Clinical Case Report

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Familial lumbar Scheuermann disease with idiopathic scoliosis in China

First case report

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Abstract

Rationale: Given that Scheuermann disease rarely occurs in the lumbar region and that the co-occurrence of Scheuermann disease and idiopathic scoliosis (IS) has not been reported—the etiology of Scheuermann disease and IS is not clear. In this case report, we present familaiar lumbar Scheuermann disease with IS, in a Chinese proband, who was successfully treated with surgery.

Patient concerns: A 16-year-old boy presented at the Second XiangYa Hospital of Central South University with a chief complaint of kyphotic deformity in the lower back for 4 years and obvious lower back pain. In addition, he complained of limited lumbar activity. And The proband's family history was obtained by routine inquiring. In this Chinese family with 17 members over 3 generations. The 3 patients (proband, proband's sister and father) shared the characteristics of vertebral wedging from L1 to L3 and a kyphosis Cobb angle of 37°, 70°, or 73°, respectively. The main deformity of the proband's mother was at T7-L1 with a Cobb angle of 102° in the coronal plane at T7-L1, thoracic kyphosis of 73°, and lumbar lordosis of 62°.

Diagnoses: Scheuermann's disease.

Interventions: Clinical history, physical examination, laboratory tests, and radiographs of those in the pedigree were recorded, and the related literature was reviewed. The proband accepted osteotomy and orthopedic surgery for treatment.

Outcomes: After 3 months of treatment, postoperative lateral radiographs showed a significantly improved sagittal vertical axis (SVA). The other patients were continued to be seen in follow-up visits.

Lessons: This series of lumbar Scheuermann patients with IS in a pedigree support the genetic contribution to Scheuermann disease. Therefore, this study provides some insight into the genetic etiology of Scheuermann disease with IS.

Abbreviations: AIS = adolescent idiopathic scoliosis, F-S-AP and LAT X-ray = full spine anteroposterior and lateral X radiographs, IS = idiopathic scoliosis, LL = lumbar lordosis, SVA = sagittal vertical axis, TK = thoracic kyphosis.

Keywords: genetic etiology, idiopathic scoliosis, lumbar vertebra, pedigree, Scheuermann disease

1. Introduction

Scheuermann disease is one of the most common reasons for hyperkyphosis in patients with deformed spine after idiopathic scoliosis (IS),[1,2] which is characterized by vertebral body wedging, vertebral endplate irregularity, diminished anterior vertebral growth, Schmorl's nodes, narrowing of the intervertebral disk spaces, and premature disk degeneration.

The prevalence of Scheuermann disease varies from 0.4% to 10%,[3,4–11] and typically occurs from 4 to 16 years of age, most commonly between the ages 12 and 15.[12–15] It is reported that it is an euchromosomal dominant genetic disease [16–19] and causes a significantly worse deformity than postural kyphosis.[20]

IS affects about 2% to 3% of adolescent women aged 10 to 16 years old.[21–23] Scoliosis is a postural deformity characterized by a lateral curvature of the spine greater than 10°, measured by the Cobb method on standing upright spinal radiographs.[24] While most cases of scoliosis are classified as idiopathic,[14] a minority of scoliosis cases are traced to structural anomalies,[25] such as wedged vertebrae or abnormal soft tissue development.

However, Scheuermann disease seldom occurs in the lumbar region, and the co-occurrence of Scheuermann disease and IS has not been reported previously in the literature. Here, we report an interesting case of familial lumbar Scheuermann disease with IS in a Chinese proband, who was successfully treated with surgery.

2. Case report

A 16-year-old boy presented at the Second XiangYa Hospital of Central South University with a chief complaint of kyphotic deformity in the lower back for 4 years and obvious lower back pain. In addition, he complained of limited lumbar activity. This patient was specified as the proband in this study.

General examinations showed that the proband was 165 cm tall, weighed 47 kg, had kyphotic deformity of the lower back and bilateral elbow flexion contracture, tested negative for the Lasegue test at 70°, and experienced no positive neurological

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Patient consent: The patients provided written consents for publishing this case report and further studies.

Competing interests: There were no competing interests in this case report.

The authors report no conflicts of interest.

