Curve characteristics in monozygotic twins with adolescent idiopathic scoliosis

3 new twin pairs and a review of the literature

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ABSTRACT – Most authors state that there is strong evidence for a genetic origin of adolescent idiopathic scoliosis (AIS). This conclusion is mainly based on the fact that the rate of concordance for AIS in monozygotic twins is significantly higher than that in dizygotic twins. However, it is of interest to determine whether all elements of scoliosis formation are genetically predetermined. If this were the case, there would perhaps be less place for closed treatment.

We surveyed the literature for monozygotic twin pairs in which both members suffered from idiopathic scoliosis and added 3 pairs from our own patient group. The total group consisted of 32 twin pairs.

We found that gender, direction of the convexity, the level of the apex and the kyphotic angle were determined more by genetic factors than the lateral Cobb angle of the scoliotic curve. This suggests that variations in the environment may affect the curve patterns in monozygotic twins.

The etiology of adolescent idiopathic scoliosis (AIS) is still unclear. Studies on concordancy for adolescent idiopathic scoliosis in monozygotic twins provide a basis for analyzing the relative influence of genetic versus environmental factors.

Twin pairs are classified as concordant, when both twins have scoliosis, or discordant when only one has a scoliotic curve (De George and Fisher 1967, Kesling and Reinker 1997). Concordancy does not mean that the severity or pattern of the curves is similar in these twins.

We surveyed the literature and added 3 new twin pairs to evaluate the similarities and differences between scoliotic curves in monozygotic concordant twin pairs. A comparison of these parameters may help elucidate the etiology and pathogenesis of AIS by indicating which characteristics of the disorder are probably caused by genetic factors and which by the environment.

Patients and methods

A literature search was done with Pubmed, using the words 'monozygotic twins' and 'idiopathic scoliosis' as search keys. Only articles on concordant monozygotic twins with sufficient data on the twins' curve patterns were included. Monozygosity was defined as twins having completely similar physical characteristics or identical DNA fingerprinting. Patients were considered to have idiopathic scoliosis only if their curve showed a lateral Cobb angle of at least 10°.

We also added 3 pairs of girls from our own patient group who had no other abnormalities of the spine on clinical and radiographic examinations. They were classified as monozygotic because of their completely similar physical characteristics– i.e., color of eyes, hair and skin. We analyzed the following parameters: direction of the convexities, lateral and kyphotic Cobb angles and location of the apex. The statistical analysis was done with the paired samples t-test.

