



Rehabilitation for Marfan Syndrome

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Abstract: Marfan syndrome is a spectrum of disorders caused by a heritable genetic defect of connective tissue that has an autosomal dominant mode of transmission. The defect itself has been isolated to the FBN1 gene on chromosome 15, which codes for the connective tissue protein fibrillin. Abnormalities in this protein cause a myriad of distinct clinical problems, of which the musculoskeletal, cardiac, and ocular system problems predominate. The skeleton of patients with Marfan syndrome typically displays multiple deformities. Mitral valve prolapses that requires valve replacement can occur as well. Given the variable expressivity of Marfan Syndrome, no single sign is pathognomonic; the diagnosis is made on clinical grounds on the basis of typical abnormalities. We reported a boy, 12 years old, referred from surgeon with diagnosis *pectus carinatum pro brace*. Chest protrusion appeared since age 6, getting bigger without any complaint but cosmetic. Other complaints on feet which looked flat, sometimes ankle sore after long distance running or futsal. He was the first child and no family history had a condition like him. His hobby was playing futsal, and daily activities were independent without assistive devices. General appearance and vital signs were normal, cardiorespiratory assessment was normal, BMI on percentile 10-25, arm span to height ratio 1.09, lens subluxation of left eye, lens dislocation of right eye, poor standing balance, inadequate toe off, thoracic hyperkyphotic, positive wrist sign, true leg length discrepancy of 1 cm (left>right), bilateral ankle ROM limitation, rigid flat feet suspected bilateral vertical talus, left hallux valgus, Marfan syndrome score 9, and normal echocardiography. In this patient, we gave semi rigid thoraco-lumbo-sacral orthosis (TLSO) with 3 points pressure system and rigid bar on protution area (custom molded). resistance exercise (F: 3x/week, I: moderate fatigue, Borg scale 13-15/20, T: 8-15 reps/set, 2-3 set/ session, T: major muscle group upper and lower extremities aerobic exercise (F: 3x/week, I: moderate to vigorous, borg scale 13-15/20, T: ≥ 60 min/session, @5-10 min warming up and cooling down (stretching), T: sport (swimming, running, cycling). The patient was referred to a surgeon for a brace. In conclusion, this case report highlights the multidisciplinary management of patients with Marfan syndrome.

Keywords: Marfan syndrome; typical abnormalities; multiple deformities

INTRODUCTION

Marfan syndrome is a spectrum of disorders caused by a heritable genetic defect of connective tissue that has an autosomal dominant mode of transmission. It is a rare condition, occurring about 1 in 5000 people. The defect itself has been isolated to the FBN1 gene on chromosome 15, which codes for the connective tissue protein fibrillin. Abnormalities in this protein cause a myriad of distinct clinical problems, of which the musculoskeletal, cardiac, and ocular system problems predominate. The skeleton of patients with Marfan Syndrome typically displays multiple deformities including arachnodactyly (ie, abnormally long and thin digits), dolichostenomelia (ie, long limbs relative to trunk length), *pectus* deformities, and thoracolumbar scoliosis.^{1,2}

There are no specific cures to treat Marfan Syndrome. The majority of medical therapy related to Marfan syndrome has been targeted at preventing cardiovascular compromise with beta blockers and afterload-reducing agents to reduce stress on the aortic and mitral valves and the aortic root.³

CASE REPORT

We reported a male patient, age 12 with suspected Marfan syndrome due to *pectus carinatum* as the chief complaint. Through comprehensive evaluation, patient had many problems so that he was managed multidisciplinary with orthopedist, ophthalmologist, and pediatric cardiologist, however, this report will focus more on discussing the management of the given rehabilitation. The rehabilitation management provided in this case report aimed to address patient complaints related to chest protrusion and leg deformities.

In history of present illness, the patient's mother reported that his breast bone became more prominent since he was 6 years. At first it was a lump on the chest sized of a chicken egg, but it grew further and started to bother cosmetically. He never complained of shortness of breath or other complaints related to the heart and lungs, but the protrusion of the bone was felt to be disturbing so he decided to seek treatment. Also, he had complaint about flat feet especially left side, and sometimes felt sore at ankle after futsal, long-distance walking, or running. He went to a surgeon due to protrusion on the chest and he was advised to get brace from a physiatrist. At school, he was able to follow the lessons well and never missed class. He was also able to participate in sports activities without any complaints, in fact he regularly participated in futsal extra-curricular activities once a week.

