



# Marfan Syndrome: Diagnostic Criteria by Body System

Marfan Syndrome Diagnostic Criteria describes features that doctors use to diagnose (decide if someone has) Marfan syndrome. Diagnostic criteria can be in many parts of the body since Marfan syndrome affects many body systems.

There are both major and minor diagnostic criteria. This sheet lists the major and minor criteria in each body system. To learn more about these criteria and how doctors use them to make a diagnosis, please read “Marfan Syndrome: Diagnostic Criteria.”

Body System	Major Criteria	Minor Criteria
Skeletal System (Bones & Joints)	<p>Major criteria of the skeletal system is when a person has at least 4 of these features:</p> <ul style="list-style-type: none"> <li>• A chest that sticks out (pectus carinatum)</li> <li>• A chest that sinks in so much as to need surgery (pectus excavatum).</li> <li>• Arm span greater than height. This means that when your arms are stretched to the side, the distance from finger tip to finger tip is greater than your height.</li> <li>• Reduced upper to lower segment ratio. This is when the length of your torso (from shoulders to legs) is shorter than the length of your legs.</li> <li>• Positive wrist sign. This is when your thumb and little finger overlap when you grasp the other wrist.</li> <li>• Positive thumb sign. This is when you put your thumb on your hand and it extends beyond the palm.</li> <li>• Curvature of the spine (scoliosis) with a curve greater than 20 degrees.</li> <li>• Vertebrae (spine bones) that slip over each other (spondylolisthesis).</li> <li>• Flat feet (pes planus).</li> <li>• Extra-deep hip sockets (where the thigh bone meets the hip). This is also called “protrusion acetabulae” and doctors find it by hip x-ray.</li> </ul>	<ul style="list-style-type: none"> <li>• A chest that sinks in, but not so much as to need surgery.</li> <li>• Very flexible joints throughout your body.</li> <li>• The palate (roof) of your mouth is highly arched and the teeth are very crowded.</li> <li>• Certain facial features such as: a long, thin face; deep-set eyes; receding chin; or down-slanting eyes.</li> </ul>



National Marfan Foundation

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of people with  
Marfan syndrome  
and related disorders

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Body System	Major Criteria	Minor Criteria
Cardiovascular System (Heart & Blood Vessels)	<ul style="list-style-type: none"> <li>The part of your aorta closest to the heart bulges out or is enlarged (ascending aortic dilation or aneurysm).</li> <li>The layers of the ascending aorta are separated and may tear (dissection of the ascending aorta).</li> </ul>	<ul style="list-style-type: none"> <li>A “floppy” mitral valve (mitral valve prolapse, or MVP).</li> <li>Enlarged pulmonary artery without any known cause, before age 40.</li> <li>Calcium deposits in the mitral valve, before age 40</li> <li>A part of the aorta—either in the chest (descending thoracic aorta) or stomach (abdominal aorta)—is enlarged, before age 50.</li> <li>Tearing (dissection) of the aorta in the chest or stomach, before age 50.</li> </ul>
Pulmonary System (Lungs)	There are no major criteria of the pulmonary system.	<ul style="list-style-type: none"> <li>Sudden collapse of the lung for no known reason (spontaneous pneumothorax)</li> <li>Extra-large air sacs at the top of the lung (apical blebs). These are found by chest x-ray.</li> </ul>
Skin	There are no major criteria of the skin.	<ul style="list-style-type: none"> <li>Skin stretch marks, not due to pregnancy or weight gain.</li> <li>Two or more hernias (recurrent hernia).</li> </ul>
Spinal Column Sac (Dura)	<ul style="list-style-type: none"> <li>Swelling of the sac around the spinal column (dural ectasia). This is found with CT or MRI scans of the back.</li> </ul>	There are no minor criteria of the spinal column sac.

#### WAYS TO LEARN MORE ABOUT MARFAN SYNDROME DIAGNOSTIC CRITERIA:

- Call the National Marfan Foundation (NMF) Resource Center at 1-800-862-7326 ext. 26. You will speak with a nurse who can answer questions and mail out information.
- Talk with your doctor. Sometimes it helps to use information from the NMF when you speak with the doctor.
- Visit the NMF website at <http://www.marfan.org>. You can print information from the “About Marfan Syndrome” page. You can also ask questions by clicking “Support Services: Ask a Question.”