marfan syndrome and associated dural ectasia be considered as a differential diagnosis of low back pain in tall individuals suspected to have Marfan syndrome.[15, 20]

**Other Areas of Systemic Involvement**

Most children with Marfan syndrome demonstrate a predilection for myopia at an early age.[9] Of these, 50% will exhibit ectopia lentis (subluxation of the ocular lenses).[23] This cause of reduced visual acuity should be investigated and corrected. Because the connective tissues and skeletal system involving fibrillin are commonly weakened, many other areas of the body are structurally compromised; as such, hernias are a frequent manifestation.[24] Within the pulmonary system, cystic changes are often noted, which can lead to a pneumothorax.[24] Other physical characteristics associated with the presence of Marfan syndrome are listed in Table 1 and in their entirety by De Paepe et al.[7]

**Cardiovascular Involvement**

Although Marfan syndrome is linked with a wide variety of musculoskeletal conditions, it is most often associated with complications involving the cardiovascular system. Fibrillin is abundant in the elastic lamellae of the aortic wall,[25] As a result of the altered fibrillin fibers, the aortic walls tend to weaken over time, caused by constant diastolic and systolic cycles leading to aortic root dilatation.[1] This can progress to serious clinical consequences of valve prolapse,[26] aortic regurgitation, aortic dissection,[9, 12, 13, 23, 27--29, 31] and other related cardiovascular abnormalities.[32] The most common cause of sudden death associated with Marfan syndrome remains aortic dissection,[3] which can occur in patients exhibiting only mild aortic dilatation.[32] In addition, the occurrence of aortic dilatation in Marfan syndrome is variable and does not develop as a function of increasing age.[39] Severe aortic dilatation can be seen in patients as young as the teens to early twenties and progress rapidly to dissection or aneurysm, as in this case. At present, there are no clear clinical predictors to assist clinicians in identifying those patients who may develop life-threatening cardiovascular anomalies.[33]

**Prognosis**

Patients diagnosed with Marfan syndrome can typically enjoy a productive life, free from impairment. The median life expectancy rate for individuals with Marfan syndrome has increased 25% between 1972 and 1990, with life expectancy currently 72 years of age.[34] This increase in life span is primarily attributable to advanced cardiac diagnostic techniques and surgical interventions.[10, 13, 34]
Management

Patients sharing the same gene as relatives with known Marfan syndrome may exhibit varying Marfanoid phenotypes. One of the goals of management is to interrupt the natural history of the physical manifestations at the earliest age possible. However, effective management may be difficult because of the phenotypic variability. Consequently, prognostication is difficult and limited to generalizations.[5, 9] However, all patients should have a thorough diagnostic work-up of organ systems known to be affected by Marfan syndrome. Prophylactic management of any organs identified as affected can then be implemented.[15]

For the spinal column, management should begin during childhood. Progression of growth abnormalities such as scoliosis occurs rapidly during the early years of development,[15] with potential danger to the thoracic cavity's organs. For scoliotic curves >20 degrees detected during the growth phase, bracing is recommended.[15] In a departure from the natural history of adolescent idiopathic scoliosis, curvatures of 30 to 40 degrees have a high predilection for progression in the adult.[17] In scoliotic curves >40 degrees, instrumentation with or without spinal fusion should be considered.[9, 15]

Because aortic dilatation is usually silent, regular clinical follow-up, which should include echocardiographic investigation, is required. Prophylactic surgical repair should occur when the aortic-root diameter reaches 55 mm during adulthood. However dissection may occur before reaching the critical diameter of 55 mm.[35] Aggressive surgical approaches and the implementation of prophylactic pharmaceuticals such as β-adrenergic blockers for the management of cardiovascular problems, have been gaining support and are pivotal in altering the natural history of Marfan syndrome.[13, 34–40] The impact of the underlying conditions affects the overall health of the patient. Therefore lifestyle changes should be introduced, and ultimately family planning and counseling should be introduced to address the genetic aspects of the condition. The diagnosis of Marfan syndrome should not be taken lightly because this condition is accompanied by significant morbidity and death.

CONCLUSION

There are several important features of this case report. First, our patient was diagnosed with a whiplash injury and treated conservatively and fully recovered. It must be realized that there is a common clinical domain within individuals exhibiting Marfan syndrome and the general population. The significant difference is that the affected connective tissues lead to weaker than normal holding elements within the joints. Although chiropractors must exercise caution when implementing manipulative therapy, as in this case, most patients, including those with Marfan syndrome, may respond favorably to this form of therapeutic intervention.