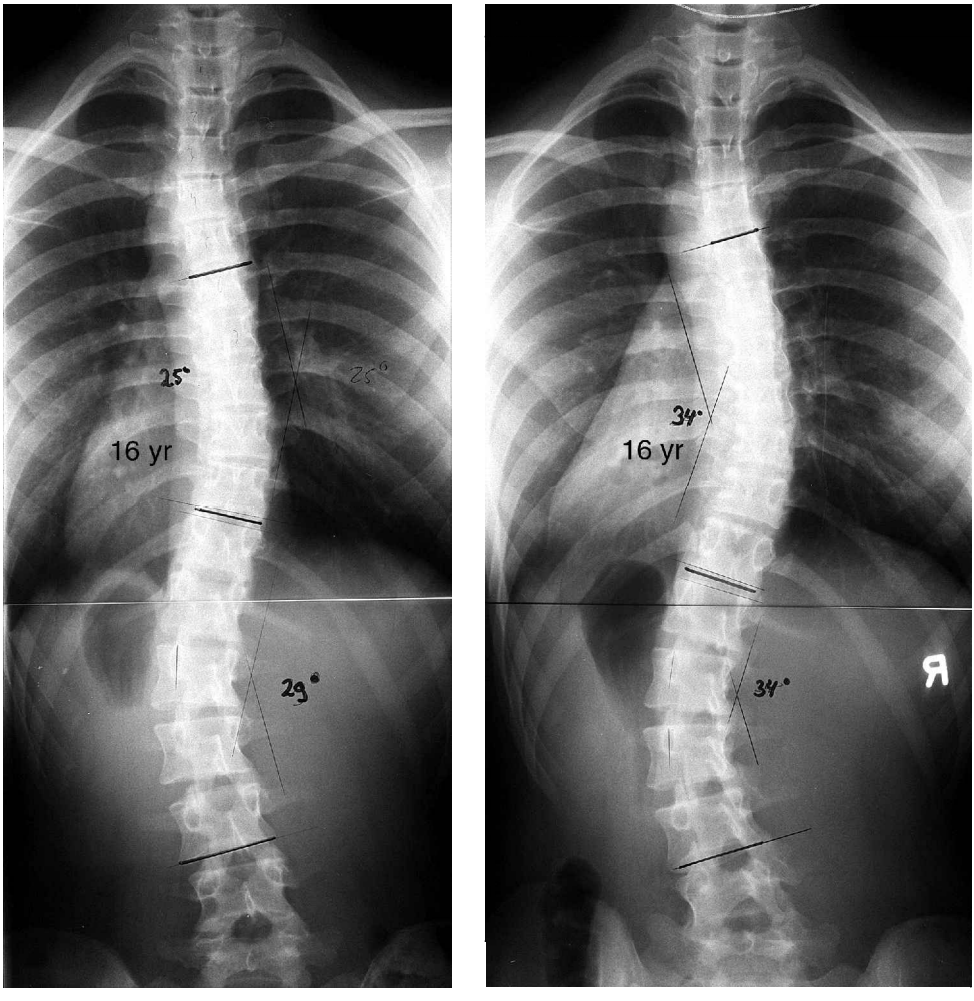

Figure 1. Pair 1.

Case reports

Pair 1. Both children were first given a brace for progressive idiopathic scoliosis at the age of 14 years. Twin I had a right convex curve of 26° and left lumbar curve of 33°. After this treatment, the thoracic curve was 25° and the lumbar curve 29°. Twin II had a right convex curve of 32° and left lumbar curve of 43°. After brace treatment, both curves were 34° (Figure 1).

Pair 2. The twins were started on brace treatment for progressive idiopathic scoliosis at age 14. Twin I had a 28° right thoracic curve with an in-brace correction of 7°. After this treatment, the thoracic curve was 18°. Twin II had a 32° right thoracic curve with an in-brace correction of 12°. After brace treatment the thoracic curve was 19° (Figure 2).

Pair 3. These twins were first seen at the age of 13 years. Twin I already had a right convex curve of 78° and underwent surgery with Harrington instrumentation. After fusion, the curve was 58°. Twin II showed a right convex curve of 24° at the same age. 2 years later, despite brace treatment, it had progressed to 49°. She was also operated on and, after fusion the curve was 35°.

Literature review

32 twin pairs were included using the above criteria. All twins were girls (Table). The mean Cobb angle in all 64 patients was 37° (SD 19°). We compared the lateral Cobb angles in each twin pair. 17 pairs had a difference in curve severity of less than

