



Department of Rehabilitation Services
Occupational Therapy

ICD.9 Code: 759.82

Standard of Care: Marfan Syndrome

Case Type / Diagnosis:

Marfan Syndrome is an autosomal dominant disorder of connective tissue with manifestations variably involving the cardiovascular, ocular, musculoskeletal, and other systems. It affects approximately 1 in 5000 people. Involvement of the cardiovascular system, particularly aortic dilatation and dissection, makes Marfan syndrome one of the most highly lethal genetic conditions.¹ Many of the physical features common to Marfan syndrome such as tall stature, scoliosis, deformities of the chest wall, and mitral valve prolapse are also relatively common in the general population. Advances in medical treatment have greatly extended life expectancy. The best hope for living into one's 70s or even 80s lies with attention to recommended medical care and changes to lifestyle. Lifestyle changes are mostly related to limitations in physical activity, both at work and during recreation.² Diagnosis of Marfan syndrome is based on the presence of at least two of four characteristic features: family history, and ocular, cardiovascular, and skeletal manifestations.³

Clinical Manifestations:

Ocular system: Ectopia lentis (lens dislocation), high myopia, and retinal detachment are features that may be found in patients with Marfan syndrome.¹

Cardiovascular system: Abnormalities of the cardiovascular system are the leading cause of early and/or sudden death in Marfan syndrome. Aortic aneurysm and dissection, mitral valve prolapse and regurgitation are most common (found in 50%-80% of patients).¹ All Marfan syndrome patients are considered at increased risk of aortic dissection. Intense isometric exertion, such as lifting free weights, induces a Valsalva maneuver and increases wall stress. This can lead to weakening of the aortic media, particularly if aortic dilatation is present.⁴

Integumentary system: Skin hyperextensibility has been found in about two-thirds of patients with Marfan syndrome, and striae distensae (stretch marks) are common in this population. Hyperextensibility of integument can also lead to inguinal, diaphragmatic, and umbilical hernias.³

Musculoskeletal system: People with Marfan syndrome are typically taller than their unaffected siblings and have long digits and extremities. They may present with abnormalities of anterior chest (pectus excavatum, pectus carinatum) and of the spine, including spondylolisthesis,² scoliosis and kyphosis.¹ Because of the high prevalence and aggressive progression of scoliosis in addition to unusual patterns of kyphosis, it is not uncommon for the patient with Marfan

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syndrome to require surgery. However, a high rate of complications following spinal surgery has been noted to occur in patients with Marfan syndrome.⁵

Joint laxity being generally the problem for these patients, occupational therapy is consulted mostly for stabilization splints, adaptation for activities of daily living (ADL), instrumental activities of daily living (IADL) and joint protection issues. However musculoskeletal manifestations can frequently include acetabular protrusion at the hip, and elbow flexion contractures.²

Pulmonary system: Spontaneous pneumothorax and apical blebs may occur.² Restrictive lung disease may also be present in setting of scoliosis.

Variations in joint mobility: Generalized hypermobility is a prominent feature of hereditary connective tissue disorders, such as in Marfan syndrome, where marked laxity and dislocations of the patella, hip, shoulder and ankle are common.⁶ Joint laxity may lead to frequent sprains. The fingers, wrists, elbows and knees are commonly hyperextensible and ligament laxity could result in problems such as (pes planus/flat foot).⁷ Positive thumb sign and wrist sign are often present. A positive thumb sign is when the entire thumb nail projects beyond the ulnar border of the hand when the hand is clenched without assistance. A positive wrist sign has the thumb overlapping the terminal phalanx of the fifth digit when grasping the contralateral wrist.¹ At its most extreme, this deformity with subsequent weight bearing over the medial aspects of the feet and ankle cause abnormal stance with secondary stresses on the other joints of the leg.⁷ Conversely, patients with Marfan syndrome may also present with congenital contractures especially at the elbows.⁸

Marfan patients at times would have features of a congenital condition known as Congenital Contractural Arachnodactily or CCA. This condition is characterized by Marfan-like appearance “crumpled” ears and congenital contracture of major joints.

