

SURGICAL TREATMENT FOR A PATIENT WITH MARFAN SYNDROME BY POSTERIOR STABILIZATION: A CASE REPORT

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Abstract

Scoliosis in Marfan syndrome is a common occurrence, constituting up to 63% prevalence. Correction is not always easy as the structural anatomy differs from the general population. We present a female 23 years old with chief complain of curved back since 10 years before admission. The patient had high stature and positive Marfan syndrome's criteria. The patient also complained of shortness of breath. The curvature was progressive becoming more severe over time. In radiographs we found that the main thoracic curve Cobb's angle of 92° and main lumbar curve Cobb's angle 76°. We performed scoliosis correction with pedicle screws and rod by posterior approach. Intra-operative blood loss was 1900 cc. Postoperatively, the patient had weakness at the lower limbs but resolved after several days and gained full motoric function after one week. Severe scoliosis is an indication for surgery to correct the spinal deformity that causes several complications such as pulmonary and cardiac complications. The result was satisfying proving that the procedure is safe and with proper peri-operative preparation, we may achieve a good functional outcome.

Keywords: Marfan syndrome, Scoliosis, Surgery

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Introduction

Marfan syndrome is a common connective tissue disorder compromising around 0.01% of the general population. The disorder is due to dominant mutation of the fibrillin-1 gene located on chromosome 15. Traditionally, diagnosis is made using the Ghent criteria although newer revised criteria has been proposed.⁽¹⁾

Spinal deformity is one of the most dreaded clinical features in Marfan syndrome that often needs correction. Although more patients will not require correction, the clinical consequences of patients with spinal deformities are often severe and troublesome. Spinal deformities in Marfan syndrome include progressive scoliosis and thoracic lordosis with concomitant loss of lumbar lordosis, and more rarely, thoracolumbar kyphosis, severe spondylolisthesis and cervical problems. Marfan syndrome is often associated with scoliosis, with two thirds of cases having the aforementioned deformity. Approximately one quarter to one half of those patients have curves severe enough to consider surgical correction after nonoperative approach. Scoliosis in Marfan syndrome has a higher prevalence of double thoracic curves and triple major curve.⁽²⁻³⁾ Almost one half of patients have kyphosis up to 50°. ⁽⁴⁾

Newer methods and instrumentation have improved the outcome of scoliosis treatment, including for patients with Marfan syndrome. The aim for treatment for scoliosis among patients with Marfan syndrome is to achieve spinal correction and good general functional outcome. As bracing is often ineffective in treating the scoliosis associated with Marfan syndrome, surgery has been considered as the current definitive treatment among patients with scoliosis more than 40°. ⁽⁵⁾ We present a case

of Marfan syndrome with a severe scoliosis corrected using the surgical posterior approach.

Case Report

Clinical condition

We present a woman 23 years old with curved back since ten years previously. At first the curvature was not severe, and the patient did not seek medical attention. Five years later, the curve worsened, and the patient sought medical attention. The patient went to an orthopaedic clinic and was suggested to undergo the operation. The family refused the operation due to financial problems. The patient then decided to take out health insurance first. The patient returned three years later with complaint of shortness of breath, back pain and fatigue. The patient could not stand for long periods. No history of neurological deficit, defecation and urinal problem was present. The patient had high stature, positive Steinberg sign and Walker-Murdoch sign. Forward flexion was 0-90°, extension was 0-25°, right lateral bending was 0-40° and left lateral bending was 0-40°. Body height of the patient was 164 cm with sitting height of 74 cm, plumb line was to the right side around 3 cm and rib hump was 5 cm (**Figure 1**). No shoulder tilt or pelvic tilt was evident, and the body-arm distance was 3 cm. From the neurological examination, no neurological deficit was present. The X-ray of the patient is shown in **Figure 2**, and radiologic parameter is shown in **Table 1**. From the physical examination, we concluded the patient to have a diagnosis of neurogenic scoliosis due to Marfan syndrome. Before the operation, we consulted a cardiologist and pulmonologist for echocardiography and spirometry and evaluated the tolerance for operation. This was the step for peri-operative preparation.



Figure 1. From the clinical picture, a curved back was present with right thoracic curve and left lumbar curve, with rib hump, no step off and no tenderness.



Figure 2. X-ray of the patient, from the left, lateral erect, AP erect, right lateral bending, left lateral bending

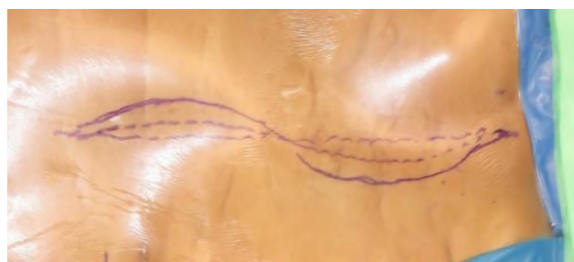
Table 1. Radiological scoliosis profile measurement of the patient