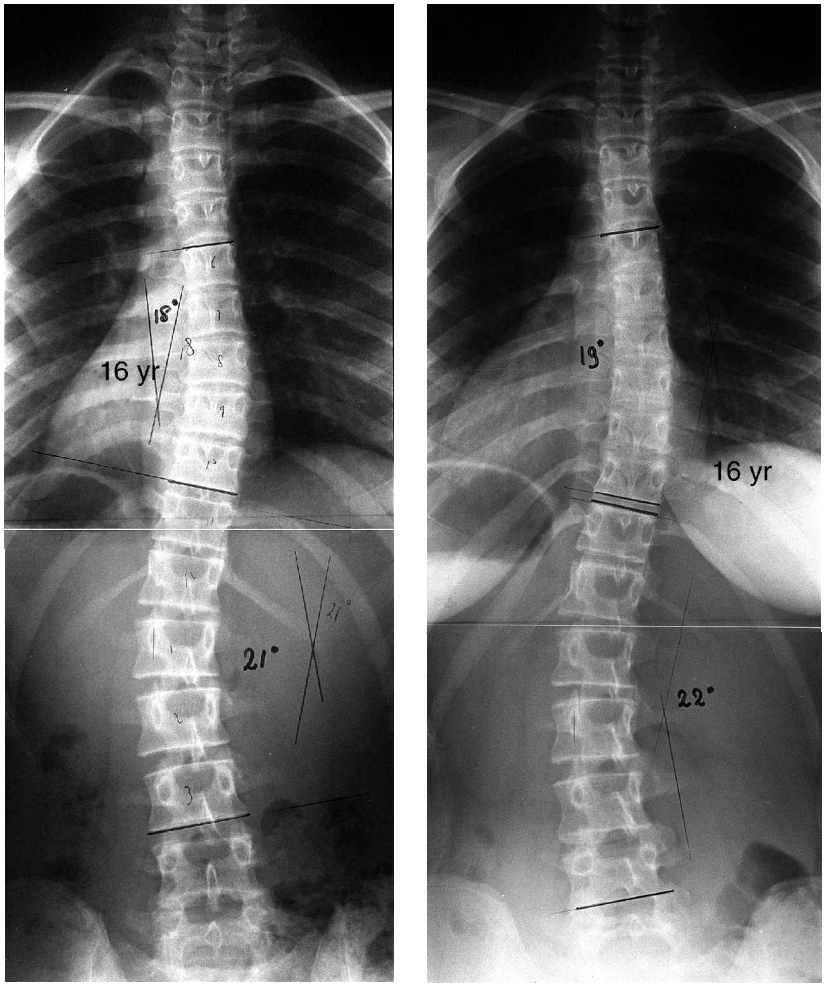

Figure 2. Pair 2.

10°, 9 pairs between 11° and 20° and 6 pairs more than 20°. The mean difference between the twin curves was 12° (SD 12°).

The direction of the convexity was similar in 24 of the 26 twin pairs in whom it had been reported. The location of the apex could be compared in 18 pairs. In 11, the apex was at the same location; 5 twins differed by one segment, 1 twin two segments, and 1 three segments.

The kyphotic Cobb angles were available in only 15 of the twin pairs. Of these, 11 had differences in the kyphotic Cobb angle of ≤ 3°; 1 between 4° and 9°; and 3 pairs had a difference of ≥ 10°.

The vertebral rotation of the apex was not mentioned in any of the articles reviewed. We assessed this parameter in our 6 patients, using Perdriolle and Vidal's method (1981). In 2 twin pairs, both members had their most rotated vertebrae in the lumbar curve and in the remaining pair, both members had their most rotated vertebrae in the thoracic curve.

Discussion

Monozygotic twins are oftener concordant for AIS than dizygotic ones (Fisher and De George 1967, Wynne-Davies 1968, Kesling and Reinker 1997, Inoue et al. 1998). Inoue et al. (1998) found concordance in 12 of 13 monozygotic twin pairs and in 5 of 8 dizygotic twins. In their meta-analysis, Kesling and Reinker (1997) reported concordance

Clinical characteristics of concordant monozygotic twins with idiopathic scoliosis

