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Work adjustment in patients with Marfan syndrome: A case report

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A B S T R A C T

Introduction: Marfan syndrome (MFS) is a connective tissue disorder with multisystem involvement, including cardiovascular and ocular complications, which can impact a patient's work capacity. While MFS may necessitate certain job restrictions, complete exclusion from professional roles may not be justified. Adjustments and careful planning can enable individuals with MFS to maintain meaningful employment within safe parameters.

Case Report: This case examines a 29-year-old male patient diagnosed with MFS, presenting with ocular issues and significant cardiovascular involvement, including aortic insufficiency and a history of valve replacement. Given his condition, ongoing anticoagulant therapy with warfarin is required, alongside annual cardiac assessments. Employment restrictions recommended for this patient include avoiding heavy manual labor, working at heights, operating hazardous tools, confined workspaces, and solitary work environments. Additionally, prescription glasses are mandatory to address his ocular complications, with regular follow-up by an ophthalmologist. Following a sleep study, no limitations were indicated for night shift work, permitting him flexibility in shift scheduling.

Conclusion: This case highlights that while patients with MFS can remain active in professional roles, individualized work modifications are essential. Work-related precautions, especially concerning physically demanding tasks and hazardous environments, should align with the patient's medical condition and treatment. Personalized occupational health assessments can enhance safety and facilitate work participation for individuals with MFS.

Keywords: Marfan Syndrome, Occupational Health, Occupational Diseases

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Introduction

Marfan syndrome (MFS) is an autosomal dominant disorder that was first identified in 1896 by a French pediatrician named Antoine Marfan (1). MFS appears to be age-related and hereditary, with significant intra- and inter-familial variation. Pathogenetic changes in FBN1, which encodes fibrillin-1, are the cause of MFS (2). De novo variants can be present in up to 25% of MFS. The most notable MFS symptoms include asymptomatic aortic root aneurysms, aortic dissections, ocular lens dislocations (ectopia lentis), and skeletal abnormalities that are characterized by long bone overgrowth (1, 4). The Ghent II nosology is used to diagnose MFS; genetic testing to confirm the existence of an FBN1 pathogenetic variant is not always necessary for diagnosis but can aid in differentiating MFS from other heritable thoracic aortic disease syndromes that can present with skeletal features resembling those observed in MFS (2, 3).

Complementary and supportive treatments for MFS aim to manage symptoms, reduce complications, and enhance quality of life, given the syndrome's wide-ranging effects on connective tissue, particularly impacting the cardiovascular, ocular, and skeletal systems (5). Cardiovascular care is crucial, as many MFS patients face risks associated with aortic aneurysms or valve issues (6). Beta-blockers or angiotensin receptor blockers (ARBs) are commonly prescribed to help control blood pressure and reduce stress on the aorta, potentially slowing disease progression (6). Regular monitoring with echocardiography is essential to track cardiovascular health, and in some cases, surgery, such as aortic root replacement, may be required (6). For skeletal concerns, including scoliosis or joint laxity, physical therapy can improve posture, flexibility, and core strength, helping to reduce discomfort and support joint stability (7). Orthopedic bracing may also be considered to manage scoliosis in children and adolescents (8). Ophthalmologic care is equally important, as MFS often affects the eyes; prescription glasses or contact lenses help address lens dislocation and other common visual disturbances (9). In severe cases, corrective surgery may be necessary to improve or stabilize vision (9). Psychological and social support, including counseling or support groups, can address the emotional impact of chronic disease and improve coping strategies (10). Nutritional guidance, emphasizing heart-healthy foods, further supports overall wellness, as does education on avoiding high-impact or contact sports to minimize the risk of injuries (8). Together, these complementary treatments provide a holistic approach, allowing patients to manage MFS's complex challenges while maintaining their quality of life (10).

Although this disease with a prevalence of 1 in 5000 people (4) can affect various organ systems and cause symptoms such as ectopia lentis in the eye, scoliosis, long bone overgrowth, arachnodactyly, and pectus deformity, it should not cause these patients to be completely excluded from the work cycle (2). In this study, an examination of a young man with MFS was conducted to gain a deeper understanding of the symptoms and complications associated with the disease and to highlight the job limitations and workplace adaptations necessary for individuals with this condition.