Occupational Therapy referral process at Brigham and Women’s Hospital:

Referrals are typically generated from the physical therapist working with the patient. The patients will already have been seen in the Brigham and Women’s Comprehensive Marfan (and related disorders) Clinic, which is a collaboration between teams from the adult genetics clinic and the cardiovascular genetics center. The cardiologists comment on the appropriate activity level for each patient. Patients may also be referred from physicians in rheumatology, cardiology, or by their primary care physician. Patients are referred to Rehabilitation Services for evaluation and treatment of various musculoskeletal issues.

Indications for Occupational Therapy Treatment:

1. Pain
2. Joint hypermobility/strains and sprains.

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3. Impaired range of motion of hand and upper extremities.
4. Splint and adaptive equipment needs.
5. Impaired hand/upper extremity function affecting ADL and/or work performance.
6. Decreased knowledge of appropriate pacing strategies with ADL and IADL as well as activity modification.

Contraindications / Precautions for Treatment: ¹

Contraindications:

- Avoid isometric exercises/strength testing and any activities that would involve Valsalva maneuver due to the increase stress on the heart.
- **See Patient/Family Education and Exercise Guidelines on page 7 & 8.**

Precautions:

- Minimize activities that involve sudden stops and rapid changes in position.
- Avoid contact type sports.
- Use of *beta-blockers*. Beta-blockers are used in the treatment of high blood pressure (hypertension). Some beta-blockers are also used to relieve angina (chest pain) and in heart attack patients to help prevent additional heart attacks. Beta-blockers are also used to correct irregular heartbeat, prevent migraine headaches, and treat tremors. Beta-blockers work by affecting the response to some nerve impulses in certain parts of the body. As a result, they decrease the heart's need for blood and oxygen by reducing its workload. They also help the heart to beat more regularly. ⁹

Commonly used brand names in the US include: Betapace (sotalol), Blocadren (timolol), Brevibloc (esmolol), Cartrol (carteolol), Coreg (carvedilol), Corgard (nadolol), Inderal (propranolol), Inderal-LA (propranolol), Kerlone (betaxolol), Levatol (penbutolol), Lopressor (metoprolol), Normodyne (labetalol), Sectral (acebutolol), Tenormin (atenolol), Toprol-XL (metoprolol), Trandate (labetalol), Visken (pindolol), Zebeta (bisoprololor), Calan, Isoptin (verapamil).

During exercise, keep the pulse rate under 100 beats/min. If not on beta-blockers, keep the pulse under 110 beats/min or per recommendations of the cardiologist.

Examination:

Medical History: Review PMH, pertinent diagnostic tests, imaging and workup, Rheumatology and/or Cardiology's notes in longitudinal medical record (LMR). Inquire about any past occupational therapy and or physical therapy interventions.

History of Present Illness: Chief complaint or mechanism of injury, date of injury or duration of symptoms, treatment to date, reason for referral, prior level of function,

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previous therapy treatment, past or current use of orthotics, and any exercise program. Also inquire about patient's own goals.

Social History: Family/social support, employment, physical activity level, hobbies, sports, ADLs and any pertinent functional limitations. Assess for any psychosocial needs.

Medications: Refer to LMR and/or Outpatient Health Screen. Inquire specifically about beta-blockers. (See previous page for commonly used brand names).

Examination:

- Pain: Described by patient on VAS pain scale. Note location, description and aggravating/limiting factors.
- Posture: May present with thoracic kyphosis and/or scoliosis. Assess for positioning needs.
- Muscle length: Assess overall muscle length tension balance. Assess joint play, end feel of muscle stretch and stability of ligaments to hold joint stable against stress.
- Palpation: To assist in determining source of pain or tissue condition.
- ROM: Select areas to test based on patient's history and involved area. Document active and passive range given that the patient may have issue with both joint flexibility (inert tissues) and muscle ability to work efficiently.
- Strength: Manual resistance and isometric grading are contraindicated. Observe active movement against gravity, for a maximum grade of 3/5. Assess as well for quality of movement and coordination.
- Function: Bed mobility, functional transfers, self-care, ADL and IADL performance including child-care and work. Use of a functional assessment measure such as The Quick DASH or the Functional Independence Measure (FIM).
- Vital Signs: Obtain parameters from physician or use Rehab Services Department parameters for surgical and non-surgical patient with cardiac disease
 - Heart rate: lower end of range: 50bpm; higher end of range: 100-110bpm
 - ¹Systolic blood pressure: 90-150
 - O₂ Saturation: > 90%
 - Respiration rate: < 30 at rest.

