

Marfan Syndrome *At a Glance*

Marfan syndrome is a genetic disorder of the connective tissue. Connective tissue crisscrosses the body, within muscles and organs, holding them in place like glue, and helping to control how the body grows. In Marfan syndrome, the connective tissue doesn't grow correctly. This condition occurs in males and females equally, in all races, and has a high degree of variability, even within a family. All organs contain connective tissue, so it can affect any part of the body. Typically this condition affects the heart, eyes, lungs, and bones. It does not affect cognitive function. Marfan condition can be life threatening.

About 1 in every 5,000 babies is born with Marfan syndrome.

Learn more about features of Marfan syndrome (NOTE: not all people with Marfan syndrome will have all of these features):

Eyes

- Myopia or nearsightedness - most common eye finding; can progress rapidly during childhood
- Displacement of the lens (ectopia lentis), seen in 60% of individuals with Marfan syndrome, is a hallmark feature
- Increased risk for retinal detachment, glaucoma, and early cataract formation
- Glasses
 - Many eye problems can be managed with glasses

Skeletal system

- Bone overgrowth
 - Tall and thin
 - Extremities (arms and legs) are disproportionately long for size of trunk
 - Overgrowth of ribs
 - Overgrowth of the ribs can either push sternum (breastbone) **in** (pectus excavatum); or push sternum (breastbone) **out** (pectus carinatum)
 - May require surgery to repair the chest wall deformity
 - Long, thin fingers
 - Scoliosis which may be mild to severe and progressive
- Facial features
 - Long narrow face with deeply set eyes
 - Down slanting eyes

- Flat cheekbones
- Small and receding chin
- Dental issues due to high arched palate, and dental crowding
- Joint laxity (flexible joints)
- Flat feet
 - Orthotics and arch supports can lessen leg fatigue and muscle cramps associated with flat feet

Cardiovascular manifestations (~90%)

- These can be severe and may require medications; this needs to be monitored.
- Dilatation of the aorta – an enlargement of the major blood vessel coming from the heart
- Predisposition for aortic tear and rupture
- Mitral valve prolapse, tricuspid valve prolapse (floppy heart valves)
- May need prophylactic antibiotics with dental work

Other features which may occur:

Skin

- Hernias
- Stretch marks that occur without major weight gain or loss

Pulmonary issues (lungs)

- Sudden collapse of the lung (Pneumothorax)
- Asthma
- Sleep apnea
- Pectus excavatum (indented breast bone) and scoliosis may reduce lung capacity
 - Lead to shortness of breath and fatigue

Spine

- Widening or ballooning of the membrane (dural sac) surrounding the spinal cord (dural ectasia)
- Can cause headaches, back/abdominal/leg pain

Headaches/Migraines

- These sometimes occur

Things to Think about in the Classroom

1. Medical / Dietary Needs

What You Need to Know

The list of possible medical problems in Marfan syndrome can be quite extensive. However, each individual usually has only some of these problems. Also, the severity of any one of these medical problems varies widely. Therefore, it is important to ask the parents about the medical issues for their child. School age children who have Marfan syndrome should have a primary care doctor and, often, specialist's visits to monitor medical conditions.

- Students may have chronic pain and require medication as needed
- Students may also require other medication for heart or asthma

What you can do

- Ensure a yearly check up in the child's Medical Home.
- Ensure up to date immunizations.
- Notify parents of changes in energy level.
- Be aware of any changes in behavior or mood or academic performance. Notify the parents.

2. Education Supports

Marfan disease does not affect intelligence. Individuals may have gross or fine motor delay and/or visual difficulties. They may have frequent absences and need help making up schoolwork.

What you need to know

Gross and fine motor delays

- Gross Motor
 - Delays in gross motor development caused by joint hypermobility
- Fine motor
 - It may be hard to hold a pencil because of loose hand ligaments
 - Individuals may have a hard time writing for a long period of time

Vision

- Vision may fluctuate
- It may be hard to read for long periods of time.
 - Individuals may have difficulty reading small or light colored fonts
 - They may have difficulty seeing the chalk board/smart board etc.

