Standard of Care: Marfan Syndrome

ICD.9 Code: 759.82

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. 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 70’s or even 80’s 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. 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.

Clinical Manifestations:

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

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). All Marfan syndrome patients are considered at increased risk of aortal 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.

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.

Musculoskeletal system: People with Marfan syndrome are typically taller than their unaffected siblings and have long digits and extremities. They also may present with abnormalities of the anterior chest (pectus excavatum, pectus carinatum) and of the spine, including spondylolisthesis, scoliosis and kyphosis. 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 syndrome to require surgery. However, a high rate of complications following spinal surgery has been noted to occur in patients with Marfan syndrome.\textsuperscript{5}

A case study by Herzka et al\textsuperscript{6} notes increased frequency of radiographic cervical spine abnormalities in patients with Marfan syndrome, notably increased atlantoaxial translation. The authors suggest that bony and ligamentous abnormalities noted in some patients with Marfan syndrome may place them at increased risk for atlantoaxial rotatory subluxation. The authors described 2 cases in which these patients did not have any radiating pain, paresthesias or weakness, and one case in which the patient experienced transient upper extremity weakness.

Also, an abnormally high prevalence of lumbosacral transition vertebrae, biconcave vertebrae, decreased bone density, and increased intertransverse process distances are present in the Marfan lumbar spine.\textsuperscript{7} These abnormalities may lead to compromise of fixation of pedicle screws or lamina hooks during spinal surgery for correction of kyphosis and scoliosis. Dural ectasia has also been noted in people with Marfan syndrome. This is a ballooning or widening of the dural sac or dural nerve sleeves, potentially resulting in bony erosion of vertebral elements and anterior meningoceles.\textsuperscript{5}

Other musculoskeletal manifestations include acetabular protrusion at the hip, and elbow flexion contractures.\textsuperscript{2}

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

\textbf{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.\textsuperscript{8} Pes planus is also a common manifestation of joint hypermobility. 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.\textsuperscript{9} Conversely, patients with Marfan syndrome may also present with congenital contractures, especially at the elbows.\textsuperscript{10}

\textbf{Chain of referral to Physical Therapy at Brigham and Women’s Hospital}:

Typically, patients with Marfan syndrome will 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 team will comment on the appropriate activity level for each patient. Patients may also be referred from physicians in pain management, orthopedics, cardiology, or by their primary care physician. Patients are referred to Rehabilitation Services for evaluation and treatment of various musculoskeletal issues. Physical Therapy’s role will also be to educate patients regarding exercises guidelines.
with individualized exercise programs based on musculoskeletal pathology and appropriate activity levels established by cardiologist.

Indications for Treatment:

1. Pain
2. Joint hypermobility/strains and sprains
3. Impaired Range of Motion of peripheral joints and spine
4. Impaired Function (decreased endurance and activity tolerance)
5. Poor Posture
6. Weak posterior (scapular, shoulder, trunk extensors) musculature
7. Footwear evaluation and recommendations re commercial vs. custom footwear and/or orthotic
8. Decreased knowledge of appropriate exercises based on activity level established by Cardiology
9. Decreased knowledge of activity modification and/or progression

Contraindications / Precautions for Treatment: ¹

Contraindications:
- avoid isometric exercises/strength testing and any activities that would involve Valsalva maneuver

Precautions:
- minimize activities that involve sudden stops and rapid changes in position
- minimize contact with other players, equipment or ground.
- 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. ¹¹

Commonly used brand names in the US include: Betapace (sotalol), Blocadren (timolol), Brevidloc (esmolol), Cartrol (carteolol), Coreg (carvedilol), Cordard (nadolol), Inderal (propranolol), Inderal LA (propranolol), Kerlone (betaxolol), Levatol (penbutolol), Lopressor (metoprolol), Normodyne (labetolol), 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 betablockers, keep the pulse under 110 beats/min

Standard of Care: Marfan Syndrome

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**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 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, previous PT, 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, ADL’s and any pertinent functional limitations.

