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Syndromic scoliosis in a patient with arthrochalasia Ehlers-Danlos syndrome corrected with a Wood-Rigo-Cheneau derotational brace

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SUMMARY

We present a boy in middle childhood with a medical history of arthrochalasiaEhlers-Danlos syndrome who was diagnosed with scoliosis as a toddler. His treatment began at a regional children's hospital, where initial spine radiographs demonstrated a 43.6° dextroscoliosis curve with the apex at L3. He was initially treated with a Boston brace, and the family was informed that MAGEC (Magnetic Expansion Control) growing rods were likely the definitive treatment due to the high likelihood of progression given the patient's large Cobb angle. However, the decision was made by the family and the Ehlers-Danlos syndrome specialist to proceed with the Wood-Rigo-Cheneau derotational brace.

BACKGROUND

Ehlers-Danlos syndrome (EDS) is comprised of a group of inherited connective tissue disorders. There are currently 14 recognised types characterised by protein and gene mutations that result in abnormal connective tissue. It has been shown to affect types I, III and V collagen, and specific mutations have been attributed in 13 of the 14 known subtypes.[1–3] Patients with EDS are generally characterised by skin hyperextensibility, generalised joint hypermobility, easy bruising, and other soft tissue, organ and orthopaedic manifestations.[2 4 5] The type relative to this case study, arthrochalasia EDS, is especially rare. In arthrochalasia EDS, variants are found in the COL1A1 or COL1A2 genes and result in the loss of exon 6 sequences from the mature messenger RNA. Patients with arthrochalasia EDS, in particular, often present with congenital hip dislocations and extreme joint hypermobility.[6]

The major criteria for diagnosis of arthrochalasia EDS include congenital bilateral hip dislocation, severe joint laxity with multiple dislocations/subluxations and skin hyperextensibility. Minor criteria include muscle hypotonia, kyphoscoliosis, mild osteopaenia on radiographs, tissue fragility and easy bruising. According to the 2017 international classification of EDS, minimal criteria suggestive of arthrochalasia EDS include congenital bilateral hip dislocation plus skin hyperextensibility, or congenital bilateral hip dislocation with severe joint laxity and at least two other minor criteria.[3]

Scoliosis is often seen in patients with EDS. Most of the time, scoliosis can be treated conservatively with casting and bracing. However, in patients with larger curves, there is a risk of continued progression if left untreated. This impacts not only the musculoskeletal system, but other vital organ systems including the cardiopulmonary system. Specific findings that are associated with curve progression include a rib vertebral angle difference of >20° and a Cobb angle of >30°.[7] Treatment options in these instances include serial casting, bracing and surgery. Serial casting is well known and is considered the standard of care since the landmark 2005 study by Mehta,[8] which found resolution of scoliotic curves in 100% of patients treated with casting before the age of 2. Several braces have also been used in the treatment of scoliosis to prevent curve progression, including the Boston brace, Milwaukee brace, Wilmington brace and Wood-Rigo-Cheneau (WRC) derotational brace. We present a case of a patient with arthrochalasia EDS and a high-degree lumbar curve to highlight the use of the WRC derotational