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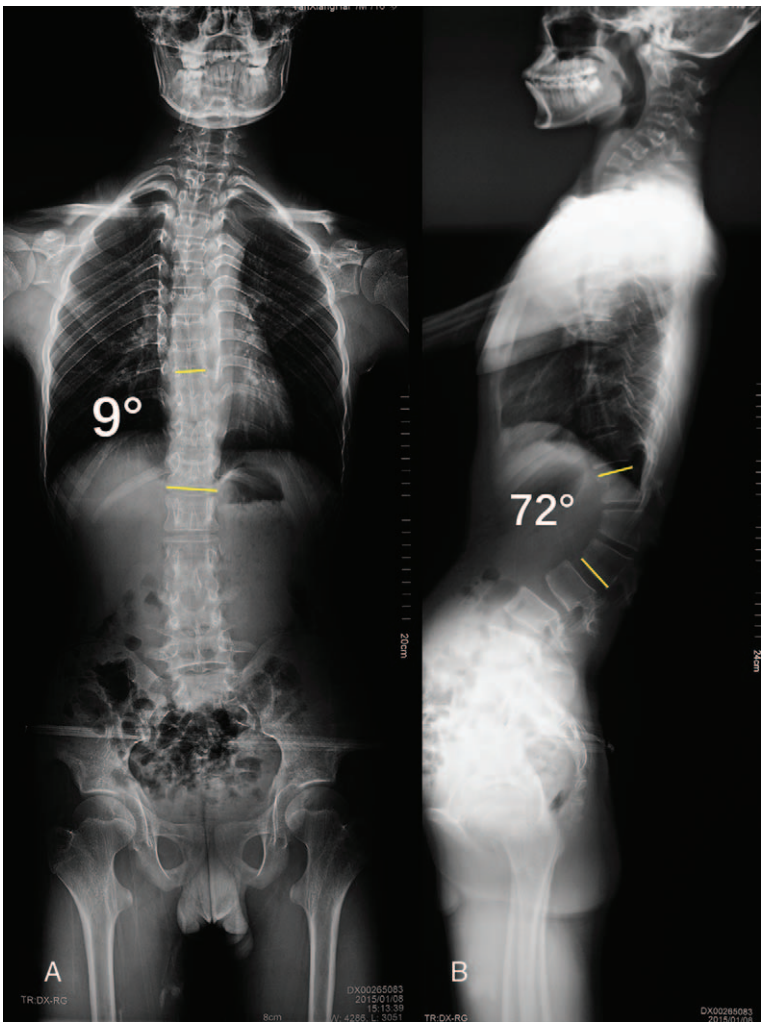

Figure 1. Upright radiographs of the whole spine of the 16-year-old proband, (A) anteroposterior radiograph showing a Cobb angle of 9° at T9 to T12. (B) sagittal radiograph showing obvious kyphosis and a Cobb angle of 73° at L1 to L3.

signs. Full spine anteroposterior and lateral (F-S-AP and LAT) x-ray examinations revealed an L1 to L3 "wedging" shape of the vertebrae and 73° localized kyphotic angulation in the lumber region. The Cobb angle of T9 to T12 on the coronal plane was 9° (Fig. 1). The patient was diagnosed with Scheuermann disease after excluding congenital kyphosis due to no hemivertebrae, incomplete segmentation, butterfly vertebrae, or other deformities present on the x-ray and computed tomography scan. In addition, traumatic kyphosis was excluded due to no history of trauma.

The proband's family history was obtained by routine inquiring. The results showed that there was a family history of scoliosis. The proband's 57-year-old father (II-3) and sister (III-5) were also confirmed to have Scheuermann disease. The proband's father showed similar characteristics to the proband in the general examination; the F-S-AP and LAT x-ray showed a "wedging" shape of the vertebrae at L1 to L3 and intervertebral space narrowing at L2 to L3 with localized kyphotic angulation of 70°, and a coronal Cobb angle of 20° at L1 to L5; there was slight displacement and instability of the lumbar vertebra; and there was a diagnosis of degenerative scoliosis (Fig. 2). The proband's 19-year-old sister did not show kyphotic deformity, pain, or dysfunction of the lower back; in addition, there were no positive neurological signs. Radiographs of the F-S-AP and LAT x-ray showed a "wedging" shape of the vertebrae at L1 to L3, a localized kyphotic angulation of 37°, and a Cobb angle of 15° at T9 to T12, with a diagnosis of IS (Fig. 3).

The proband's 51-year-old mother had a thoracic scoliotic deformity for 40 years, without back pain or lumbar dysfunction. She had no treatment or disease progression in the last 30 years. Physical examinations showed thoracic vertebral curvature on the left kyphotic deformity of the thoracic region, a positive Adam forward-bending test with even shoulders, same hip and leg length, negative neurological signs, and no cafe-au-lait macules spots. Radiographs showed scoliosis of the thoracic vertebrae with a "C" shaped curve (Fig. 4). Magnetic resonance imaging did not show any manifestations of syringomyelia. She was diagnosed with IS because she refused further examinations.