**Medications:** Refer to LMR and/or Outpatient Health Screen. Inquire specifically about betablockers. (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. Identify if patient is frequently in a sitting position and observe this. Inquire about sleeping positions including number of pillows under head and/or pillow arrangements used for comfort, elevation or support.
- **Muscle length:** Hip flexors, hamstrings, and gastrocnemius because of these muscles’ influence on posture.
- **Palpation:** To assist in determining source of pain or instability.
- **ROM:** Select areas to test based on patient’s history and involved area; may include spinal and peripheral joints.
- **Strength:** Manual resistance and isometric grading are contraindicated. Observe active movement against gravity, for a maximum grade of 3/5.
- **Function:** Bed mobility, transfers, stairs
- **Gait:** Pattern/deviations, need for assistive devices, appropriateness of current device, safety of movement, footwear
- **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
  - Systolic blood pressure: 90-150
  - O2 Saturation: > 90%
  - Respiration rate: < 30 at rest

**Evaluation / Assessment:**

**Standard of Care: Marfan Syndrome**

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Establish Diagnosis and Need for Skilled Services

**Problem List:** See indications for treatment section

**Prognosis:** Variable as outcomes will be affected by age, prior function, comorbidities, history of injuries, social support

**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 activity tolerance and knowledge of safe exercises/sports activity levels; increase knowledge of self-monitoring vitals and rating of perceived exertion (RPE)  
   See annex.
5. 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. The physical therapist will consider all of the patient’s impairments whether they are disease or age based and will determine a comprehensive assessment and rehabilitation plan for each patient.

**Treatment Planning / Interventions**

<table>
<thead>
<tr>
<th>Established Pathway</th>
<th>___ Yes, see attached.</th>
<th><em><strong>X</strong></em> No</th>
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</thead>
<tbody>
<tr>
<td>Established Protocol</td>
<td>___ Yes, see attached.</td>
<td><em><strong>X</strong></em> No</td>
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**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 of hamstrings, iliopsoas, rectus femoris, gastroc/soleus, periscapular muscles
- ROM: select joints and movements based on patient’s history and limitations found during evaluation. ROM exercises should be performed against gravity.
• Closed kinetic chain exercises to increase strength, joint stability, proprioception and function, such as stair climbing, single-leg stance, mini-squats (avoid Valsalva maneuver)
• Transfer/gait training with/without assistive device
• Patient education re exercise guidelines, general conditioning, activity selection and modification. Discuss patient’s current activities and activity levels, and simulate those activities in clinic while monitoring vitals and RPE. Also teach patient how to self-monitor heart rate and RPE. Heart rate will approximate RPE X 10 +20 to 30 beats/min. During exercise, keep the heart rate under 100 beats/min. If not on betablockers, keep the heart rate under 110 beats/min.

**Frequency & Duration:** At the therapist’s discretion, based on evaluation findings. Patient will benefit from HEP and exercise 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*

• **“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, most likely because it can elicit higher rate-pressure products than does treadmill exercise testing, 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.

• Activities that risk rapid changes in atmospheric pressure (such as scuba diving, flying in unpressurized aircraft)
• Exercise programs (even if recreational in nature) that require systematic and progressive levels of exertion and are focused on achieving higher levels of conditioning and excellence, such as cycling, running and rowing. These individuals are also advised against systematic training during which they are extended beyond the physical limits imposed by their underlying disease and the average aerobic state expected at that age.

• 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

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 PT appointments are scheduled more than 30 days apart.

Other Possible Triggers: Change in 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.

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

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ANNEX

Borg Rate of Perceived Exertion Scale (RPE)\textsuperscript{17}

6  No exertion at all

7  Extremely light (7.5)

8  Very light

9  Light

10  Somewhat hard

11  Hard (heavy)

12  Very hard

13  Extremely hard

14  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
REFERENCES


17. Department of Health and Human Services, Centers for Disease Control and Prevention. Available at: www.cdc.gov/nccdphp/dnpa/physical/everyone/measuring/perceived_exertion.htm