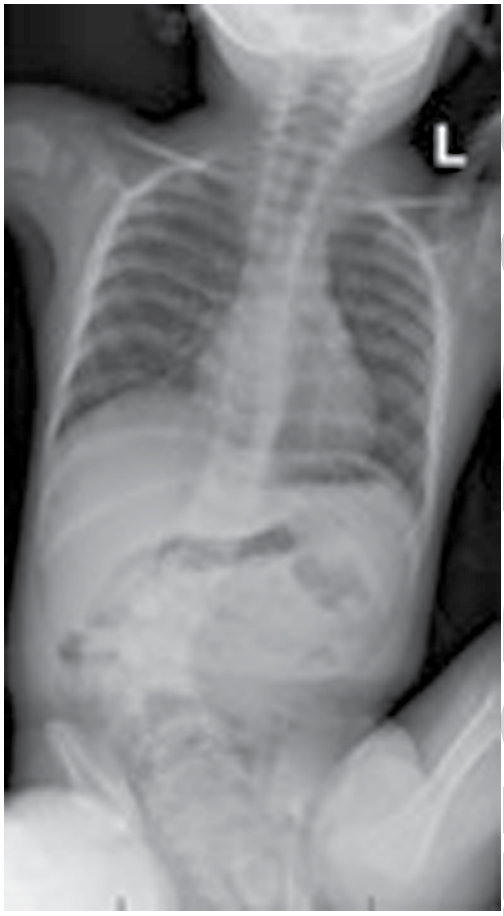

Figure 1 Anteroposterior radiograph of the spine demonstrating a 43.6° lumbar dextroscoliosis with an apex at L3 prior to the initiation of bracing.

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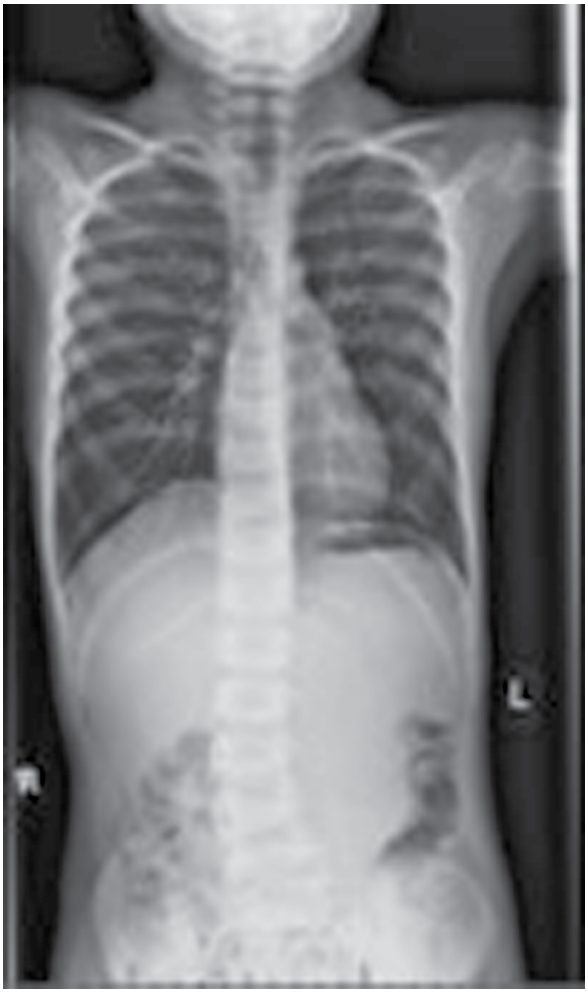

Figure 2 Anteroposterior radiograph of the spine demonstrating a 14° lumbar dextroscoliosis with an apex at L3 following treatment with the Wood-Rigo-Cheneau derotational brace.

brace as an effective conservative treatment option in syndromic scoliosis. To our knowledge, no study has evaluated the efficacy of the WRC brace as treatment for syndromic scoliosis.

CASE PRESENTATION

We present a boy in middle childhood with a medical history of arthrochalasia EDS. Genetic testing was performed which identified a significant finding: a pathogenic variant, specifically the c.279 G>A mutation within the COL1A2 gene, consistent with a diagnosis of arthrochalasia EDS. His parents noted a progressive curvature of his spine, and he was subsequently diagnosed with scoliosis before the age of 2. Spine radiographs obtained at an outside children's hospital demonstrated a 43.6° lumbar dextroscoliosis with an apex at L3 (figure 1). He was initially treated with a Boston brace, and the family was informed that MAGEC (Magnetic Expansion Control) growing rods were likely the definitive treatment due to the high likelihood of progression given the patient's large Cobb angle. Four months later, the patient's EDS specialist suggested the WRC brace as an alternative, supported by a second opinion at a different academic children's hospital. At the visit with the second spine specialist, the family was informed that the WRC brace would be ineffective secondary to the patient's underlying connective tissue disorder; the family was also recommended surgery. Through shared decision making with their EDS specialist, the family elected to proceed with conservative management using the WRC brace, in conjunction with weekly physical therapy.

