Genetic Education Materials for School Success

Marfan Syndrome

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Acknowledgment:

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Disclaimer:

This is not intended to be medical or clinical advice. Any recommendations here should be reviewed for the appropriateness for your individual child and their unique situation. Also, please note that each document should include the date that it was last reviewed or updated. Research and guidelines that have been released after that date may represent more up-to-date recommendations.

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 or elastics, and helping to control how the body grows. In Marfan Syndrome, the connective tissue doesn't grow correctly. Marfan syndrome is typically inherited from an affected parent. 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, but not cognitive function.

Common Features of Marfan Syndrome:

- Tall, thin build with very long arms and legs
- Long, thin fingers
- Long, narrow face with deeply set eyes
- Flexible joints that might sprain or dislocate easily
- Possible bony abnormalities like a curved spine or sunken ribcage
- Eye problems
  - Nearsightedness
  - Possible cataracts or displacement of structures within the eye
- Fragility of some internal organs
  - Possible rupture of lungs or aorta (the major blood vessel from the heart)
- Heart murmur from floppy heart valves
- Easily tired or short of breath
- May have chronic pain

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In Depth

Medical and Dietary Considerations

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 specific medical issues for their child. School age children who have Marfan syndrome may be followed by a variety of specialists to monitor their medical conditions. Students may:

- have chronic pain and require medication as needed
- require other medication for heart or asthma issues
- often need to avoid strenuous exercise and contact sports, to reduce the risk of injury

Eyes

- Myopia or nearsightedness - most common eye finding
- Vision changes can progress rapidly during childhood
- Increased risk for
  - retinal detachment or displacement of the lens
  - glaucoma and early cataract formation

Skeletal System

- Tall, thin build - Arms and legs are disproportionately long for size of trunk
- Bone overgrowth of ribs - can either push sternum (breastbone) ***in*** (pectus excavatum); or push sternum ***out*** (pectus carinatum)
  - May require surgery to repair the chest wall deformity
- Long, thin fingers and toes
- Scoliosis which may be severe and progressive
- Facial features
  - Long narrow face with deeply set or down slanting eyes
  - Flat cheekbones and small and receding chin
- Dental issues due to high arched palate, and dental crowding
- Joint laxity (hypermobile joints) that are at increased risk of injury
- Flat feet
  - Orthotics and arch supports can lessen leg fatigue and muscle cramps associated with flat feet

Cardiovascular Manifestations

If present, may be severe and could require medications

- Dilatation of the aorta – an enlargement of the major blood vessel coming from the heart. Increased risk for aortic tear and rupture

- Enlarged heart or abnormal heart rhythm
- Mitral valve prolapse or tricuspid valve prolapse (floppy heart valves)

Lungs

- The lungs can have weakened areas of tissue that can rupture. This can cause spontaneous collapse of the lung (Pneumothorax)
- Increased likelihood of asthma or sleep apnea
- Possible increased risk of lung infections.
- Rib cage abnormalities and scoliosis may reduce lung capacity
    - Lead to shortness of breath and fatigue

What you can do

- Encourage at least yearly checkups in the child's Medical Home. Obtain updated medical information and review any plans at least yearly.
- Ensure up to date immunizations.
- Repeat vision assessment annually if not documented by primary care.
- Obtain medications and instructions for any heart, lung, or other medical condition.
- Consider developing a pain management plan.
- Inform parents of any suspected injury even if you do not think the mechanism could have caused significant injury.
- Notify parents of changes in energy level, behavior, mood, or academic performance.

Education Supports

It is important to have high learning expectations for children who have Marfan syndrome, utilizing their strengths and interests as a foundation. Encourage use of the core educational curriculum and modify it to meet the child's individual needs.

What you need to know

Marfan syndrome does not affect intelligence. Individuals may have gross or fine motor delays and/or visual difficulties which negatively impact school success. Students may also become easily fatigued during the day. They may have frequent absences and need help making up for missed schoolwork.

Gross and Fine Motor Issues

- Delays in gross motor development may be caused by joint hypermobility
- 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

Make sure seats and desks are appropriately sized for student's height and limb length. Be aware that student may get easily fatigued and need more frequent breaks. Consider modifying the amount of output required for the student and allowing child to keep a second set of books at home.

Gross and Fine Motor Issues

- 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

Behavior and Sensory Support

What you need to know

Individuals with Marfan syndrome may look "different" than their peers which may affect their self-esteem.

- Taller than average
- Very skinny and lanky
- Stretch marks
- Scoliosis
- Chest wall deformities

- 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 and may interfere with ability to focus or sit for long periods of time
- Students who are significantly taller than their peers may be treated as if they are older than their actual age

What you can do

- Allow privacy in changing areas 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
- Allow extra breaks and/or rests
- Make sure those interacting with the student are aware of the condition and the child's age

Physical Activity, Trips and Events

What you need to know

Marfan syndrome can cause a variety of problems, and each child will have unique problems and limitations. Most children with Marfan syndrome 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.