Physical examinations were, as follows: General status *compos mentis*, general condition looked good. Vital signs were within normal limits. Cough ability was adequate, and single breath count test was 34. Chest expansion 4-4-5 cm. Anthropometry status: body weight: 39 kilograms, body height: 159 cm, BMI: 15.6 kg/cm²; arm span: 174 cm. CDC: WFA percentile 50; HFA percentile 90; BMI for age percentile 10-25.

Related to general systems, thorax inspection revealed *pectus carinatum*, but other examinations were within normal limit.

Mobility showed that supine to side lying: independent; side lying to sitting: independent; sitting to standing: independent; walking and running: indoor and outdoor independent, without assisted devices; stairs and hopping: independent. Gait assessment: asymmetric stride and step length, shorter on left leg (LLD 1cm left <right), asymmetric step/ base width, toeing out right >left, asymmetric arm swing, Trendelenburg gait (+)/pelvic drop on left side, lack of toe off (inadequate pre swing).

Status localis of upper extremities: Look: no deformity, arm span 174 cm (arm span to height ratio = 1.094); Feel: no tenderness, no mass, no inflammatory sign, normal muscle tone, normal sensory; Move: full ROM all joints without pain, no spasticity, muscle strength functional (MMT 5/5); physiological reflex ++/++, pathological reflex (-), wrist sign (+). Status localis of lower extremities: Look: no genu deformity, bilateral flat feet (medial arch (-)), bilateral ankle pronation (+), left hallux valgus (+), overlapping toe and 2nd left digit, AHI n/t. Feel: no tenderness, no mass, no inflammatory sign, normal muscle tone, normal sensory,



Figure 1. Body posture



Figure 2. Patient's hand



Figure 4. Patient's feet

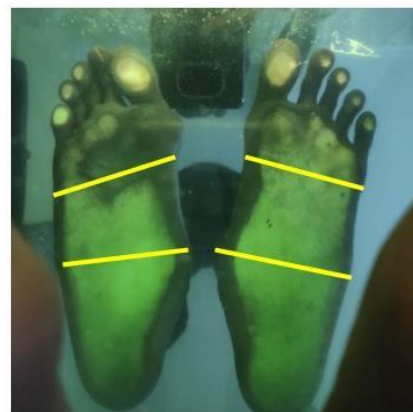


Figure 5. Patient's footprints



Figure 6. Patient's chest

Pedis Xray AP-Lateral:

Institution	RSUP PROF DR RD KAN MANADO
Other Patient ID	
My Comment(s)	
Study Comments	Pectus Caviartum dan FLAT

Foto Pedis sinistra AP/Oblique/Lateral :

- Longitudinal arch pedis sinistra tampak mendatar
- Tidak tampak fraktur dan destruksi tulang
- Mineralisasi tulang baik
- Celah sendi yang tervisualisasi baik
- Jaringan lunak sekitar kesan baik

=====[Conclusion]====
Flat foot sinistra

Vertical Talus

Figure 7. Radiology finding of patient's foot

TLL 97 cm/96 cm, ALL 95 cm/ 94 cm, LLD 1 cm, bony protusion on median plantar left foot, painless. Move: full ROM all joints without pain exclude on ankle, no laxity, no spasticity, muscle strength functional (MMT 5/5). Right ankle: dorsoflexion 5-10°, plantarflexion 40°. Left

ankle: dorsoflexion (-), plantarflexion 30°. Physiological reflex ++/++, negative pathological reflex. Jack Toe raise test (-) patient could not do toe-raise (rigid flatfoot), CSI >45%.