Evaluation / Assessment:

Establish Diagnosis and Need for Skilled Services.

Problem List: See indications for treatment section.

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Prognosis: Variable as outcomes will be affected by age, prior function, comorbidities, history of injuries, social support and personal insight and motivation.

Goals:

1. Decrease pain and increase independence with symptom management.
2. Demonstrate independence with joint protection techniques, use of orthotics/braces.
3. Increase ROM and improve self-correction of posture.
4. Increase knowledge of energy conservation techniques, work simplification, and pacing. Increase awareness of available assistive devices that may be of help. Increase activity tolerance and knowledge of safe exercises/sports activity levels; increase knowledge of self-monitoring of vitals and rating of perceived exertion (RPE).¹⁰ *See annex.*
5. Increase independence in performance of ADL and IADL including work.
6. Independence with HEP and activity modification/progression.

Age Specific Considerations:

Age specific considerations in this population include the normal physiological changes that occur with aging, compounded with any existing comorbidities.

Treatment Planning / Interventions

Established Pathway ___ **X** No

Established Protocol ___ **X** No

Interventions most commonly used for this case type/diagnosis:

- Postural training: sitting, standing and sleeping positions. May benefit from positioning supports such as pillows, wedges, rolls, corsets and braces as comfort measures.
- Stretching: correct any muscle length imbalances periscapular and pectoralis due to the chest issues and elbow muscles due to the potential for elbow contractures
- ROM: select joints and movements based on patient's history and limitations found during evaluation. ROM exercises should be performed against gravity as tolerated in order to maintain functional strength.
- Performance of functional activities designed to increase strength, and joint stability (avoid Valsalva maneuver).

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- Patient education regarding exercise guidelines, general conditioning, activity selection and modification. Review ADL and IADL for level of exertion and need for pacing and self monitoring. Heart rate will approximate $RPE \times 10 + 20$ to 30 beats/min.¹⁰

During exercise, keep the heart rate under 100 beats/min. If not on beta-blockers, keep the heart rate under 110 beats/min.¹ As documented previously, MD recommendation is of prime importance for patient-specific parameters to guide treatment.
- Use splints and or wraps to support and stabilize lax joints when performing activities. These splints are used to support/stabilize a proximal lax joint while allowing increased function distally. Examples of such splints would be: Static wrist splints, CMC thumb splints, buddy straps, digit positioning splints as well as wrist and thumb wraps. An appropriately applied splint designed specifically to supplement prescribed exercises may augment an active exercise or ADL program. Allowing greater function than would otherwise take place, this kind of splint often supports weaker muscles, stabilizes adjacent (usually proximal) joints, particularly the wrist, maximizes tendon amplitude at a given joint, or uses or negates a tenodesis effect.¹¹
- Night positioning splints as well as mobilization splints may be used to address the problem of elbow contracture which affects patients with Marfan syndrome. After careful assessment of the musculoskeletal as well as the neurovascular presentation of the joint, splints can be used to minimize incidence of and or correct some elbow contracture problems. If the contracted elbow has a soft end feel, a corrective approach should be taken. If not, steps should be taken to maintain available range for maximum function.

However as for all elbow treatment, strong manipulation should be avoided and splinting should not irritate the superficially located ulnar nerve. It is important to keep in mind that elbows do not respond well to aggressive or forceful therapy. A slow continuous force that stretches the damaged contracted tissue must be distinguished from the sudden, quick maneuver (manipulation) that tears tissue and may evoke a response that results in further contracture. Prolonged gentle splinting improves elbow range of motion through remodeling of contracted soft tissue structures.¹¹

Due to the fact that Marfan patients are of a tall and unusually long stature, prefabricated splints likely will not fit them properly. Consequently, splints and/or braces may need to be customized.

- Given the emphasis on cardiac involvement with Marfan patients, throughout their treatment and especially after cardiac surgery, the occupational therapist's program should include discussions regarding home and work adaptations, activity

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analysis/task modification for upgrading and downgrading activity level, use of adaptive equipment, pacing/energy conservation, as well as lifestyle changes. Equipment and adaptation recommendations may vary from medicine bottle cap changes to relocating a bedroom downstairs and use of higher toilets with arms.