OUTCOME AND FOLLOW-UP

After 3.5 years, the patient's Cobb angle decreased to 8°. His Cobb angle was stable under 15° on brace for 12 months. At the age of 6, after being out of brace for 6 months, his curve measured 14° (figure 2). Although he is brace-free, given his young age, future vertical growth, extraordinary amount of joint hyperflexibility and low muscle tone, he will continue to be followed closely in the clinic until skeletal maturity (Risser 4). If his Cobb angle were to progress towards 20°, the WRC brace would be reinstituted. It is important to note that the patient did not experience any major complications from wearing the brace and was able to participate in physical therapy without physical limitations.

DISCUSSION

Scoliosis can be successfully treated with either observation or conservative management, including casting or bracing. Serial casting can be an effective means of decreasing Cobb angles, especially if started before the age of 2 years.[7 9] Although casting increases compliance due to inability to remove them easily, there are concerns regarding their safety because there is a need for young children to undergo general anaesthesia multiple times for cast changes, which could lead to significant negative neurodevelopmental effects.[10 11]

Bracing can be considered an alternative conservative option to control curve progression as it eliminates the need for general anaesthesia and its potential neural toxicity. It is also cheaper and more convenient for patients and their families.[12 13] The Boston brace has been considered the gold standard, but other options include the Milwaukee brace, the Wilmington brace and the less widely known WRC brace. Literature supports the use of bracing as an effective means of preventing surgery in adolescent idiopathic scoliosis (AIS); however, no consensus exists for syndromic scoliosis.

Traditional bracing aims to correct scoliosis in the coronal plane only. However, scoliosis is a three-dimensional spinal deformity. Schlösser *et al*[14] describe the initiation of scoliotic deformity as an anterior vertebral overgrowth or an anterior lengthening of the spine. As this occurs, it creates a wedging effect that places tension on the spinal column. The normal thoracic kyphosis is then replaced with thoracic lordosis. In order for the body to balance the head over the pelvis, the spine rotates, causing scoliotic deformity.

Using computer-aided design/computer-aided milling (CAD/CAM) technology, three-dimensional braces such as the WRC brace can be customised to correct the deformity in all planes using derotation forces.[11] The well-described principles of correction by Sauvagnac and Rigo[15] include (1) regional derotation with cranial and caudal counter-rotation forces; (2) lateral and ventrodorsal contacts for frontal and sagittal plane alignment guidance; and (3) a special mechanism to fight against the structural lordotisation of the thoracic region, which depends on the level, shape and orientation of the contact areas of the brace. The level of the main derotational force is the apical region of the main curve or curves, with counter-rotation forces acting proximal and distal to the apex.[15] In essence, a three-point system is created to correct the curve in the frontal plane by applying force at the apex of the curve and counterforce proximal and distal to the apex. As for the sagittal plane, a normal physiological sagittal plane is dependent on pelvic incidence and lumbar lordosis. When designing the WRC brace, the objective is to use pads on either side of the spine to best restore physiological

sagittal balance.[16] Furthermore, the pads are placed in an oblique plane compared with a single plane to provide forces for derotation in the transverse plane.[17]

In the study performed by Thometz and Liu,[12] nine patients with infantile idiopathic scoliosis were treated using an elongation bending derotation brace, with a mean age at treatment of 11 months. Four patients were fully corrected to curves <10°, and the remaining five patients showed curve improvement from a mean of 57° to a mean of 21°, supporting the use of CAD/CAM technology.[12]

Minsk *et al*[18] compared the well-known Boston brace with the less-known WRC brace in patients with AIS. Patients with initial curves of between 25° and 40° were retrospectively reviewed from their initial treatment using either Boston or WRC brace until skeletal maturity or surgery. Patients treated with the WRC brace experienced significantly lower rates of spinal surgery and had lower mean and percentage of major curve progression compared with patients treated with the Boston brace.