As our case resume, a 12-year-old boy, referred from a surgeon with diagnosis of *pectus carinatum* pro brace. The chest protusion appeared since age 6, and getting bigger without any complaint but cosmetic. Other complaints on feet, looked flat, ankle sore after long distance running or playing futsal. General appearance and vital signs were normal, cardiorespiratory assessment normal, BMI on percentile 10-25, arm span to height ratio 1.09, standing balance poor and inadequate toe off, thoracic hyperkyphotic, wrist sign (+), true leg length discrepancy 1 cm (left>right), bilateral ankle ROM limitation, rigid flat feet susp vertical talus bilateral, left hallux valgus, Marfan syndrome score 9, echocardiography normal. The clinical diagnosis was Marfan syndrome. The functional diagnosis was *pectus carinatum*, thoracic hyperkyphotic, limitation of ankle ROM (dorsi-plantar flexion bilateral), rigid flat feet suspected as vertical talus, and hallux valgus sinistra, True LLD 1 cm, Lens subluxation OS and dislocation OD. Rehabilitation problems were *pectus carinatum*, bilateral rigid flat feet, hallux valgus sinistra, true LLD 1 cm, psychological, blurred vision ec lens subluxation/dislocation of right and left eyes.

The goals of therapy were short term: working diagnosis was established and patient/caregiver was well educated, psychological problem was corrected, *pectus carinatum* was improved, feet deformities were corrected. Long term: *Pectus carinatum* was corrected, recurrency prevention, maintenance of good cardiorespiratory function (physical performance) and quality of life, and early detection and adequate management for complications of Marfan syndrome.

The prognosis was, as follows: *ad vitam: dubia ad bonam*. Marfan syndrome cannot be cured but therapy is more aimed at overcoming existing problems and complications. *Ad sanationam: malam*. Marfan syndrome cannot be cured because the etiology is in the form of a genetic disorder, so the current management is to overcome the problems that arise. *Ad functionam: pectus carinatum: dubia ad bonam*, because *pectus carinatum* can be treated conservatively with a brace (patients and caregivers refused surgery) and at a child's age (immature skeletal bone) the prognosis is quite good with regular use. Feet deformities: *malam*, because the deformity that occurs was an anatomical problem where the main management recommended is surgery. However, the patient and caregiver refused surgery, so the prognosis was *malam*, only management for ankle fatigue and improvement of gait could be performed.

DISCUSSION

Marfan syndrome is an autosomal dominant heritable disorder of fibrillin-1 (FBN1) with predominantly ocular, cardiovascular, and musculoskeletal manifestations that has a population prevalence of approximately 1 in 5–10,000. There is no apparent enrichment in any ethnic or racial group. Males and females are affected with equal frequency. The protein encoding fibrillin-1, known as the FBN1 gene, is located on chromosome 15.1 Fibrillin-1 is the main component of elastic matrix microfibrils, which have a role in the connective tissue of the cardiovascular and musculoskeletal systems^{1,2}

The musculoskeletal manifestations of MFS, which to date have received less attention, can also have a significant impact on the quality of life and are likely to become more important as the age of the MFS population increases. Aortic root dilatation and ectopia lentis are the two cardinal clinical features of MFS, but on their own, they are insufficient to confirm the diagnosis. Rather, diagnosis is made using a combination of features stratified on the presence or absence of a family history of MFS.⁴

Radiography, family history, and genetic testing are used in the diagnosis of MFS. It is important for orthopaedic surgeons to recognize the signs of MFS; however, a diagnosis should come from a geneticist. Genetic specialists are aware of the indications and implications of genetic testing and of the differential diagnoses. Prompt diagnosis and treatment are essential to optimize outcomes. For those diagnosed as MFS, their care will involve routine echo-monitoring (usually yearly) and a review of skeletal manifestations and symptoms over many decades.⁵

An overlooked but important effect of the musculoskeletal complications of MFS is their impact on daily functioning and quality of life. In Speed's study in 2017 involving 245 MFS participants, 89% reported pain, most commonly beginning in the back (50.6%). Of these, only 46.6% were satisfied with their current pain management.⁴

Symptoms associated with pain included stiffness, difficulty walking, muscle spasms, and muscle weakness, and there was a significant association between pain and the presence of kyphosis, degenerative disc disease, osteoarthritis, and dural ectasia. The diagnosis of a chronic and potentially life-threatening disease such as MFS comes with its own psychological burden that varies widely depending on personal circumstance, particularly when the manifestations and severity of the condition vary widely.¹¹

For parents of children with MFS, there are a number of practical issues that they find hard to manage and for which the schools are often ill-equipped to deal with (due to lack of knowledge of the disease). Focus on academic work may be affected by upper limb fatigue when writing and poor visual acuity (when there is lens involvement).¹²

