

The Role of Exercising in a Pair of Female Monozygotic (High-Class Athletes) Twins Discordant for Adolescent Idiopathic Scoliosis

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Study Design. The report of 2 cases and review of the literature.

Objective. To report the cases of a pair of female monozygotic (high-class athletes) twins discordant for adolescent idiopathic scoliosis.

Summary of Background Data. The relation between scoliosis and exercising is rather unclear. The latter has often been considered both as a therapeutic means and a causative factor of the former. The existence of genetic predisposition in the development of adolescent idiopathic scoliosis is commonly accepted. According to the best of our knowledge, this is the first report of a pair of female monozygotic (high-class athletes) twins, discordant for adolescent idiopathic scoliosis.

Methods. A pair of 13.5-year-old female monozygotic twins, high-class level athletes of synchronized swimming, was clinically examined during a school screening program. Both girls were observed in the standing erect position for asymmetries of the lateral contours of the trunk, shoulders, and scapulas and their limb's length was measured. The "forward bending test" was performed to determine the existence of rib hump asymmetry.

Results. One of the sisters was considered to be suspicious of suffering from scoliosis. The radiologic evaluation that followed confirmed the existence of adolescent idiopathic scoliosis (left thoracolumbar curve of 32° as measured by the Cobb angle). The clinical and radiologic evaluation of her sibling failed to reveal the existence of any spinal deformity.

Conclusion. Adolescent idiopathic scoliosis seems to be a multifactorial skeletal disorder. The role of exercising and heredity in its development remain controversial.

Key words: adolescent idiopathic scoliosis, exercising, monozygotic twins, exercising and adolescent idiopathic scoliosis, etiology of adolescent idiopathic scoliosis.
Spine 2008;33:E607–E610

The exact cause(s) of adolescent idiopathic scoliosis (AIS) remain(s) more or less unclear. Extensive scientific research has not managed (so far) to adequately uncover the actual pathogenetic mechanisms that are implemented in the development of AIS.

AIS often develops in several members of large families, suggesting that its incidence is influenced, to some extent, by genetic factors as well.¹ Studies on concordance for AIS in monozygotic twins provide an excellent basis for analyzing the relative influence and importance of genetic *versus* environmental factors in the etiology of AIS.^{2–4} If a pair of monozygotic twins is suffering from an entirely genetic disorder, both siblings ought to be affected in a similar way, because their genome is identical. Therefore, in theory, if a pair of monozygotic twins is "exposed" to a given environmental factor, they should react in an identical or at least similar pattern. The relation between AIS and exercising (a typical extragenetic or "environmental" potentially causative factor) is rather ambiguous. The latter has often been considered both as a therapeutic means and a causative factor of the former.

We report the cases of a pair of female monozygotic twins (high-class synchronized swimming athletes) discordant for AIS. To the best of our knowledge, this is the first report of such a case.

■ Case Report

A pair of monozygotic, female, white, 13.5-year-old twins was examined during a school screening program. Monozygosity was initially defined as twins having completely similar physical characteristics (*i.e.*, brown color of eyes and hair, identical skin type, same facial characteristics), nearly indistinguishable fingerprints and their parents' report of finding a single placenta at birth. It was further validated by performing blood tests that revealed identical blood group and histocompatibility antigens and negative reaction in mixed lymphocyte culture. Both girls had exactly the same height (171 cm), almost identical body weight (59.5 and 60 kg, respectively), the same prevalent extremity (right), and almost simultaneous (reported) onset of menstruation. Their medical history was free and none in their family was suffering from AIS. No history of major trauma was reported either. Both girls were high-class athletes (second and third place winners at the national championship). They continuously and systematically practiced synchronized swimming for the last 5 years before their examination, following an al-

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Acknowledgment date: June 1, 2007. First revision date: November 25, 2007. Second revision date: March 5, 2008. Acceptance date: March 8, 2008.

The manuscript submitted does not contain information about medical device(s)/drug(s).

This study has been approved by the Institution's Review Board and was conducted in accordance with the World Medical Association Declaration of Helsinki of 1975, as revised in 1983 and 2000. After the patients' parents were fully informed, they consented that data concerning their children could be submitted for publication.