Reference	Pairs	Gender	Age studied years	Curve pattern Curve 1	Apex	Curve 2	Apex	Cobb angle in degrees Lateral curve 1	Lateral curve 2	Kyphotic	Rotation curve 1	Rotation curve 2
Depalma 1961	1	F	13	Right T5-L1	T9			45				
				Right T5-L1	T9			56				
Cordorniu 1958	2	F	13	Right Thoracic	T7			35				
				Right Thoracic	T6			42				
Esteve 1958	3	F	11	Right Thoracic	T7			46				
				Right Thoracic	T7			58				
Murdoch 1959	4	F	16	Right T5-T11	T8			46				
				Right T5-T11	T8			55				
Gaertner 1979	5	F	13	Right T4-T11	T8			58				
				Right T6-T11	T8			28				
	6	F	13	Right T5-T12	T8			74				
				Right T5-T12	T8			44				
Fisher 1967	7	F	13	Right T5-T12	T8	Left T12-L4		67	47			
				Right T4-T11	T8	Left T12-L4		70	49			
	8	F	18	Right T5-T11	T8			43				
				Right T6-T12	T9			22				
	9	F	14	Left T5-T11	T8	Right T11-L3		41	46			
				Left T7-T12	T9			17				
	10	F	15	Left T6-T12	T9			12				
				Right T6-T12	T9			25				
	11	F	21	Right T2-T10	T6	Left T10-L4		87	84			
				Right T6-T12	T9			80				
	12	F	35	Right T5-T10	T7			42				
				Right T6-T10	T8			26				
Carr 1990	13	F	19	Right Thoracic				20				
				Right Thoracic				25				
	14	F	13	Right Thoracic				42				
				Right Thoracic				30				
Kesling 1997	15	F	14	Right T6-T11	T8	Left T12-L4		19	20			
				Left T6-T10	T8	Right T11-L2		19	16			
	16	F	12	Right T6-T11	T9	Left T12-L4		17	13			
				Right T7-T11	T9	Left T12-L3		18	15			
Vercauteren 1972	17	F	8	Right Thoracic	T7			23				
				Right Thoracic	T9			12				
Inoue 1998	18	F	22	Double				51	43	–8		
				Right Thoracic				68		–7		
	19	F	15	Right Thoracic				32		53		
				Double				31	32	36		
	20	F	18	Right Thoracic				64		19		
				Right Thoracic				53		20		
	21	F	19	Right Thoracic				32		38		
				Right Thoracic				25		32		
	22	F	14	Double				30	20	32		
				Right Thoracic				21		32		
	23	F	18	Right Thoracic				18		26		
				Double				25	22	28		
	24	F	14	Right Thoracic				25		0		
				Right Thoracic				27		0		
	25	F	12	Right Thoracic				80		26		
				Right Thoracic				40		15		
	26	F	11	Right Thoracic				42		0		
				Double				31	25	0		
	27	F	16	Right Thoracic				18		0		
				Right Thoracic				25		0		
	28	F	13	Right Thoracic				25		14		
				Right Thoracic				28		13		
	29	F	15	Double				18	10	28		
				Right Thoracic				15		25		
Van Rhijn 2001 (Authors' data)	30	F	16	Right T6-T11	T9	Left T12-L4	L1	25	29	21	0	17
				Right T6-T12	T9	Left L1-L4	L2	34	34	20	5	18
	31	F	16	Right T6-T10	T8	Left T11-L3	L1	18	21	9	0	12
				Right T6-T11	T8	Left T12-L5	L2	19	22	12	0	17
	32	F	13	Right T4-T10	T8			24		20	15	
				Right T6-L2	T9			78		50	30	

in 27 of 37 monozygotic pairs but in only 11 of 31 dizygotic twins. Most authors regard discordancy between monozygotic twins as evidence of an environmental origin (Carr 1990), and concordancy as due to genetic factors (Gaertner 1979). Kesling and Reinker (1997) found a higher concordance for AIS in monozygotic than in dizygotic twins and concluded that there is strong evidence for a genetic etiology in AIS.

However, it is also important to determine whether there are differences in the severity and pattern of the curve between members of concordant twin pairs. For instance, if a strong genetic factor is found for curve severity, scoliosis may be less influenced by closed (e.g., brace) treatment.

Two studies have evaluated the differences in Cobb angle between twins. Inoue et al. (1998) concluded that there was a genetic factor in curve severity because in 8 of 12 monozygotic twin pairs the difference between lateral Cobb angles was less than 10°. Kesling and Reinker (1997) compared 20 sets of monozygotic twins to 16 sets of dizygotic twins. The correlation coefficient for the lateral Cobb angle in monozygotic twins was 0.74, in dizygotic twins it was 0.40. No information was given concerning the mean values of the Cobb angles.

In our study, only half of the twin pairs showed a difference in lateral Cobb angles of less then 10°. These findings suggest that curve severity may be affected by the environment.

Kesling and Reinker (1997) noted that monozygotic twin pairs had very similar curve patterns. Inoue et al. (1998) compared patterns in 12 monozygotic twins and confirmed that in most pairs, the shape of the scoliosis was similar. Although 6 of 12 pairs showed discordant curve patterns.

In our study, nine tenths of the apices were at the same level or within one segment of the corresponding twin. Furthermore, the direction of convexities was similar in nine tenths of the twins. Most twins showed hardly any differences in kyphotic Cobb angle. These findings suggest a genetic influence on curve pattern. However, due to incomplete data, we could not compare the curves in our survey using a proper classification system.

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