The differential diagnoses of this case were Loeys-Dietz syndrome (LDS), an autosomal dominant condition that includes many features of Marfan syndrome and Shprintzen-Goldberg syndrome (SGS): The phenotype of SGS is distinctive but shows significant overlap with LDS and Marfan syndrome. Major distinctions include the unique and highly penetrant developmental disability in SGS, with less frequent and milder cardiovascular manifestations than in either Marfan syndrome or LDS.¹³

Scoliosis may require bracing or surgical stabilization; repair of *pectus* deformity is largely cosmetic. Functional deficits or pain associated with protusio acetabulae may respond to physical therapy, analgesics, or anti-inflammatory medications. Orthotics and arch supports can lessen leg fatigue, joint pain, and muscle cramps associated with pes planus.

There are no known effective therapies for symptomatic dural ectasia. Medications that reduce hemodynamic stress on the aortic wall, such as beta blockers (β blockers) or angiotensin receptor blockers (ARBs), are routinely prescribed. Therapy is generally initiated at the time of diagnosis with Marfan Syndrome at any age or upon appreciation of progressive aortic root dilatation even in the absence of a definitive diagnosis.^{2,14}

Pectus carinatum is characterized by an anterior protrusion of the sternum and the adjacent costal cartilages. There are two subtypes: The chondro-gladiolar subtype is most common (92–95% of cases), and involves a protrusion of the body of the sternum (gladiolus).¹⁴

In children, *pectus carinatum* is the second most common chest wall deformity, affecting males about four times more frequently than females. Compressive orthotic bracing is a time sensitive therapy. A chest wall becomes more rigid (less compliant) with age, and the optimal age for bracing is between 10 and 15 years. Compressive orthotic bracing is the first line therapy for most children with a compliant *pectus carinatum*, but may be less effective in patients with a non-compliant chest wall or asymmetry.⁸ Bracing may also be effective as part of a hybrid procedure in older patients with a mature skeleton. These patients can be treated with a minimally invasive procedure to excise around 70% of the abnormal cartilage in a flat ridge using with a small oscillating saw.⁸ Surgery is usually not needed and only offered if bracing does not result in the desired correction or if the child has completed puberty and the skeleton is too stiff to respond to bracing. Surgical repair (Ravitch procedure) is done through a horizontal chest incision across the mid chest. In this repair, the abnormal costal cartilages are removed, preserving the lining that covers the outside of the cartilage, allowing the sternum to be pushed downward in a more normal position. In chest expansion, the child breathes in as deeply as he/she can, pulling shoulders back while taking in a breath. The child should then hold his/her breath for as long as possible.⁸

Physiotherapists also assess endurance, speed, fatigability, pain and ability to walk on different terrains, with a focus on assessing function, not just structural abnormalities. Foot exercises to strengthen intrinsic foot muscles have been suggested to be helpful in the management of pes planus. It has been reported that these exercises can change plantar pressure distribution and

thereby relieve the pain. Specific exercises include: walking up on the metatarsal heads ('tip-toes'), walking on the heels, activities to improve the dynamic arch such as walking barefoot on soft sand, flexing the toes (eg picking up a tissue with the toes), rolling a ball under the arch of the foot while sitting, pretend piano playing with the toes, great toe dorsiflexion, and encouraging climbing and other gross motor activities. Insoles act as a temporary relief for flat feet.^{11,15}

The goal is not usually to permanently reverse the changes in the foot and ankle, but rather to help limit progression of the deformity and reduce the rate of chronic, secondary complications up the kinetic chain. Life expectancy has increased from 47 years to 75 years.^{11,15}

CONCLUSION

We reported a 12-year-old boy suspected Marfan syndrome, known as *pectus carinatum*. The prominent breast bone on his chest began to appear at age six and had grown since then. He also complained about flat feet and ankle sores after futsal, long-distance walking, or running. The patient was advised to get a brace from a physiatrist. Physical examinations showed good general condition, vital signs were within normal limits, and thorax inspection revealed *pectus carinatum*. His mobility was independent, and he was able to follow lessons and participate in sports activities without complaints. Moreover, his lower extremities showed no deformity, bilateral flat feet, and a bony protrusion on the median plantar left foot. The patient was referred to a surgeon for a brace. This case report highlights the multidisciplinary management of patients with Marfan syndrome.

Conflict of Interest

The authors affirm no conflict of interest in this study/

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