Case Report

The patient, a 29-year-old man working as a hospital caretaker, visited the occupational medicine clinic for annual examinations. He was a tall man with elongated hands and a thin face with visible misalignment in his teeth. In physical examinations, his height was measured to be abnormally tall, due especially to the abnormal length of the extremities (figure 1 – A). His facial features included a long, narrow face and a high, arched palate. Clinical tests for the evaluation of hyperextensiveness included the wrist (Walker-Murdoch) sign (Figure 1 - B) and the thumb (Steinberg) sign (Figure 1 - C), which were both positive.

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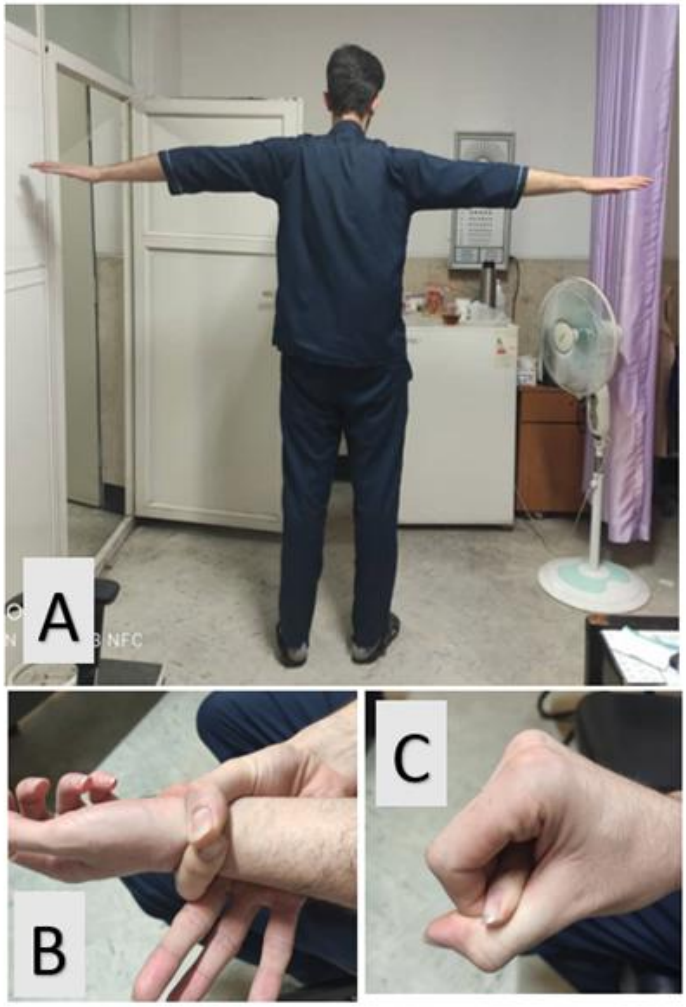


Figure 1. Clinical features of patients with MFS. Abnormal length of the extremities (A). Clinical tests for the evaluation of hyper extensibility included the wrist (Walker-Murdoch) sign (B) and the thumb (Steinberg) sign (C).

A cardiac examination revealed a midline scar on the sternal, which was caused by an aorta valve replacement surgery. A metallic valve sound was audible through the stethoscope. Blood pressure readings were identical in both arms, and the pulse was symmetrical in the arms and feet. In his past medical history, he mentioned that because of progressive exertional dyspnea, he underwent a cardiovascular examination that showed moderate AI. The patient had undergone valve replacement surgery (mechanical leaflet AV prosthesis) in 2016 and is still undergoing treatment with warfarin, losartan, and propranolol. The patient's 3D echocardiography results before and after open heart surgery are shown in Table 1. The abnormally migrating band detected in the patient provided the genetic evidence for the presence of MFS. His history indicated that he was exempted from military service due to severe visual acuity depletion.

Ethical consideration

Ethical issues including adherence to established ethical standards, ensuring honesty in literature collection and data reporting, obtaining written informed consent from the participant in accordance with the Declaration of Helsinki guidelines, and following principles for conducting interventions involving human subjects were considered.

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Table 1. The patient's three-dimension echocardiography result before and after open heart surgery.