No funds were received in support of this work. No benefits in any form have been or will be received from a commercial party related directly or indirectly to the subject of this manuscript.

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Figure 1. The twin girls observed in the standing erect position-dorsal view. The girl suffering from adolescent idiopathic scoliosis is on the right.

most identical professional training schedule of at least 20 h/wk as they were members of the same athletic team.

Both girls were observed in the standing erect position for asymmetries of the lateral contours of the trunk, shoulders, and scapulas (Figures 1 and 2). Clinical examination failed to reveal any site of tenderness along the spinous processes of their vertebrae, vertebrocostal junctions, sacroiliac joints, or any other part of their bodies. Both girls were able to perform full range of motion (active and passive) movements (spine, hip joints) without reporting any pain. No discomfort was reported occurring during their training schedule either. The “forward bending test” was performed next to determine the existence of a rib hump asymmetry and the length of their limbs was measured (Figure 3). As one of the sisters was considered to be suspicious of suffering from scoliosis, they both underwent further standard radiologic evaluation that confirmed the existence of AIS in the “suspicious” one (left lateral convex thoracolumbar



Figure 2. The twin girls observed in the standing erect position-lateral view. The girl suffering from adolescent idiopathic scoliosis is on the right.



Figure 3. The twin girls performing the “bending test.” The girl suffering from adolescent idiopathic scoliosis is on the right.

curve of 32° as measured by the Cobb angle, from T11 to L3 vertebrae, with the apex at the T12–L1 disc space). Both clinical and radiologic evaluation of her sibling failed to reveal the existence of AIS (Figures 4 and 5). As the T12 costae of the scoliotic twin seemed to be shorter than her sister’s and L5 appeared to be somewhat tilted (thus implying the existence of sacral inclination), we obtained a “Ferguson” view of the sibling suffering from scoliosis that failed to reveal any significant pathology (Figure 6).

The girl found to suffer from AIS was given a thoracolumbar Boston brace. An in-brace correction of 8° was achieved and no progression of the scoliotic curve was noticed on her latest follow-up visit, 24 months later. Her sister is still free of AIS. Both children remain under constant medical observation.

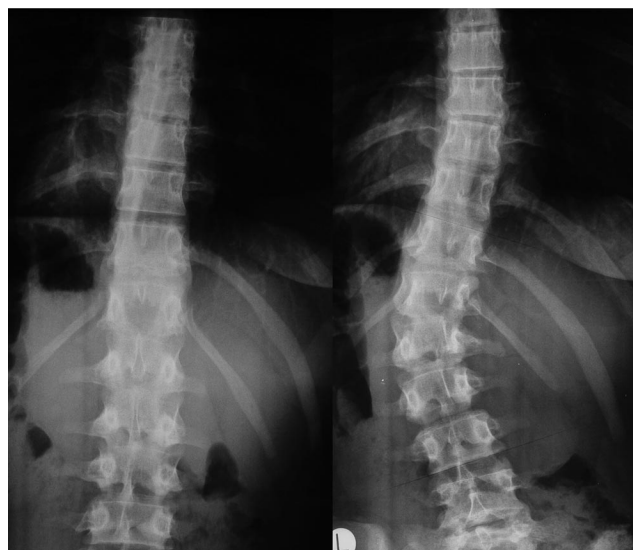


Figure 4. The standard posteroanterior radiographs of the spines of the twins revealed the existence of a left lateral convex thoracolumbar curve of 32° in only one of them.



Figure 5. Standard laterolateral radiographs (right: scoliotic twin).

■ Discussion

The actual cause(s) of AIS remain(s) more or less unspecified. Several theories have been proposed, and many factors have been associated with the development of AIS; nevertheless, both its exact pathogenetic mechanism

and the existence of possible contributing factors to its development remain (at the moment) unclear.

Studies in twins are of considerable value in the investigation of genetic and environmental etiologic factors influencing or predisposing to a certain disorder.^{1,2} Given the fact that they share an identical genome, monozygotic twins ought to be concordant for any given genetic disorder. Nevertheless, the expression of most inheritable disorders seems to be affected from environmental factors as well. Therefore, discordance between monozygotic twins may be presumed to be environmental in origin.⁴ This presumption, however, has recently been the subject of controversy.