Rivett *et al*[19] looked at the effectiveness of the WRC derotational thoracic-lumbar-sacral-orthosis (TLSO) scoliosis brace in 51 female patients with AIS and Cobb angles between 20° and 60°. The study looked at the effectiveness of the WRC brace with regard to brace compliance. The patients also followed a specific scoliosis-focused exercise programme. One study group, labelled the compliant group, wore the brace at least 20 hours a day (average of 21.5 hours a day) and exercised three times a week or more (average of four times a week). The other group, labelled the non-compliant group, wore the brace less than 20 hours a day (average of 12 hours a day) and exercised less than three times a week (average of 1.7 times a week). The results were significant in that the compliant group improved on average 10.19°±5.5°, while the non-compliant group showed deterioration of 5.52°±4.3° on average.[19]

Ovadia *et al* [20] looked at whether early bracing using the WRC brace would decrease advancement of AIS. Their study included 93 patients with AIS and an average Cobb angle of 31.97° and Risser score of 1.07. The patients were followed until they reached a Risser score of 4, with no vertical growth greater than 1 cm for 6 months and then for 12 months after. The patients were instructed to wear the WRC brace 20–22 hours a day. They found that 83.8% of the patients (n=79) who used the brace as prescribed showed not just lack of progression of their Cobb angle at 28.97° (p=0.022), but a slight improvement in their curve. Those in the treatment failure group (n=14) showed, on average, a worsening Cobb angle to 55.93° (p=0.0001). These results support the use of the WRC brace in patients with AIS who were compliant, showing a halting of scoliosis progression and even mild improvement of their Cobb angle.

Babaee *et al*[10] evaluated the effectiveness of brace treatment using either a TLSO or a Milwaukee brace in patients with IIS until skeletal maturity or spinal fusion. They found that bracing was successful in more than two-thirds of patients with IIS curves, preventing surgery before the age of 10.

In conclusion, EDS comprised a group of inherited connective tissue disorders, with 14 recognised types characterised by protein and genetic variants that result in abnormal connective tissue. In the present case, the patient was diagnosed with arthrochalasia EDS based on clinical information and genetic testing. There is sparse research on arthrochalasia EDS. Current literature highlights certain clinical manifestations such as congenital bilateral hip dislocation, severe generalised hypermobility, recurrent dislocations and skin hyperextensibility.[5 21 22] In addition, Ayoub *et al*[21] highlight less common manifestations, including scoliosis.[5] Early diagnosis with genetic testing will help guide families and providers in the treatment of the various signs and symptoms associated with arthrochalasia EDS.

Patient's perspective

After the child was in the second spica cast, we noticed the child's back might be turning. We brought this to the attention of the doctors at a well-known children's hospital who eventually agreed it was scoliosis. Unfortunately, the child had to be treated for his hip dysplasia first. This was concerning for a first time parent to add another diagnosis. We tried the Boston brace first, which is basically a tube and did nothing as the child's curve continued to increase. The well-known children's hospital wanted to start magnetic rod surgery, but we did not so we obtained a second opinion, which was the WRC brace, which is amazing. The child has never complained of pain from the scoliosis. There was some mild discomfort from the brace, but the child got used it. Why is the WRC brace amazing- my child had a rib hump, curve was over 50° and now it's between 11 and 20° and rub hump is gone. As the child begins to curve again with growth, we fully expect the WRC brace to achieve the correction that the child has a historically experienced. As a parent, I found it best to advocate for my child to have the least restrictive and least invasive methods especially when taking the complex medical history into consideration, a.k.a. history of bilateral hip dysplasia and Ehlers Danlos syndrome.