	Cobb angle	UEV	LEV	Apex
Proximal thoracic	15°	Thoracal 2	Thoracal 4	Thoracal 3
Main thoracic	92°	Thoracal 5	Thoracal 11	Thoracal 7
Lumbal	76°	Thoracal 12	Lumbar 4	Lumbar 2
Riisse	R5			

*UEV: upper end vortex, LEV: lower end vortex



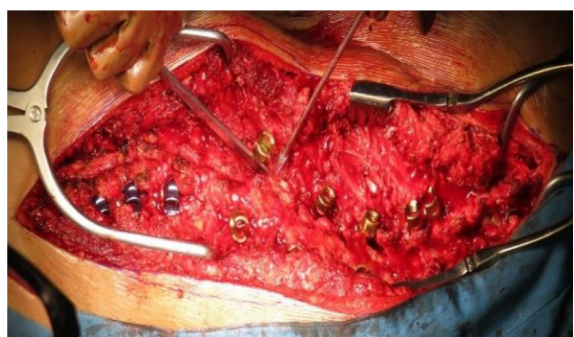
Step 1. Incision design was made. From the picture, we can see the scoliosis curve (a dash-dotted line), midline (inside dots), and incision landmark (middle dots).



Step 2. After incision layer by layer, the whole spine was exposed. The rotational deformity is clearly observed.

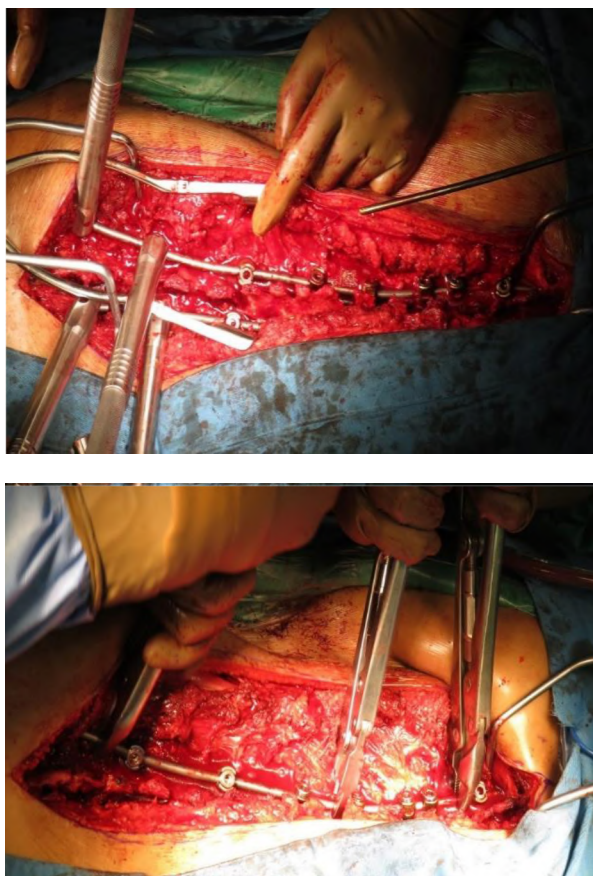


Step 3. Pedicle screws are inserted into Thoracal 3, 5, 8, 10, and 12 (monoaxial), and Lumbar 2, 3 and 4 (polyaxial). The pedicles are around 4.5 to 5.5 mm in diameter.



Step 4. Facetectomy and release of interspinous ligament are done to further free the vertebral body. Knubble was used to harvest bone graft.

Figure 3. Surgical procedure step by step



Step 5. A rod is placed afterwards. Translational (the upper figure) and rotational (the lower figure) correction are done.

Figure 3. Surgical procedure step by step (Next)



Figure 4. From left to right, clinical picture showing the patient had a good functional outcome, and the AP and lateral x-ray postoperatively.

Table 2. Pre-operative and postoperative comparison of Cobb's angle

	Pre-operative	Post-operative	Angle Difference
Proximal thoracic	15 ⁰	3.9 ⁰	11.1 ⁰
Main thoracic	92 ⁰	46.2 ⁰	45.8 ⁰
Lumbal	76 ⁰	32 ⁰	44 ⁰

Surgical technique

We performed scoliosis correction using pedicle screws and rod by posterior approach. The surgical steps for treating the scoliosis are shown in **Figure 3**. Intra-operatively the blood loss was 1900cc, with additional transfusion of 750cc of packed red cell and 500 cc of fresh frozen plasma. After the operation on the third day, more transfusion was given due to a drop in the hemoglobin. Immediately after the operation, patient complained of weakness on both of her legs; however, the weakness improved. On the seventh day, she was able to stand on both of her legs and walk with the help of assisted walking device. The postoperative X-ray is shown in **Figure 4**. The comparison of Cobbs' angle pre- and postoperative is shown in **Table 2**. Three months after the operation the patient was able to walk without assisted device, without complaint of shortness of breath or back pain.