What you can do

Gross and fine motor

- PT and orthopedic braces may help
- Allow extra time for assignments and/or tests
- Limit handwriting
- Allow to dictate or verbally take test
- Use a computer or assistive software

Vision

- Large print books
- Materials could be contrasting and clear
- Sit near board
- Tests/homework in large print

3. Behavior and Sensory Support

What you need to know

- Individuals with Marfan syndrome may look “different” than their peers.
 - Taller than average
 - Very skinny and lanky
 - Stretch marks
 - Scoliosis
 - Chest wall deformities
 - Foot problems.
- Feeling as though they look different may affect self-esteem.
- Back braces, and orthotics may further affect self-esteem and confidence.
- Individuals may be unable to participate in many activities, can lead to feelings of isolation
- Individuals with Marfan syndrome may experience pain.
 - May be chronic may interfere with ability to focus or sit for long periods of time

What you can do

- Allow privacy in changing if needed
- Encourage discussions about differences and acceptance of differences within classroom
- Provide explanation of the condition and how it affects individuals
- Encourage participation in activities as appropriate
- Allow access to nurse and medications as needed
- May need to take breaks and/or rests

4. Physical Activity, Trips and Events

What you need to know

Marfan syndrome can cause a variety of problems and each child will be unique with problems and limitations. Most children with Marfan will need a custom designed exercise program. The child's physicians and child's parents should set parameters for exercise. It is important for individuals with Marfan syndrome to have an opportunity for physical activity to optimize physical and mental health.

Learn more about bodily systems:

Musculoskeletal system

- The child may be tall and have long limbs
 - Increased height may lead people to treat older than actual age.
- The child may be very thin and may lack muscle bulk and strength
- Joints may be hyper extensible and prone to dislocating. They may have joint contractures.
- Combination of underdeveloped muscles and joint hypermobility can contribute to poor coordination and delay in acquiring gross and fine more skills
- Scoliosis may limit that range of motion of back
 - May need brace which limits movement even more
- Chest wall deformities (pectus) are usually just cosmetic, but occasionally may affect normal function of lungs
 - They may require surgery or a brace
 - This may cause problems in lifting, exercise capacity, and range of motion
- High narrow and highly arched palate
 - May not be able to use standard mouth guard

Ocular system (eyes)

- May require glasses
- Child may have difficulty with visual perception and with hand eye coordination
- May have difficulty in following the flight of an object
- May have difficulty to tolerating bright light

Cardiovascular System (heart)

- Children with Marfan syndrome will have regular echocardiograms to monitor the size of their aorta.
 - Spontaneous aortic rupture (without a trauma) is rare in school aged children
- Exercise modifications and beta-adrenergic blockers medications are part of management
 - They help by reducing the force with which blood is pumped from the heart thus reducing stress on aorta.
 - Beta blockers may cause-fatigue, feeling sleepy, and reduced ability to concentrate.
 - Repair or replacement of mitral valve may be needed in school age child
 - If child on blood thinners, they may be prone to spontaneous bleeding and easy bruising.

Pulmonary System (lungs)

- The lungs can have weakened areas of tissue that overinflate and can break. This can cause the lungs to collapse.
 - Improper breathing techniques can put further stress on lungs and may lead to lung collapse
 - Lungs may be underdeveloped with reduced capacity for gaseous exchange.
 - Children with this lung problem may have reduced exercise tolerance

What you can do

Exercise modifications

- Reduce the stress on the heart and major blood vessels by limiting activities involving endurance, weight lifting or intense competition
- Minimize opportunity for head and chest trauma
- Reduce stress on joints
- Accommodate possible reduced lung capacity
- Take care with all physical activity
- Exercise to point of exhaustion should be prevented to avoid:
 - Increases in heart rate
 - Increase in blood pressure
 - Force of muscle contractions

- Be aware of those individuals on a blood thinner; a forceful collision may result in a heavy bleed

General

- Encourage academic and artistic activities
- Allow child to help with coaching or team management if there are restrictions
- Consider 504 for physical activity modifications

Explore activities that are of interest to the child AND safe. What accommodations can be made to keep the child safe? Here are some examples of activities that might work for some children who have Marfan syndrome:

K-3

- Movement exploration activities

Grades 4-12

- Archery
- Pool activities
- Board games
- Bowling
- Bicycling
- Croquet
- Dance/rhythms
- Darts
- Golf
- Gymnastics (balance activities)
- Horseshoes
- Relaxation exercises
- Shuffle board
- Walking
- Aquatics/water activities

Learn more about modifications:

There is not a single exercise program that works for each child with Marfan syndrome. A child may need help developing a realistic self-concept of abilities and limitations.