Befor open heart surgery	After open heart surgery
Mild LV[1] enlargement with preserved systolic function (EF[19]:50-55%) No resting RWMA[2] No LVH[3] Normal LV[1] diastolic function Normal RV[4] size with normal systolic function Prolaptic PMVL[5] with mild MR[6], no MS[20] Bicuspid AV[7] (mediolateral oriented) with mild to moderate highly eccentric jet AR[8], no AS[9], dilated aortic root and STJ[10] (annuloaortic ectasia), annulus:3.2 cm, sinus of Valsalva: 4.2 cm, proximal part of ascending aorta: 4.7cm, distal part: 3.5 cm Mild TR[11] (TRG[12]: 20 mmHg), no TS[13] Trivial PI[14], no PS[15] No PH[16] (PASP[17]: 25 mmHg) No pericardial effusion	Normal LV[1] size (regarding to BSA[18]) with preserved systolic function (EF[19]:50%) No resting RWMA[2] No LVH[3] Normal LV[1] diastolic function Normal RV[4] size with normal systolic function Prolaptic PMVL[5] with mild MR[6], no MS[20] Mechanical bileaflet AV[7] prosthesis with normal leaflet motion and acceptable hemodynamic study (PPG[21]: 28 mmHg, MG[22]:18 mmHg, DVI[23]:0.4), patent aortic dacron tube graft with no leakage from distal part. Mild TR[11] (TRG[12]: 20 mmHg), no TS[13] Trivial PI[14], no PS[15] No PH[16] (PASP[17]: 25 mmHg) No pericardial effusion
[1] LV: Left Ventricle, [2] RWMA: Regional Wall Motion Abnormalities, [3] LVH: Left Ventricle Hypertrophy, [4] RV: Right Ventricle, [5] PMVL: Posterior Mitral Valve Leaflet, [6] MR: Mitral Valve, [7] AV: Aortic Valve, [8] AR: Aortic Regurgitation, [9] AS: Aortic Stenosis, [10] STJ: Sino Tubular Junction, [11] TR: Tricuspid Regurgitation, [12] TRG: Tricuspid Regurgitation Gradient, [13] TS: Tricuspid Stenosis, [14] PI: Pulmonary Insufficiency, [15] PS: Pulmonary Stenosis, [16] PH: Pulmonary Hypertension,[17] PASP: Pulmonary Artery Systolic Pressure, 18 BSA: Body Surface Area, [19] EF: Ejection Fraction, [20] MS: Mitral Stenosis, [21] PPG: Peak Pressure Gradient, [22] MG: Mean Transvalvular Pressure Gradient, [23] DVI: Doppler Velocity Index	

Genetic counseling was done, and a pedigree was drawn. He is the third child, and his older sister and mother's appearances are both Marfanoid, without cardiac complications (Figure 2). A genetic study was considered in the mentioned case, and we do whole exome sequencing by Illumina Nextseq 550 to cover all genes and exomes related to Marfan and Marfanoid diseases (Figure 3). A pathogenetic variant in the FBN1 gene was found (NM_000138.4: c.493C>T, p. Arg165Ter). We did Sanger sequencing to approve this variant in the proband and his family. A heterozygous condition was shown in the result of sequencing in his mother and his sister with Marfan disease, and wild type was reported in the rest of his family without manifestation of any sign or symptoms of MFS. The patient's current position as a hospital caretaker involves tasks that could potentially pose risks considering his Marfan syndrome. Given his use of anticoagulants, aortic valve replacement, and poor vision, specific job tasks may need to be evaluated and adjusted to ensure his safety. Hospital caretakers may be required to engage in manual labor, work with sharp tools, or navigate confined spaces, all of which could increase their risk of injury or strain. The final decision is conditional. Fit: The patient's role as a hospital caregiver could be suitable if certain modifications are made to accommodate his health condition. This includes:

•Avoiding heavy manual labor or tasks that involve lifting heavy objects.

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•Steering clear of working at heights or in confined spaces where physical exertion or accidents could lead to serious complications (e.g., bleeding due to anticoagulant use).

Minimizing exposure to situations where eye injuries could occur, such as cleaning tasks involving splashing liquids or working with hazardous chemicals.

