Marfan Syndrome

What is Marfan syndrome?

Marfan syndrome is an inherited condition. It affects the tissues that connect parts of the body.

How common is it?

One in 5,000 people in the United States have Marfan syndrome. It affects boys, girls, and people of all races equally. It is inherited in three-quarters of cases, but some cases are new in a family.

What are some common characteristics of children with Marfan syndrome or of Marfan syndrome as children present with it?

- Children with Marfan syndrome may be tall and thin with long arms, fingers, and legs. They may be more prone to having a curve in the spine (scoliosis), chest bones that stick out or cave in, flat feet, hernias, or crowded teeth. Not all children will have all these characteristics. Their arm spans might be longer than their heights. Their skin might have stretch marks and might heal with unusual scars. They may have trouble with sleep and may be more tired than average, especially at older ages.
- Children with Marfan syndrome have normal intelligence and, with increased medical monitoring, should have a normal life span.

Who might be on the treatment team?

- Pediatrician/primary care provider in the medical home
- Physical therapists
- Ophthalmologists (eye doctors)
- Cardiologists (heart doctors)
- Geneticists (gene doctors)
- Orthopedic surgeons (muscle and bone surgeons), as needed

What adaptations may be needed?

Medications

- Children with Marfan syndrome may be on medication that lowers blood pressure to reduce the strain on their blood vessels. The blood pressure medication can cause fatigue or dizziness. Medications that can increase blood pressure, such as decongestants, caffeine, and stimulant medications for attention-deficit/hyperactivity disorder, should generally be avoided.
- Staff who will be administering medications should have medication administration training (see Chapter 6).

Physical Environment and Other Considerations

Some children with Marfan syndrome might need to avoid vigorous physical activity, but normal play is encouraged. They can have loose joints and might get hand pain after prolonged grasping of writing tools such as crayons. If the child has vision problems, he or she might need special glasses or accommodations (see Visual Impairments Quick Reference Sheet [page 201]). Ask the child's doctor if he or she needs any restrictions on activities such as blowing up balloons.

What should be considered an emergency?

- Call emergency medical services (911) immediately for any of the following symptoms:
  - Aortic aneurysm or rupture can be a life-threatening emergency. It might start with chest pain, loss of consciousness, numbness, or color change (pale or blue).
  - Sudden changes of vision or unexplained flashes of light might mean a lens dislocation, which should be treated urgently. Sudden onset of chest pain could mean a pneumothorax, which is an air leak around the lung, indicated by sudden onset of chest pain.
- Plans should be made to address these emergency conditions if they occur during transportation.

What are some related Quick Reference Sheets?

Visual Impairments (page 201)

What are some resources?

National Heart, Lung, and Blood Institute: "What is Marfan syndrome?" (Web page), www.nhlbi.nih.gov/health-topics/marfan-syndrome

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