Ideally the child should have a non-competitive isokinetic activity preformed at a non-strenuous aerobic pace where he/she can stop and rest when tired and have no forceful contact with other players, equipment, or ground. There should be a minimal chance of sudden stops and/rapid change in direction.

It is generally recommended that people who have Marfan syndrome stay at an aerobic level of work at 50-60% of their maximal heart rate (if no evidence of aortic dilatation the physician

may permit activity at higher levels of intensity). Physicians may recommend a graded exercise program.

- Examples include; Brisk walking, leisure biking, slow jogging, shooting baskets, slow tennis, use of 1-3 pound weights
- They shouldn't strain and should use low-tension settings on equipment, such as treadmills or exercise bikes
- Individuals must learn to not test limits. This is very hard for children.

Other Modifications:

General

- May be hard to fit into desks, chairs
- Hard to sit comfortable for a long period of time
- Allow to stand
- Equipment: a child may need a brace
- May affect maneuverability, flexibility, speed, and endurance
- Provide privacy in changing to avoid embarrassment
- Check what restrictions with brace may be
- If chest wall surgery occurs, ask what the restrictions are.
- Mouth guards
 - May require special one
- If hernias are present, individuals may need supportive gear and instruction in proper lifting, and minimize lifting and climbing
- Clothing should be supportive and appropriate to avoid increased risk of falling
- If glasses: as with other children with glasses they should be shatterproof and rounded
- If individual has contact lens, these lenses are at increased risk of falling off because of flatter lens
- May need sun glasses for activities outside or in brightly lit places

Cardio

- Encourage children to take part in noncompetitive activities performed at a sub maximal level of effort.
 - Instruct in safe levels of intensity and duration
 - Should be taught never to "push through" symptoms
 - Provide adequate time for gradual warm-up and cool down
 - Monitor level of exertion more closely under extreme weather conditions because this may add additional stress and may affect child's endurance and exertion level

- Many children are on beta blockers which will depress heart rate so heart rate is not indicator of exertion level (those on beta blockers <7 years of age should keep prolonged heart rate under 120/min)
- Older children keep heart rate under 100 beats/min
- Select activities to promote concept of self-competition to minimize the effects of peer pressure for the child to exceed physical exertion limits
- Instruct in self-monitoring technique.
 - Provide opportunities for practice of exercise self-monitor
 - Instruct in relaxation techniques, safety, and breathing
- May need additional time to get to class
 - Limit extra movement between classes
- Extra books and/or a second locker may be needed to accommodate lifting/carrying restrictions.

Musculoskeletal

- Strengthening exercises for both muscles and ligaments
 - Address overall lack of bulk and muscle tone
 - Good for joint hyper extensibility (underdeveloped muscles)
 - Avoid heavy weights and intense isometric exertion
- Pain (chronic)
- Muscle fatigue
- May require physical therapy
- Modifications to improve coordination
 - May have poor eye hand and eye foot coordination
- Provide opportunities to practice visual tracking of objects in motion and develop other sequential perceptual motor skills.
 - Fine motor
 - Gross motor
 - Balance
 - Spatial
 - Body awareness
- May be able to enhance child's body mechanics and improve posture with appropriate exercises.

Modifications for intensity, endurance, and fatigue

- Decrease duration of an activity
- Decrease size of playing area
- Frequent time out periods
- Permit participation at child's own rate
- Eliminate competitive and emotional stress factors
- Reduce weights

- Use transportation and support devices
- Sitting or lying down position may be better than standing
- Use relaxation techniques and relaxing music
- Incorporate proper breathing techniques
- Should not carry heavy backpacks

To address collisions and contact concerns:

- Assign zones of play
- Use individual activities
- Use singles vs. doubles in racquet games
- Use foam or Nerf balls when appropriate
- Group children according to size, abilities, and needs
- Provide clear concise directions, rules, and regulations
- Provide areas free of obstacles, barriers, or hazards
- Proper padding of facilities and equipment

To address visual and perceptual motor limitations

- Use brightly colored objects
- Use soft objects (Nerf)
- Use Velcro to assist on catching
- Decrease distances
- Change implement to decrease speed of flight and movement (is whiffle ball)
- Provide clear and close visual fields
- Increase size of implement (softball)
- Provide playing areas free of hazards
- Use appropriate illuminations
- Familiarize child with play area prior to start of the activity.

