MARFAN SYNDROME

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WHAT IT IS ?

Marfan syndrome (MFS) is an autosomal dominant heritable disorder of connective tissue that involves primarily the skeletal, ocular, and cardiovascular systems. Connective tissue supports many parts of your body. It as a type of "glue" between cells that:

PHelps bring nutrients to the tissues
PGives tissues form and strength
PHelps some tissues do their work.

HOW MANY ARE AFFECTED?

1 per 5,000 people is affected by Marfan's syndrome worldwide.

WHAT CAUSES IT?

Marfan syndrome is caused by defects in a gene called fibrillin-1. Fibrillin-1 plays an important role as the building block for connective tissue in the body. In most cases, Marfan syndrome is inherited, which means it is passed down through families. However, up to 30% of patients have no family history, which is called "sporadic." In sporadic cases, the syndrome is believed to be caused by a new gene change.

WHO GETS MARFAN SYNDROME?

Men, women, and children can have Marfan syndrome. It is found in people of all races and ethnic backgrounds.

WHAT ARE THE SYMPTOMS?

Marfan syndrome affects people in different ways. Some people have only mild symptoms, and others have severe problems. Most of the time, the symptoms get worse as the person gets older.

Skeleton: People with Marfan syndrome are usually tall with long, thin arms and legs and spider-like fingers called arachnodactyly. When they stretch out their arms, the length of their arms is greater than their height. They may have:

✓Bones (arms, legs, fingers, and toes) that are longer than normal
✓A long, narrow face
✓Crowded teeth because the roof of the mouth is arched
✓A breastbone that sticks out or caves in
✓A curved backbone
✓Flat feet.

Heart and blood vessels: Most people with Marfan syndrome have problems with the heart and blood vessels, such as:

☛A weak part of the aorta (the large artery that carries blood from the heart to the rest of the body). The aorta can tear or rupture.
☛Heart valves that leak, causing a "heart murmur." Large leaks may cause shortness of breath, fatigue, and a very fast or uneven heart rate.

Eyes: Some people with Marfan syndrome have problems with the eyes,such as:

☛Nearsightedness
☛Glaucoma (high pressure within the eye)at a young age
☛Cataracts (the eye's lens becomes cloudy)
☛A shift in one or both lenses of the eye
☛A detached retina in the eye.

Skin: Many people with Marfan syndrome have:

☛ Stretch marks on the skin.These are not a health problem.
☛A hernia (part of an internal organ that pushes through an opening in the organ's wall).

Nervous system: The brain and spinal cord are covered by fluid and a membrane. The membrane is made of connective tissue. When people with Marfan syndrome get older, the membrane may weaken and stretch. This affects the bones in the lower backbone (spine). Symptoms of this problem include:

*Painful abdomen
*Painful, numb, or weak legs.

Lungs: People with Marfan syndrome do not often have problems with their lungs. If symptoms in the lungs do arise, they may include:

*Stiff air sacs in the lungs.
*A collapsed lung if the air sacs become stretched or swollen.
*Snoring or not breathing for short periods (called sleep apnea) while sleeping.

HOW IT IS DIAGNOSED?

There is no single test to diagnose Marfan syndrome,it may need,

†Medical history
†Family history (any family members who have Marfan syndrome or who died at a young age from heart problems)
†A physical exam, including the length of the bones in the

arms and legs There may be hypermobile joints and signs of aneurysm, collapsed lung and heart valve problems.

†An eye exam, including a "slit lamp" test and may show defects of the lens or cornea,retinal detachment and vision problems

†Heart tests such as an echocardiogram every year to look at the base of the aorta.

†Fibrillin-1 mutation testing (in some people).

HOW IT IS TREATED?

There is no cure for Marfan syndrome, but certain activities can help treat and sometimes prevent related problems.

Skeleton

✓Getting a yearly exam of the spine and breastbone

✓Using a back brace or having surgery for severe problems.

Heart and blood vessels

✓Getting regular checkups and echocardiograms

✓Seeing a doctor or going to an emergency room for pain in the chest, back, or abdomen

✓Wearing a medical alert bracelet

✓Taking medicine for heart valve problems

✓Having surgery to replace a valve or repair the aorta if the problem is severe.

Eyes

☛Getting yearly eye exams

☛Wearing eyeglasses or contact lenses

☛Having surgery if needed.

Lungs

☛No to smoking

☛Consult a doctor if there is any problem with breathing during sleep.

Nervous system

†Taking medicine for pain if the membrane of spinal cord swells.

Diet

†Eating a balanced diet can help to maintain a healthy lifestyle, even though no vitamin or supplement can slow, cure, or prevent Marfan syndrome.

WHAT DO PREGNANT WOMEN WITH MARFAN SYNDROME NEED TO KNOW?

Women with Marfan syndrome can and do have healthy babies. Because pregnancy can stress the heart, pregnant women should see an obstetrician and other doctors familiar with Marfan syndrome, to help prevent problems with heart while pregnant.

WHAT ARE SOME OF THE EMOTIONAL AND PSYCHOLOGICAL EFFECTS OF MARFAN SYNDROME?

A genetic disorder can cause social, emotional, and financial stress. It often requires changes in outlook and lifestyle. People with Marfan syndrome may feel many strong emotions, including anger and fear. They may also be concerned about whether their children will have Marfan syndrome. Genetic counseling may also help to learn about the disease and the risk of passing it on to the children.

WHAT IS THE ROLE OF NURSE ?

The nurse's role in caring for people with Marfan syndrome is varied and depends largely upon each individual's symptoms and particular health problems. Apart from specialised care afforded to each affected organ, the nurse's main role is to provide support and education - both for patients and their families and loved ones. Nurses can provide emotional support, which will be vital during any hospital admissions, and assist with education on subjects such as family planning and genetic counselling, fitness and exercise, diet and nutrition, and any special precautions or follow-up treatment and examinations required. Feelings of isolation, resentment for being 'abnormal' and depression can be common among this group of patients, so nurses should focus on positive aspects of people's lives by encouraging them to join groups appropriate to their physical capabilities, follow a healthy diet and lifestyle, and lead as full and active a social and personal life as possible.

WHAT IS THE PROGNOSIS?

Heart-related complications may shorten the lifespan of people with this disease. However, many patients survive well into their 60s. Good care and surgery may extend the lifespan further.

WHAT ARE THE POSSIBLE COMPLICATIONS?

Complications may include:

† Aortic regurgitation
† Aortic rupture
† Bacterial endocarditis
† Dissecting aortic aneurysm
† Enlargement of the base of the aorta
† Heart failure
† Mitral valve prolapse
† Scoliosis
† Vision problems

HOW TO PREVENT IT?

Spontaneous new gene mutations leading to Marfan (less than 1/3 of cases) cannot be prevented. Those who have Marfan syndrome must consult doctor at least once every year.

ANY SUPPORT GROUPS?

National Marfan Foundation -- www.marfan.org

REFERENCES

Doyle J, Dietz III H. Marfan syndrome. In: Kliegman RM, Behrman RE, Jenson HB, Stanton BF. Nelson Textbook of Pediatrics. 19th ed. Philadelphia, Pa: Saunders Elsevier; 2011:chap 693.

Pyeritz RE. Inherited diseases of connective tissue. In: Goldman L, Schafer AI, eds. Cecil Medicine. 24th ed. Philadelphia, Pa: Saunders Elsevier; 2011:chap 268.