Although the aunt of the proband had lower back kyphosis and the grandmother of the proband had thoracic scoliosis (Fig. 5), they failed to receive a diagnosis and treatment for a variety of reasons.

The proband accepted osteotomy and orthopedic surgery for kyphosis at T10-L4, followed by pedicle screw fixation and bone graft surgery, after general anesthesia. After 3 months of treatment, postoperative lateral radiographs showed a significantly improved sagittal vertical axis (SVA) (Fig. 6).

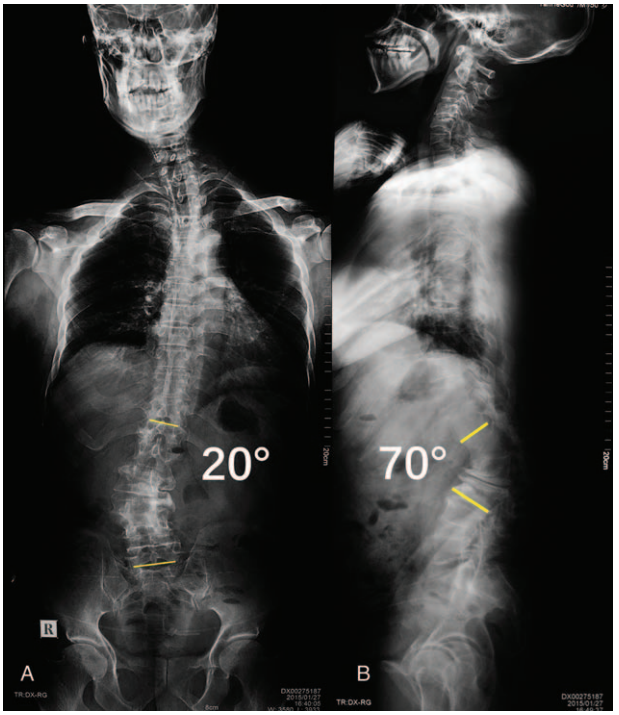

Figure 2. Upright radiographs of the whole spine of the proband's father. (A) Anteroposterior radiograph showing a Cobb angle of 20° at L1 to L5. (B) Sagittal radiograph showing a Cobb angle of 70° at L1 to L3 and with intervertebral space narrowing at L2 to L3.

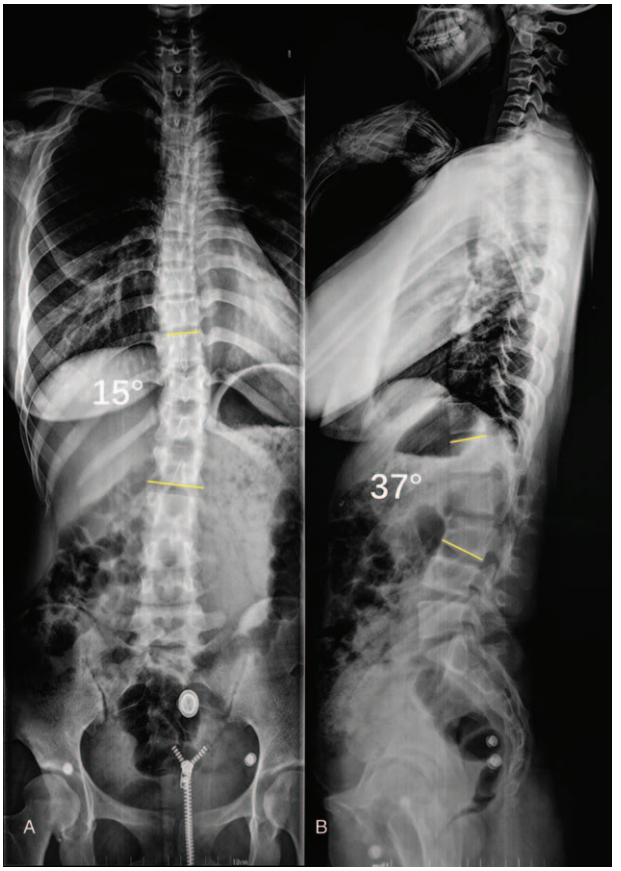

Figure 3. Upright radiographs of the whole spine of the proband's older sister. (A) Anteroposterior radiograph showing a Cobb angle of 15° at T10-L1. (B) Sagittal radiograph showing a Cobb angle of 37° at L1 to L3.

