

MASS PHENOTYPE

MASS Phenotype is a connective tissue disorder that is similar to Marfan syndrome in that people with the condition have the similar **M**itral valve, **A**orta, **S**kin, and **S**keletal features. People with MASS phenotype do not have lens dislocation and do not show progressive and dangerous aortic root enlargement, hallmark features of Marfan syndrome.

MASS phenotype is similar to Marfan syndrome, but does not have dangerous aortic root enlargement.



What other names do people use for MASS phenotype?

MASS phenotype is also known as MASS syndrome.

How prevalent is MASS phenotype?

There aren't any good estimates regarding the prevalence of MASS phenotype.

What are the characteristics of MASS phenotype?

The features of MASS phenotype are:

- Mitral valve prolapse (MVP), a condition in which the mitral valve of the heart closes properly, but then flops backward, which allows blood to leak into the chamber from which it came instead of moving forward. This leaking is called regurgitation
- Aortic root diameter may be at the upper limits of normal for the person's body size, but it does not worsen over time. In addition, it does not increase the risk of aortic dissection.

- Skin stretch marks unrelated to weight change (striae).
- Skeletal (bone and joint) features of Marfan syndrome (including scoliosis, chest wall deformities, and joint hypermobility).

What causes MASS phenotype?

MASS phenotype can be caused by a change in the FBN1 (fibrillin-1) gene, the same gene that causes Marfan syndrome. It can be inherited within families (passed down through generations). It is autosomal dominant, which means that someone with MASS phenotype has a 50/50 chance of passing the gene to each child. There may be other genes that can cause MASS phenotype.

How is MASS phenotype diagnosed?

MASS phenotype can only be diagnosed after a comprehensive evaluation by an ophthalmologist, cardiologist, and geneticist (or other doctor) who look for features of a connective tissue disorder throughout the body. Both a slit lamp eye exam and an echocardiogram should be done to exclude eye lens dislocation and aortic enlargement, respectively. Echocardiograms should be performed intermittently to exclude emerging Marfan syndrome. Family history is also important. If one individual in the family shows development of an aortic root aneurysm, the diagnosis would change to Marfan syndrome for all affected people in the family.

Is there a genetic test for this gene?

Currently, there isn't a gene test that can differentiate MASS phenotype from Marfan syndrome.

How is MASS phenotype managed?

Like Marfan syndrome, MASS Phenotype affects different people in different ways. It is important that the treatment focus on the specific features that are present, regardless of the actual diagnosis.

Always talk to your doctor about your specific needs. Here are some key points about the treatment of MASS phenotype.

Treatment of mitral valve prolapse

If there aren't any symptoms related to mitral valve prolapse, such as shortness of breath, exercise intolerance, or abnormal heart rhythm (arrhythmia), medical treatment is not needed. However, the ascending aorta and mitral valve should be monitored for enlargement and function by echocardiography every year.

Endocarditis, a bacterial infection, is a concern for people with mitral valve prolapse. This can occur during routine dental procedures or minor surgery that could introduce bacteria into the bloodstream. The American Heart Association has guidelines about who needs antibiotics before a procedure to reduce the risk of infection. Talk to your doctor to determine if antibiotics are recommended for your specific situation.

People with rapid heart rhythms associated with mitral valve prolapse can benefit from treatment with drugs called beta blockers. Other drugs can be considered, and options should be discussed with a cardiologist. If mitral valve prolapse is associated with severe valve leakage, this can cause the heart to enlarge and to show reduced function. Medications such as angiotensin converting

enzyme (ACE) inhibitors or angiotensin receptor blockers (ARBs) can reduce the strain on the heart in some people.

People with serious symptoms may require surgery to repair or replace the mitral valve.

Treatment of the aorta

The aortic root diameter in people with MASS phenotype may be at the upper limits of normal for body size, but there is no progression to aneurysm or increased risk of a tear in the aorta (dissection). However, it is recommended that the ascending aorta be monitored by echocardiography for enlargement yearly during childhood and, if the aorta stays stable, less frequently into adult life.

Treatment of bone and joint problems

Many of the skeletal features that are similar to Marfan syndrome do not require treatment in most individuals. In some cases, some treatment is needed.

Scoliosis

Scoliosis is often more severe if it is related to a connective tissue disorder than in people who have scoliosis from an unknown cause. Bracing is sometimes used in people with curves in the 20°-40° range who have a reasonable side profile and whose skeletal growth is not yet complete. Surgery is an option when the curve is 40°-50° or greater, the side profile is abnormal, and there is back pain due to degenerative changes resulting from the scoliosis. Children need to be monitored carefully during the time they are growing. Adults who have a curved spine must also be monitored because, even between the ages of 15 and 75, there is a risk that the curve will progress. Annual spinal exams are recommended if there is a significant curvature or pain.

Chest Bone

Usually, when the chest bone sinks in (pectus excavatum) or protrudes outward (pectus carinatum), no treatment is necessary. However, surgery may be needed if it impacts how the heart and lungs work, which is very rare. Surgery should not be considered until most growth is complete.

Stretch marks

Stretch marks do not require medical treatment. They may fade over time.

What is the life expectancy for someone with MASS phenotype?

MASS phenotype does not affect life expectancy.

Do you have questions? Would you like more information?

- Call our help center, 800-862-7326, ext. 126 to speak with a nurse who can answer your questions and send you additional information.
- Visit our website at marfan.org. You can print information that interests you and ask questions online.