Discussion

Spinal deformity characteristic in Marfan syndrome

The spectrum of spinal deformity is wide ranging from lumbar lordosis, or thoracic kyphosis to severe scoliosis. The classic Marfan syndrome features include vertebral scalloping, higher prevalence of lumbosacral transitional vertebrae, lengthened process distance and a reduction in pedicle width; therefore, causing the mean pedicle width to be smaller than the smallest pedicle screw. Laminar thickness is also smaller than normal controls. In our patient, we tended to use a smaller pedicle screw with 4.5 to 5.5 mm in diameter due to smaller size in the pedicle. ^(2, 4, 6)

Scoliosis is prevalent in Marfan syndrome, reaching a fascinatingly 63% of patients. ⁽⁷⁾ Although similar to cases of idiopathic scoliosis, Marfan syndrome with scoliosis usually

presents with deformities that are more severe and progressive. A higher prevalence of double thoracic curves and triple major curves has been noted among patients with Marfan syndrome. A severe scoliosis case may also cause problem in cardiac and pulmonary systems. The threshold of curve progression is below 40⁰ at the age of maturity, but the Ghent criteria includes Marfan syndrome with a scoliosis curve more than 20^{0.2}. In our patient, the main thoracic curve was 92⁰. The curvature was progressing at a great rate that after ten years, the patient had more than 90⁰ of angle. The patient also complained of shortness of breath, which probably manifested due to her scoliosis. After the correction, the patient gradually diminished the complaint, a few months after follow-up no further shortness of breath was observed, and the motoric function returned to normal with grade 5 in each of the limbs. Unfortunately the lung function was not performed postoperation.

Indication of surgery

Spine surgery is indicated for approximately 10 to 15% of cases. Braces may be indicated for less severe cases, but due to its progressiveness usually surgery is more appropriate. Scoliosis progresses faster among patients with Marfan syndrome than the general population, with mean progressivity at the age of 3 years old and mean progression of 19±17⁰ per year. ⁽⁷⁾ Curves greater than 40⁰ traditionally are noted to be more progressive and will give significant clinical impact for the patient. At this rate, brace treatment was not indicated for the patients. Brace treatment is usually indicated for patients with curvature less than 20⁰ but with the understanding that in the future, a scoliosis correction by surgery may be needed. ⁽¹⁾ Including our patients, we proposed the need for scoliosis correction per surgery so

that we may achieve a more acute correction; thus, correcting the associated deformities and cancel the notorious complications. Although an anterior release and discectomy before the posterior instrumentation may improve correction in severe scoliosis in Marfan syndrome, we performed posterior stabilisation only. This is in line with the study by Silvestre et al. which stated satisfactory stabilisation of scoliosis could be achieved by posterior instrumentation alone among patients with Marfan syndrome.⁽³⁾

In the past, due to the respiratory insufficiency and cardiology problems caused by the syndrome, surgery was not an exciting option, especially scoliosis correction. However, due to improved surgery techniques and supporting systems, this is no longer justifiable; instead, this makes surgery become one of the mainstay treatments of those living with Marfan syndrome to definitively correct the deformity and control the rate of associated complications.⁽³⁾ Based on this optimism, we proceeded to surgery, especially choosing the posterior approach; we further reduced the risk associated with pulmonary deficiency or cardiology; thus, marking the safety of this procedure.

In our study the blood loss was 1900 cc with additional transfusion of 750 cc of packed red cell and 500 cc of fresh frozen plasma. This was higher than the mean reported blood loss in idiopathic scoliosis, with a range of 800-1400 cc.⁽⁸⁻⁹⁾ This was probably due to the higher severity of the scoliosis curvature and length of operation to correct the deformity. However, this was lower than the mean blood loss in a serial study of scoliosis among patients with Marfan syndrome by Jones, which reached 2400 cc.⁽⁵⁾ After the surgery, the thoracic curve achieved 45.8° (~50%) and lumbar curve achieved 44° (~56%) correction. The correction was acute at first causing the patient to have neuropraxia; thus, causing sudden weakness in both the lower extremities. The weakness; however, was temporary and after a few days the patient had fully gained the motoric function. The rate of this surgical complication is unclear in recent literature reports.⁽¹⁰⁾ The correction was also in accordance with the recommendation given

by Jones that the correction achieved should be around 50-60% to avoid curve decompensation.⁽⁵⁾ However, the patient did not complain of numbness or neuropathic pain afterwards.

The need for treatment guidelines for spinal deformity

Overall, spinal deformity correction has shown good results, providing good functional and physiological outcomes for patients. Patients were satisfied and we can hope for a longer life for the patient. Nevertheless, the treatment can be considered late as the patient was 23 years old and already had a curved back for 10 years. Therefore, a guideline consensus for surgery may be needed to improve the overall outcome. However, as the literature for this kind of deformity is still lacking, more research on comparing the clinical outcome of patients treated early or late may be needed first. Guidelines will be needed to help clinicians make decisions on a daily basis

Conclusion

Severe scoliosis with Marfan syndrome could be corrected acutely and safely by surgery using the posterior approach. The procedure is important to improve functional and physiological outcomes for the patient.

CONFLICT OF INTEREST

The authors declare they have no conflict of interest.

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