3. Discussion

In this case report, we present a Chinese patient with serial lumbar Scheuermann disease with IS; this proband was successfully treated by surgery. In this pedigree, the mother of the proband (II-4) was diagnosed as having adolescent idiopathic scoliosis (AIS), and the grandmother (I-4) of the proband showed the same signs as the mother according to oral descriptions without medical records. The paternal aunt (II-5) and the paternal uncle (II-7) of the proband showed no obvious signs of scoliosis. The characteristics of the pedigree described in our report fit the criteria of an allosomal inheritance pattern.

The inheritance patterns of both diseases presented in this case were consistent with known reports. Interestingly, the proband's sister (III-5) was diagnosed with both Scheuermann disease and AIS. It remains unclear if this is a pure coincidence or patients with AIS or Scheuermann disease have the same genetic mutations. It has been reported that patients with AIS or Scheuermann disease have a family history of these diseases, but these patients were not confirmed to be diagnosed with both Sheuermann' disease and AIS.[26]

A high relevance in the incidence of Scheuermann disease has been demonstrated among monozygotic twins.[17] Similarly, it has been reported that the onset of Scheuermann disease may be inherited.[16,18,25] In 2001, an autosomal dominant mode of inheritance of Scheuermann disease was confirmed by segregation analysis for 90 pedigrees from Barnaul; in addition, a succession of IS and Scheuermann disease was found in consecutive generations. Moreover, a classic case of Scheuermann disease in male monozygotic twins has been reported supporting the theory of a genetic contribution in classic

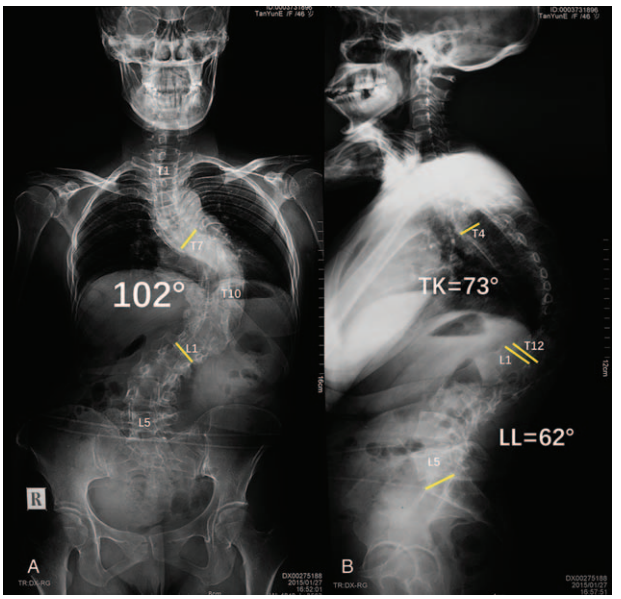

Figure 4. Upright radiographs of the whole spine of the proband's mother. (A) Anteroposterior radiograph showing a Cobb angle of 102° at T7-L1. (B) Sagittal radiograph showing thoracic kyphosis (TK) of 73° and a lumbar lordosis (LL) of 62°.

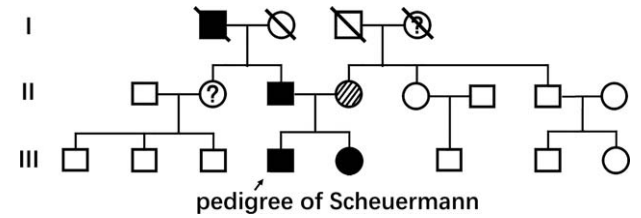

Figure 5. Pedigree chart of the patient. III-4 represents the proband. I-1 represents the grandfather of the proband. The family members said that the grandfather of the proband had similar signs as the patient and the patient's father. However, this could not be further traced due to his death. I-4 represents the grandmother of the proband, who had a hunchback from a young age, and the specific diagnosis was not clear. II-2 represents the paternal aunt of the proband, who had no confirmed diagnosis due to lack of imaging data. II-4 represents the mother of the proband, who was diagnosed with scoliosis according to her medical history; she refused further examinations of magnetic resonance imaging and computed tomography.

Scheuermann disease. Furthermore, it has been reported that the overall prevalence of Scheuermann disease was 2.8% and that the male-to-female ratio was approximately 2:1 in a cohort study among twins including nearly 35,000 individuals.[3] However, to date, all the reports refer to the classic thoracic or thoracolumbar segments, and few cases have been reported for the lumbar region.[2,27] In the current report, with a confirmed diagnosis for the proband, proband's father, and proband's older sister, it can be speculated that there is a genetic etiology in the onset of Scheuermanndisease.

