Marfan Syndrome

Marfan Syndrome is a connective tissue disorder caused by a gene *mutation* (change). Connective tissue is found throughout the body and helps support tissues and body organs. This means Marfan Syndrome can affect nearly all organ systems in your body. It can affect people of all races and ages. Marfan Syndrome can be *inherited* (passed on) from a parent who has it.

MEDICAL PROBLEMS OF MARFAN SYNDROME

- Serious heart problems are associated with Marfan Syndrome:
  - A leaky *mitral valve* (a heart valve that is located between the top and bottom parts of the heart) caused by a mitral valve *prolapse* (not closing all the way). A mitral valve leak can cause shortness of breath, dizziness, fatigue, or abnormal heart beats. Medication or surgery may be needed to help with these symptoms.
  - A weak *aorta* (the large artery carrying blood away from the heart to the rest of the body) can also cause problems. If the aorta enlarges greatly or tears, it can cause death. Warning signs of aortic enlargement may range from no symptoms to having pain or pressure in the chest or back. If aortic enlargement is discovered, surgery is usually necessary.
- Many people with Marfan Syndrome are near-sighted or have other eye problems. Some of these include:
  - *Dislocated* (out of place) eye lenses.
  - Detached retina.
  - Glaucoma.
  - Cataracts.
- Bone and joint (skeletal system) problems.
  - Long arms and legs.
  - Long, thin fingers.
  - Curvature of the spine *(scoliosis)*.
  - Chest is *oncave* (inward forming) or sticks out (pigeon chest).

DIAGNOSIS

Diagnosis (learning what is wrong) of Marfan Syndrome can be made by:

- Doing a complete physical exam and taking a complete family history.
- CT or MRI imaging to assess aortic size. The aorta should be followed and monitored closely to watch for enlargement.
- Echocardiogram. A sound wave imaging picture that looks at the heart, its valves, and the aorta.
- Electrocardiogram (EKG). This test checks your heart rate and rhythm.
- Slit lamp examination. This is an eye test that can check to see if your lenses are out of place.

TREATMENT

- There is no cure for Marfan Syndrome. Medical problems associated with Marfan Syndrome are followed and treated by your caregiver as needed.
- It is important to follow up with your caregiver on a regular basis after the diagnosis of Marfan Syndrome is made. Your caregiver may use the same tests that he or she used to diagnose the syndrome as part of your follow-up care. Heart conditions associated with Marfan Syndrome will need to be watched closely.
- Heart medicine called Beta-blockers may be used to reduce blood pressure. These help lower the stress on the heart and aorta. Beta-blockers can also control irregular heartbeats.
- It may be necessary to take *prophylactic* (preventative) *antibiotics* (medications that kill bacteria) if dental or genitourinary procedures are performed in people with mitral valve prolapse or those who have artificial heart valves.
- Avoidance of contact sports and strenuous exercise may be necessary to lessen the strain on the aorta. Your caregiver can discuss your activity limitations. If you have Marfan Syndrome, check with your caregiver if you are unsure which activities may be at risk for you.

MAKE SURE YOU:

- Understand these discharge instructions.
- Will monitor your condition.
- Seek immediate medical care if necessary.

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