Marfan Syndrome
For Healthcare Providers

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools (www.negenetics.org).

Physical characteristics and/or symptoms

Note: not all people with Marfan syndrome will have all of these features.

Common features

Eyes:

- Myopia
- Ectopia lentis
- Increased risk for retinal detachment, glaucoma, and early cataract formation

Skeletal system:

- Bone overgrowth
  - Tall and thin
  - Extremities are disproportionately long for size of trunk
  - Overgrowth of ribs
    - Pectus excavatum
    - Pectus carinatum
  - 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
- Flat feet

Cardiovascular manifestations:

- Dilatation of the aorta
- Predisposition for aortic tear and rupture
- Mitral valve prolapse, tricuspid valve prolapse

- 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:

- Pneumothorax
- Asthma
- Sleep apnea
- Pectus excavatum and scoliosis may reduce lung capacity

Spine:

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

Headaches/Migraines

Recommended Routine Surveillance

- Annual echocardiograms
- CT/MRA scans in young adulthood
- Annual ophthalmologic exams
- Management of joint laxity

Emergency Protocols

In Marfan syndrome, most emergencies are cardiac and related to the risk of aortic dissection. However, there can also be eye-related emergencies with retinal detachment, and lung-related emergencies with pneumothoraces. The Marfan Foundation has resources available for an emergency preparedness kit to compile information in case of an emergency. https://www.marfan.org/expectations/treatment/emergencies

Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Cardiologist:
    - Dilatation of aorta
    - Mitral valve prolapse
    - Predisposition for aortic tear and rupture

    - Tricuspid valve prolapse
    - Enlargement of proximal pulmonary artery
- Geneticist / Genetic Counselor:
    - Diagnosis
    - Coordination of care
    - Genetic risk for family
    - Clinical trials
- Neurologist:
    - Headache/migraines
- Ophthalmology:
    - Myopia
    - Ectopia lentis
    - Retinal detachment
    - Glaucoma
    - Cataracts
- Orthopedist:
    - Scoliosis
    - Pectus excavatum/carinatum
    - Joint laxity
- Pulmonologist:
    - Lung Bullae
    - Pneumothorax
    - Asthma
    - Sleep apnea
    - Pectus excavatum and scoliosis may reduce lung capacity
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Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

My patient_____ has been diagnosed with Marfan syndrome. Marfan syndrome is characterized by abnormalities of the skeletal, ocular, and cardiovascular systems. Medical complications with Marfan syndrome may include management of joint laxity and cardiac abnormalities. Individuals with Marfan may need to avoid contact and competitive sports as well as isometric exercises and any activities that may cause joint pain and/or injury. Medical emergencies can occur involving the heart, eyes, and/or lungs. Because of these, _____ needs the following accommodations.

- Emergency Preparedness Kit from the Marfan Foundation: https://www.marfan.org/expectations/treatment/emergencies

Seven Important Aspects of School Life

"Marfan Syndrome at a Glance" will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions

Resources

Medline Plus Reference
https://medlineplus.gov/genetics/condition/marfan-syndrome/

GeneReviews
http://www.ncbi.nlm.nih.gov/books/NBK1335/

AAP Health Supervision Guidelines
http://pediatrics.aappublications.org/content/pediatrics/132/4/e1059.full.pdf

Additional resources including support group information can be found on the main website.