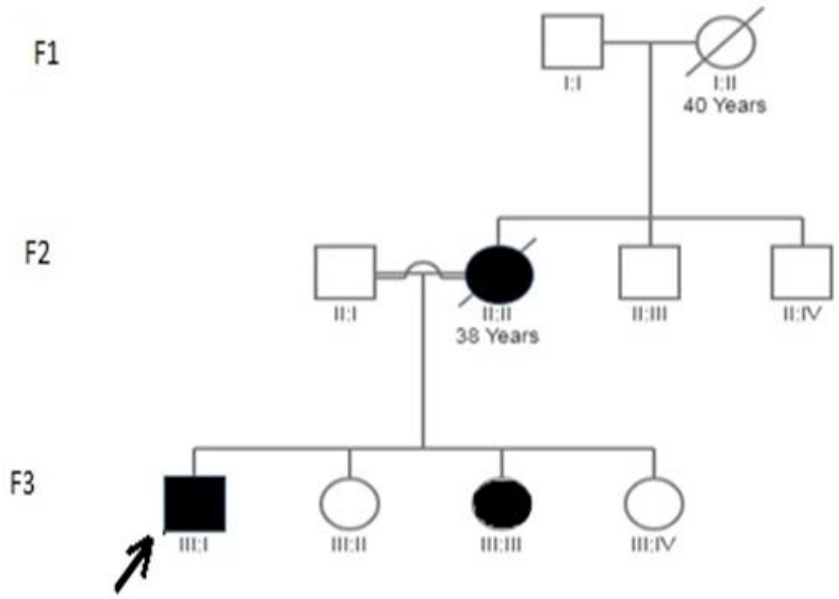


Figure 2. pedigree of patient with MFS.

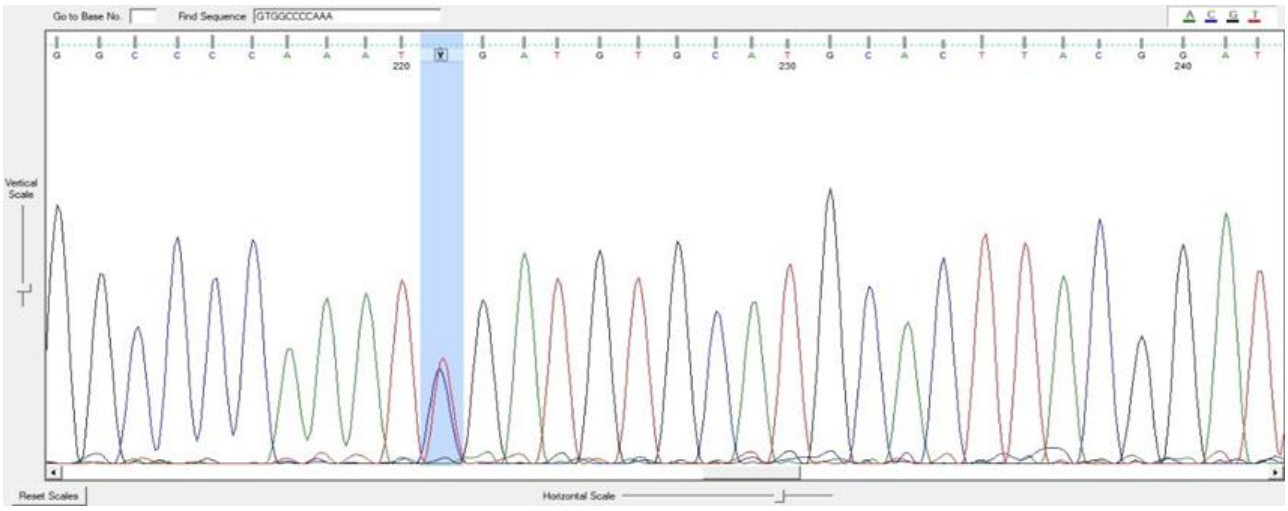


Figure 3. Detection of Heterozygous c.493C>T Mutation in MFS Proband.