Musculoskeletal System

- The child may be tall and have long limbs
- The child may be very thin and may lack muscle bulk and strength
- Combination of underdeveloped muscles and joint hypermobility can contribute to poor coordination and delay in acquiring gross and fine more skills
- Joints may be hyper extensible and prone to dislocating. They may have joint contractures.
- 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

Eyes

- Increased risk for eyeball injury with minimal trauma
- 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 in tolerating bright light

Cardiovascular System

- 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 the aorta.
    - Beta blockers may cause fatigue, sleepiness, and reduced ability to concentrate.
    - Repair or replacement of mitral valve may be needed in school age child
    - If the child is on blood thinners, they may be prone to spontaneous bleeding and easy bruising.

Lungs

- The lungs can have weakened areas of tissue - can cause collapsed lungs.
    - 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

There is no single exercise program that works for each child with Marfan syndrome. In general, try for non-competitive, isokinetic activities done at a non-strenuous aerobic pace where he/she can stop and rest when tired and with no forceful contact with other players or objects. Customize activities that are of interest to the child AND safe for their condition.

Note, a child may need help developing a realistic understanding of their abilities and limitations. They must learn to not test limits which can be very hard for children. For more fragile children, encourage academic and artistic activities. Allow child to help with coaching or team management if there are restrictions

General Considerations for Marfan Syndrome in PE:

- Minimize risk of trauma especially head and chest
- Consider safety goggles to protect eyes
- Limiting activities involving endurance or intense competition. Avoid exercise to the point of exhaustion
- Strengthening exercises for both muscles and ligaments
    - Address muscle tone - Good for joint hyper extensibility
    - Avoid heavy weights and intense isometric exertion. Use low-tension settings on equipment
- Examples of safer activities include: Brisk walking, leisure biking, slow jogging, shooting baskets, slow tennis, use of 1–3-pound weights

Possible Modifications For Active Games/Sports

- Decrease duration of an activity or size of playing area

- Frequent time out periods
- Permit participation at child's own rate
- Eliminate competitive and emotional stress factors
- Sitting or lying down position may be better than standing when possible
- Assign zones of play to decrease collisions risk
- Use singles vs. doubles in racquet games
- Use foam or Nerf balls when appropriate
- Group children according to size, abilities, and needs
- Provide areas free of obstacles, barriers, or hazards
- Proper padding of facilities and equipment

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

Emergency Planning

What you need to know

Emergency plans will be individually determined, based on the student's medical needs. In Marfan syndrome, aortic dissection is not common in children, but it can occur. Collapsed lung and detached retina (structure at the back of the eye) are also medical emergencies that require prompt action.

What you can do

Seek guidance from the child's doctor about the potential risks. Identify what symptoms to watch for and what actions to take. In general, the following symptoms might necessitate a trip to the Emergency Department: chest pain, sudden shortness of breath or difficulty breathing, loss of consciousness, turning blue. Sudden vision changes including flashing lights, spots in their vision, or blindness also present a medical emergency. Make sure the EMTs and ED staff know the child has Marfan Syndrome and any other medical information available.

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Resources

Medline Plus

MedlinePlus is an online health information resource for patients and their families and friends. It is a service of the National Library of Medicine. This link will provide a good medical overview of the condition without being too technical.
https://medlineplus.gov/genetics/condition/marfan-syndrome/

Marfan Syndrome Foundation

The Marfan Syndrome Foundation is a nonprofit organization devoted to education, research and support for individuals with Marfan Syndrome and related disorders. Check out the useful information under the “Resources” and “Living with Marfan” tabs. www.marfan.org

Marfan Syndrome Foundation - School Concerns

From the Marfan Syndrome Foundation page, this is a very helpful page that includes sections for teachers, for the schools nurse, and also guidelines for safe physical activity.
https://marfan.org/resources/school-concerns/

Info for School Nurse and Primary Care Staff

Marfan Syndrome is a complex condition that will be best managed by good collaboration between the school nursing staff and primary care staff as part of the student's team.

Gene Reviews is a peer-reviewed point-of-care reference accessible via the National Library of Medicine. The entry for Marfan Syndrome provides a comprehensive summary of the condition and describes some ongoing surveillance and management issues.
https://www.ncbi.nlm.nih.gov/books/NBK1335/

HIPAA Concerns

Sometimes concerns by primary care staff about violating HIPAA regulations can hamper care by limiting access to necessary medical information. Please note that if care is intended to be delivered by school staff, clarifying medical orders or medication instructions represents continuation of medical care and is not a violation of HIPAA. To avoid any issues like this, ensure that the parents have signed a release of information form at the primary care office. The school nurse should keep a copy of that release in the student's file, if possible, to facilitate sharing of information in the event that there is difficulty obtaining needed information.

Example letter for physician to provide to school

"My patient_____ has been diagnosed with Marfan Syndrome. This is a genetic connective tissue disorder that affects multiple organ systems. This condition may require an Individualized Education Program (IEP) or 504 plan.

Medical complications with Marfan Syndrome can include recurrent joint injuries, chronic pain and fatigue, risk of aortic rupture, collapsed lungs, damage to the structures of the eye, abnormal heart valves or heart rhythm. Because of these, _____ needs the following accommodations:"

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