Secondly, one of the most interesting aspects of the case was the implementation of manipulative therapy in a patient with a thoracic aortic aneurysm. This clinical condition has long been considered an absolute contraindication to manipulative therapy. It has been postulated that an increase in force and pressure about the weakened vascular structures may lead to progression and rupture of the aneurysm. In this case, no such phenomenon occurred. In this case, compressive manipulative forces applied to the thoracic spine failed to have a negative impact on the structural integrity of the thoracic aortic aneurysm. Indeed, the postsurgical analysis of the aortic tissue demonstrated the presence of chronic tissue, with no evidence of acute injury. We do not suggest that it is appropriate to implement manipulative therapy in patients who knowingly have an aneurysmal defect. These conditions remain a significant, urgent medical situation. Rather, we report a case where, fortunately, the expected side effects were not realized. The clinical presentation of this patient was not accompanied by the more overt physical manifestations usually associated with Marfan syndrome, which would increase suspicion for the potential contraindications for manipulation. If the heart murmur was not detected on subsequent evaluation, our patient may have had a major life-threatening event caused by progression of the aortic aneurysm.

Finally, the chiropractor must be aware that for the patient with Marfan syndrome, many organ systems may be at risk because of serious structural changes. The cardiovascular system is most often associated with morbidity and death caused by Marfan syndrome. Regular echocardiographic assessments must be undertaken to evaluate the cardiovascular system for the presence of any structural abnormalities. In the adult spine, neurologic compromise is rare; however, the risk of neurologic damage is speculated to be higher than in the general population. In addition, dural ectasia should be considered when treating the lumbar spine. When evaluating children, the chiropractor should monitor any existing scoliosis and ensure prompt referral if the scoliosis reaches a critical measure of <ge>20 degrees. Reduced visual acuity should be investigated, and if detected, a referral for further evaluation should be made.

Submit reprint requests to: Jeffrey Tuling, DC, Applied Health Sciences Research Unit, University of Waterloo, BMH 1602, Waterloo, Ontario N2L 3G1, Canada.
Table 1. Physical characteristics that may present with Marfan syndrome or related variants

<table>
<thead>
<tr>
<th>Condition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pectus carinatum/excavatum</td>
</tr>
<tr>
<td>Reduced upper to lower segment ratio or arm span to height ratio greater than 1.05</td>
</tr>
<tr>
<td>Wrist and thumb signs</td>
</tr>
<tr>
<td>Scoliosis of &gt;20 degrees or spondylolisthesis</td>
</tr>
<tr>
<td>Reduced extension at the elbows (&lt;170 degrees)</td>
</tr>
<tr>
<td>Medial displacement of the medial malleolus causing pes planus</td>
</tr>
<tr>
<td>Protrusio acetabula (ascertained on radiographs)</td>
</tr>
<tr>
<td>Joint hypermobility</td>
</tr>
<tr>
<td>High arched palate with crowding of teeth</td>
</tr>
<tr>
<td>Facial appearance of dolichocephaly, malar hypoplasia, enophthalmos, retrognathia</td>
</tr>
<tr>
<td>Down-slanting palpebral fissures</td>
</tr>
<tr>
<td>Ectopia lentis</td>
</tr>
<tr>
<td>Dissection of ascending aorta</td>
</tr>
<tr>
<td>Dilatation of ascending aorta with or without aortic regurgitation</td>
</tr>
<tr>
<td>Spontaneous pneumothorax</td>
</tr>
<tr>
<td>Striae atrophicae not associated with marked weight changes, pregnancy, or repetitive stress</td>
</tr>
<tr>
<td>Recurrent or incisional herniae</td>
</tr>
<tr>
<td>Dural ectasia (by computed tomography or magnetic resonance imaging)</td>
</tr>
<tr>
<td>Family/genetic history</td>
</tr>
</tbody>
</table>

Fig 2. Graphic representation of Fig 1 that more clearly defines the heart, vascular structures, and dissecting aneurysm. RV, Right ventricle; LV, left ventricle; AV, aortic valve; MV, mitral valve; LA, left atrium.
Fig 1. Echocardiograph of the heart and vascular structures demonstrating dissecting aortic aneurysm of 78 mm (arrows).

REFERENCES


By Jeffrey R. Tuling, DC, Clinical Science Resident, Division of Post-graduate Studies and Research, Canadian Memorial Chiropractic College, Toronto, Canada.; Edward T. Crowther, DC, Assistant Professor, Division of Clinical Education, Canadian Memorial Chiropractic College, Toronto, Canada. and Phyllis McCord, DC, MD, Associate Professor, Division of Clinical Sciences, Canadian Memorial Chiropractic College, Toronto, Canada.