No confirmed independent pathogenic factor for IS has been identified. It has been reported that both AIS and neuromuscular scoliosis share similar pathogenic characteristics.[28] AIS is considered to be genetically heterogeneous, and there are different inheritance patterns in different pedigrees.[29] Some pedigrees demonstrate a multi-gene inheritance pattern, while others show an autosomal dominant inheritance pattern. It is believed that the cause of IS is multifactorial, including genetic predisposing factors as well as metabolic, hormonal, and biomechanical factors.[30–32]

The pathogenic mechanisms of Scheuermann disease as well as AIS are unclear. It is worth following up and studying the onset and development of both diseases among the offspring of II3 and II4, especially regarding cartilage differentiation and formation. The DNA samples from every individual in this pedigree were reserved for further studies.

4. Conclusion

Although it can happen, Scheuermann disease seldom occurs in the lumbar region or with IS. However, it can be treated by an appropriate orthopedic surgery. The series of lumbar Scheuermann patients with IS in the pedigree presented herein also support the genetic etiology of Scheuermann disease. The family history of patients with hyperkyphosis should be carefully assessed to exclude the possibility of Scheuermann disease in clinical practice.

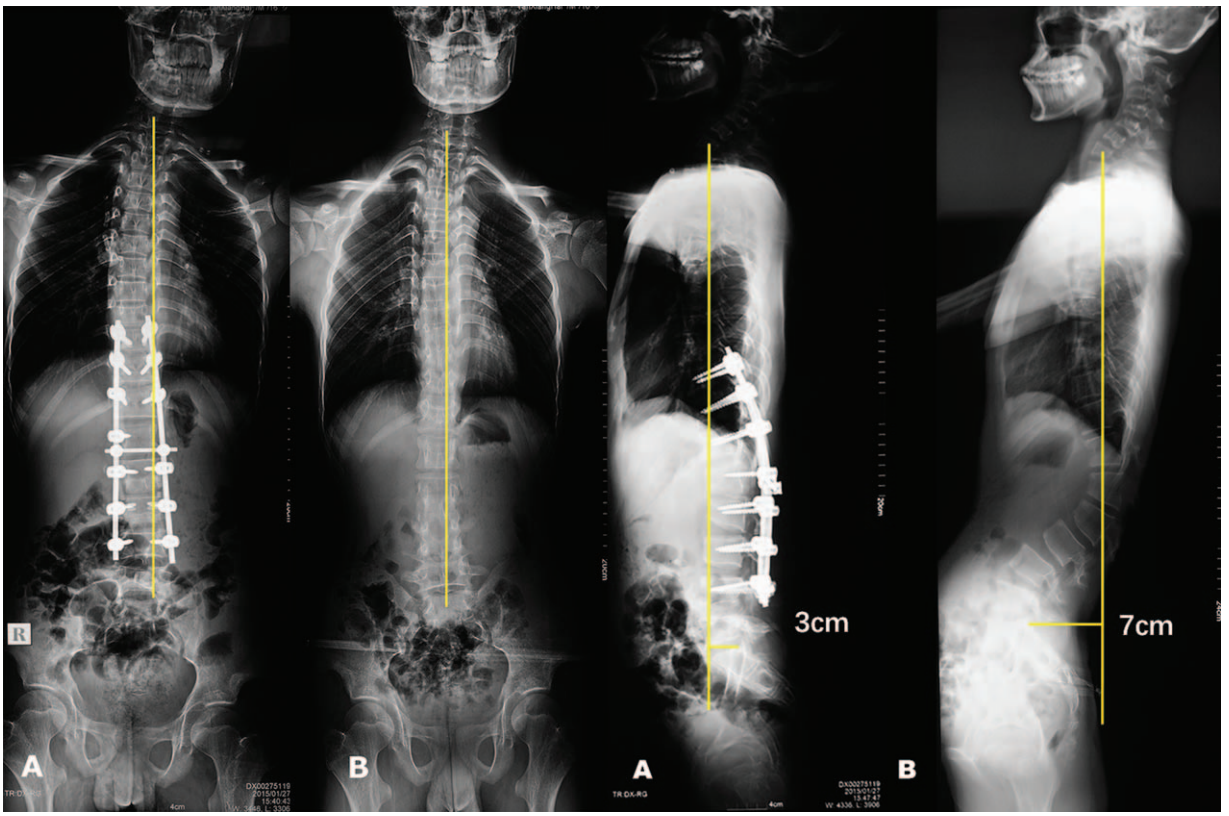

Figure 6. Preoperative and postoperative lateral radiographs of the proband were compared. (A) Postoperative anteroposterior and lateral radiographs; (B) preoperative anteroposterior and lateral radiographs.

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