Discussion

This patient presented with mild to moderately high eccentric jet aortic regurgitation (AR), a typical complication of MFS. While the full diagnostic criteria for MFS were not met, the patient's family history, genetic testing, and aortic root replacement surgery supported the diagnosis, consistent with a GenTAC registry score of 3 points (5). Aortic disease in MFS significantly impacts the safety of certain occupational tasks. The risk of aortic dissection or worsening aortic insufficiency necessitates avoiding physically strenuous jobs, particularly those involving heavy lifting, prolonged isometric

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exercises, or activities that could significantly elevate blood pressure and heart rate (10, 11). For this patient, tasks requiring significant physical exertion should be avoided, as they could increase the stress on the aorta (11). The patient is also receiving warfarin therapy, which increases the risk of bleeding. Occupational adjustments must account for this, avoiding jobs where there is a risk of injury or bruising, such as roles that involve working with sharp tools, heavy machinery, or at heights. These patients should be placed in environments where the risk of trauma is minimal, and personal protective equipment should be used as necessary (11). Severe visual impairment, common in MFS due to complications such as ectopia lentis and retinal detachment, limits the patient's ability to work in environments where sharp vision is crucial, such as jobs involving exposure to physical, chemical, or biological hazards (9). Jobs like welding, hazardous manual work, or healthcare roles that carry a risk of eye injury should be avoided unless appropriate ocular protective devices are used (11). MFS patients are also prone to developing obstructive sleep apnea, which can affect their suitability for shift work or jobs requiring high levels of attention, such as driving (11). It is recommended that diagnostic measures, such as polysomnography, be conducted to evaluate sleep quality and determine if adjustments are needed for roles involving nighttime shifts or long hours of vigilance (11). Patients with MFS would need to keep the intensity of workouts and physical movement within the low to moderate metabolic equivalent of extended tasks. Competitive and contact sports can put individuals with MFS at an expanded chance of damage. According to these explanations, the recommendations are provided in Table 2 (7, 8).

Table 2. Occupational Medicine Recommendations for Patients with Marfan Syndrome.

1	Avoid physically strenuous jobs or intense isometric activities
	Intense physical exertion can increase heart rate, blood pressure, and stress on the aorta, which could elevate the risk of aortic dissection. Jobs requiring heavy lifting or sustained physical strain should be avoided.
2	Avoid roles with a high risk of injury or trauma
	Since patients on warfarin therapy have an increased bleeding risk, they should avoid jobs involving sharp tools, heavy machinery, working at heights, or confined spaces where injury could occur
3	Limit exposure to environments with potential for eye injury
	Visual impairments such as lens dislocation or retinal detachment require avoidance of roles in high-risk environments, such as welding or work involving exposure to hazardous materials. Ocular protective equipment is recommended if such roles are necessary.
4	Consider positions with low to moderate physical activity
	Tasks that require low levels of exertion, such as office work or managerial roles, may be more suitable for individuals with Marfan syndrome to reduce stress on their cardiovascular system.
5	Evaluate for obstructive sleep apnea
	Jobs that require alertness, such as driving or night shifts, may require additional diagnostic testing to assess sleep quality and ensure that the patient can safely perform these tasks.
6	Avoid occupations with rapid changes in atmospheric pressure
	Marfan syndrome patients are at risk of lung collapse in situations with rapid atmospheric pressure changes, such as scuba diving or flying in unpressurized aircraft. These types of jobs should be avoided.

Conclusion

In this study, a 29-year-old man with Marfan syndrome (MFS) involving ocular and cardiac complications, including aortic insufficiency and valve replacement, was examined. Given the cardiac involvement and history of valve replacement,

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ongoing treatment with warfarin and annual echocardiographic evaluations under a cardiologist's supervision were recommended. Heavy manual labor, working at heights, handling dangerous or sharp tools, working in confined spaces, and solitary work environments were advised against. The use of prescription glasses and regular ophthalmologic follow-up for eye health management were also required. Based on sleep test results, no limitations were identified for night shift work. In general, patients with MFS should not be excluded from professional roles; however, individualized work modifications are essential to reduce the risks associated with their condition and medications. Occupational health and safety guidelines should be carefully adapted to align with the patient's medical status, with particular emphasis on avoiding physically demanding tasks and environments where there is an elevated risk of injury.

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Conflict of interest

The authors declare that no conflict of interest exists.

Authors' contributions

Conceptualization, writing– Review & Editing: NK, SS, Methodology: NK, LK, Validation, Resources: SS, LK, Formal Analysis: LK, NK, Investigation: NK, SL, Data Curation: SL, SN, writing– Original Draft Preparation: LK, SN, Visualization: LK, SL, SN, Supervision, Project Administration: NK, Funding Acquisition: No Funding.

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