Frequency & Duration: Varies based on evaluation findings. Patients will benefit from a home exercise program (HEP) with clear instruction, and follow up visit(s) may be indicated for re-evaluation and exercise progression.

Patient / Family Education and Exercise Guidelines:

These activities should be avoided by patients with clinically diagnosed genetic cardiovascular disease, such as Marfan's syndrome ⁴

- **“Burst” exertion (or sprinting)**, characterized by rapid acceleration and deceleration over short distances. Exercise of this type is encountered in a variety of sports, such as **basketball, soccer and tennis**.
- Extremely adverse environmental conditions, which may be associated with alterations in blood volume, electrolytes, and state of hydration and thereby increase risk, such as greatly elevated or particularly cold temperatures disproportionate to that which the athlete is accustomed to in temperate climates, high humidity, or substantial altitude.
- **Snow shoveling** has repeatedly been associated with increased cardiovascular events, ^{12 13} most likely because it can elicit higher rate-pressure products than does treadmill exercise testing, ¹⁴ because it is often performed out of necessity by unfit individuals, and because some cardiac patients develop angina at lower rate-pressure products, suggesting a coronary vasoconstrictor response during exercise in cold temperatures. ¹⁵
- Activities that risk rapid changes in atmospheric pressure (such as **scuba diving, flying in unpressurized aircraft**) ¹
- Exercise programs (even if recreational in nature) that require systematic and progressive levels of exertion and are focused on achieving progressively higher levels of conditioning and excellence, such as **cycling, running and rowing**. These individuals are also advised against systematic training during which they are extending beyond the physical limits imposed by their underlying disease and the average aerobic state expected at their age.

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- Excessive participation in sporting activities that otherwise would be regarded as recreational if performed in moderation, eg, **downhill skiing** continuously over an entire day versus more limited and selective skiing over the same time period.
- Intense static (isometric) exertion, such as **lifting free weights**, may prove to be adverse by inducing a Valsalva maneuver or by increasing wall stress and weakening of the aortic media. Other activities that may involve isometric work are **climbing steep inclines, gymnastics, pull-ups**.
- Extreme sports (such as **hang gliding and bungee jumping**) are activities that are best avoided because they require the expenditure of particularly substantial physical energy and incur psychological demands that are exceedingly unpredictable, placing individuals with genetic cardiovascular diseases in compromised circumstances in which the likelihood of injury is substantial and the possibility of rescue from a traumatic or cardiovascular event is greatly reduced.
- Activities involving **high emotional stress** (increased heart rate and blood pressure)
- **Strenuous aerobic pace** or working/exercising at a level where one is pushing beyond his or her capability to breathe normally and use oxygen efficiently.

Recommendations and referrals to other providers: Return to referring MD, especially if cardiac issues are suspected. If chest pain of cardiac origin is suspected and emergent, refer to emergency room or call emergency services.

Re-evaluation / assessment

Standard Time Frame: Re-evaluate every 30 days, or at every follow up visit if OT appointments are scheduled more than 30 days apart.

Other Possible Triggers: Change or worsening of symptoms, failure to respond to treatment, or onset of cardiac chest pain.

Discharge Planning

Commonly expected outcomes at discharge: Patient will have met goals with focus on self-management of symptoms, and exercise and activity modification and progression.

Transfer of Care (if applicable): If no improvement or progress towards goals, return to referring MD for further medical management.

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Patient's discharge instructions: Exercise guidelines, education re activity modification, HEP, postural correction, self-management of symptoms and splint care. Follow up with referring MD as needed.

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ANNEX

Borg Rate of Perceived Exertion Scale (RPE)^{10 16}

6 No exertion at all

7

Extremely light (7.5)

8

9 Very light

10

11 Fairly light

12

13 Somewhat hard

14

15 Hard (heavy)

16

17 Very hard

18

19 Extremely hard

20 Maximal exertion

Interpretation of scale:

9 on the scale corresponds to “very light” exercise. For a healthy person, it is like walking slowly at his or her own pace for some minutes.

13 on the scale is “somewhat hard” exercise, but it still feels OK to continue.

17 “very hard” is very strenuous. A healthy person can still go on, but he or she really has to push him- or herself. It feels very heavy, and the person is very tired.

19 on the scale is an extremely strenuous exercise level. For most people this is the most strenuous exercise they have ever experienced.

Borg RPE scale

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