For all the parents that complain that their child will not wear a brace 23 hours a day, it was explained in great detail to the child that if the child did not wear the brace, the child would have to have back surgery which was explained in age-appropriate terms. The child wore the brace mostly without complaint, understanding the consequences. Unfortunately, when the child ran a high fever or had skin breakdown, the child had to take a break from the brace, yet still achieved correction. The orthotist did closely monitor how skin breakdown and fevers were handled in regard to brace wear time. I personally also think WRC brace is more humane than infantile back casting as WRC reduces the skin breakdown as skin can be monitored, washed, lotioned and treated.

One of the biggest problems we did encounter was the issue of insurance not wanting to cover the brace due to it being out of network. This resulted in me having to learn how insurance works and jump through a lot of hoops to achieve coverage. Never underestimate the power of a care manager from your health insurance company as they can help with that though this is not advertised.

In conclusion, WRC brace has been completely worth it as my boy in middle childhood is presently functioning at age-appropriate levels without restriction. Scoliosis is barely visible; rib hump has disappeared. My boy in middle childhood has no pain. Currently, the child does not need a back brace again yet. When the child reaches the point of needing a back brace again, I will turn to the WRC and we will use it again as I firmly believe that this is the best treatment available for my child and the results speak for themselves.

One aside, my boy in middle childhood has been in PT for years to aid in strengthening of back muscles and also is kept in private classes, currently gymnastics, basketball and swimming. I believe regular exercise to strengthen core muscles (especially for people with EDS) is very helpful along with the WRC brace in achieving the goal of a straight back. The child was originally very floppy in muscle tone after coming out of the spica cast years ago and the child has exceeded expectations of all of medical providers in function.

Continued

Patient's perspective Continued

My boy in middle childhood presents as a normal boy, who keeps up with the other children with no problem. In fact, one would not know the child was any different by looking at the child unless the child is demonstrating the child's hypermobile talents or shows off the foot orthotics that are worn for pes planus of both feet. Whenever a WRC brace is needed again, we will have support from the school if extra help is needed at that point, but this has yet to be needed. This is very stressful for a parent to go through, but the results have been totally worth it. This is the best case scenario for the child- not needing to have back surgery at all. I have been told the child may eventually need back surgery, possibly when the child is done growing, but even if that ends up being the case- that is one surgery, not many beginning at age 3. We all remain positive the child will not and expect surgery to not be needed at all since the child has remained stable without a back brace over the last year.

The WRC brace has allowed my child to achieve correction, live a pain free and unrestricted life and avoid potential complications from back surgery. I firmly believe WRC brace should be a standard of care, more widely known and advertised as it could really benefit babies and children with scoliosis. It would also help if some providers wouldn't scare off parents about the price, tell them it won't work, and jump straight to surgery. I am glad we did not listen and went for the second opinion. The chance of correction was worth it and it was achieved in the least restrictive and least invasive way possible.

It was also nice to take my child on vacation and be able to remove the brace while physically at the beach (it was worn at all other times except bath) and the doctor knew we were doing this so the child could enjoy the vacation.

Learning points

- There are several subtypes of Ehlers-Danlos syndrome (EDS) and patients with the arthrochalasia type often present with excessive joint laxity compared with patients with other subtypes.
- There is evidence-based support on the use of Wood-Rigo-Cheneau (WRC) brace in patients with low Risser scores and mild to moderate Cobb angles (20°–45°).
- With a treatment plan to continuously wear the brace 23 hours a day, regular visits to the spine specialist for assessment of radiographic and physical changes, as well as seeing a WRC brace-trained orthotist for assessment of fit and function, the WRC brace has the potential to become a very viable conservative option for patients with syndromic scoliosis.
- Since scoliosis is a complex condition that causes spinal deformities in three distinct planes, we believe the WRC brace should be considered a useful non-operative treatment modality as it addresses the three-dimensional deformity created by scoliosis not well addressed by other long-accepted Boston-style braces.

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