Learn more about things to avoid:

Avoid

- Contact sports and sports with sudden exertion, and/or risk of falling or impact
 - A blow to a chest or the strain of jumping/straining can results in serious injury or even aortic rupture
 - Gymnastics, diving, skating can be risky
- Competitive sports and/or demanding endurance activities
 - Golf is an exception (but may not be able to carry the bag)
- Activities that place excessive stress on the joints
 - Can cause dislocation or damage to joint surface

- Isometric sports
 - Weight lifting can create high blood pressure
- Agents that stimulate cardiovascular system (i.e. decongestions and caffeine)
- For those at risk for recurrent pneumothorax
 - Avoid breathing against resistance
 - Playing brass instrument
 - Positive pressure ventilation –i.e. SCUBA diving
 - Lungs may not be able to withstand stress from pressure gradients

5. School Absences and Fatigue

What you need to know

- Individuals may experience mental and physical fatigue
- May be absent due to medical procedures

What you can do

- Make accommodations for resting or taking break
- Have peers share class notes
- Monitor work so that it is challenging, but there are attainable and realistic goals
- Plan for absences and consider tutoring
- Communication with parents is important to meet these challenges

6. Emergency Planning

What you need to know

Emergency plans will be individually determined, based on behaviors and medical issues. It is important to mention new signs, symptoms, or pain to the child's parents. Please see Marfan syndrome website for examples if emergency plans.

- Although RARE, aortic dissection (rupture) can occur in children with Marfan syndrome.
 - It is important to recognize the symptoms; Aortic rupture is typically painful and may be described as a “tearing chest pain to goes through to the back.” Less commonly, an individual may suddenly collapse or faint. It is important to know aortic dissection may be pain free and the only symptoms may be a shortness of breath.

- Other signs: nausea, paralysis, parasthesia (tickling, numbness, prickling of skin)
- Spontaneous Pneumothorax (collapsed lung)
 - Sudden shortness of breath and chest pain can also be indication of spontaneous collapse of the lung
 - Other symptoms include; cough, fatigue, rapid breathing or heart beat, or the skin turning a bluish color
 - Other signs of cardiac or pulmonary problems are fatigue and irregular heart rate
 - Any of these symptoms may be serious and child should be transported to ER
- Retinal detachments
 - If there are any sudden vision problems, including flashing lights, spots in vision, blurred vision, translucent specks of various size, shape, and consistency in eye, blindness in a part of eye or sudden loss of vision should see the school nurse. Symptoms may occur gradually or suddenly
 - If child is on blood thinners, they may develop excessive bruising or spontaneous bleeding.

7. Resources

GeneReviews

Learn more about the genetics of Marfan Syndrome

<http://www.ncbi.nlm.nih.gov/books/NBK1335/>

The Marfan Syndrome Foundation

Education, Research and Support for individuals with Marfan syndrome and related disorders

www.marfan.org

Need-to-Know Information for Teachers:

<https://marfan.org/resource-library/need-to-know-information-for-the-school-nurse/>

Need-to-Know Information for School Nurses:

<https://marfan.org/resource-library/need-to-know-information-for-the-teacher/>

Physical Activity Guidelines:

<https://marfan.org/resource-library/physical-activity-guidelines/>

Classroom Accommodations for Students with Visual Issues

Classroom Accommodations for Students with Visual Issues is a resource from Boulder Valley Vision Therapy, P.C. (<https://www.bouldervt.com/>), in Boulder, Colorado. It describes a variety of variety of issues and offers strategies.

<https://www.bouldervt.com/wp-content/uploads/sites/478/2015/12/227-Course-Handout-CCIRA-2016.pdf>

Medline Plus

Consumer-friendly information about human genetics from the U.S. National Library of Medicine
<https://medlineplus.gov/genetics/condition/marfan-syndrome/>