On the one hand, the incidence of monozygotic twins concordant for AIS has been reported to rise as high as in 93% of all reported cases¹ and that seems to be a strong evidence for the genetic origin of AIS.⁵ The latter is further supported by an article published by Kesling *et al*,⁶ which showed a statistically significant difference in the concordance between monozygotic and dizygotic twins. Therefore, one might expect that all reported monozygotic twins are concordant for AIS, regardless of the severity and the pattern of the scoliotic curve that seem to be influenced by extragenetic factors as well.³ This, however, may certainly not be the case as many other contributing factors (the way monozygosity is determined,



Figure 6. The "Ferguson" radiographic view of the scoliotic twin failed to reveal any pathologic entity.

the existence of different intrauterine environments, the occurrence of mutation after fertilization and/or the initial cleavage, the influence exerted by environmental factors after birth⁶) may play a potentially important role in the etiology of AIS. This “paradox” is further enhanced in the light of a recently published article by Andersen *et al.*² The authors performed a population-based twin study with 11,740 complete pairs of twins and reported a proband-wise concordance of 0.25 for monozygotic pairs. They concluded that both genetic and environmental factors must be important in the etiology of AIS.

The exact relation between AIS and exercising is rather ambiguous. The latter has been considered both as a therapeutic means and a causative factor of the former. There are few studies in the literature that try to evaluate the potential connection between exercising and scoliosis. The prevalence of scoliosis has been reported to rise rather high among leading athletes. Becker *et al.*⁷ examined 336 swimmers (193 women and 173 men) and reported a total incidence of structural scoliosis of 6.9% in each team. Warren *et al.*⁸ reported 18 out of a group of 75 in total professional female dancers to suffer from AIS. They attributed this extremely high prevalence of scoliosis (24%) to the relatively more delayed onset of menstruation in athletes with AIS when compared with the healthy ones and to the high percentage of a positive family history in the dancers suffering from AIS. Tanchev *et al.*⁹ estimated the prevalence of scoliosis among 100 female rhythmic gymnasts to rise as high as 12%. The authors compared these results with those of a school screening program (with a reported prevalence of AIS of 1.1%) conducted by them as well.

The study of a pair of female monozygotic twins seems to be ideal in order to determine whether a certain environmental factor may contribute in the development of a certain disorder or not, especially when taking into account of course, the suggestion that there is a significant genetic component in the etiology of AIS. Because our 13.5 years-old twins female patients share a common genetic background, are exposed to the same environmental factor (synchronized swimming) and in the same manner (almost identical professional training schedule of at least 20 h/wk for the, “crucial” as far as the development of AIS is concerned, 5 years before their initial examination), one might expect that they would show concordance with AIS. This “expectation” is further enhanced by the fact that monozygotic twins have a good chance (ranging from 25%² up to 73%⁶ or even 94%¹) of being concordant for AIS. Nevertheless, this anticipation is not confirmed in the hereby reported cases.

It is certainly true that we cannot reach secure conclusions about the potential relation between AIS and exercising based only on a pair of patients. Nevertheless, the reported cases of elite athletes twins discordant for AIS questions the reported by several authors “cause and effect” relationship between AIS and exercising.

AIS is probably a multifactorial skeletal deformity. It seems that several factors, as exercising and heredity, are involved in its development. It is our belief that more studies are needed to better understand the potential connection (if any) between exercising and AIS.

■ Key Points

- Studies on monozygotic twins are of great value in the understanding of genetic and environmental etiologic factors.
- Exercising is considered to be a therapeutic means and a causative factor of adolescent idiopathic scoliosis.
- Concordance for adolescent idiopathic scoliosis in monozygotic twins rises as high as 93%.
- Because monozygotic twins share a common genetic background, their “response” to a certain environmental factor should be identical or at least almost similar.
- The hereby reported pair of monozygotic twins questions the theory that exercising might be a single etiologic factor of adolescent idiopathic scoliosis.

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