

BONES AND JOINTS IN MARFAN SYNDROME

Marfan syndrome often causes problems in the bones and joints—in fact, these are often the features that first lead a person to suspect Marfan syndrome and seek a diagnosis. These features (called skeletal features) happen when bones grow extra-long or ligaments (connective tissue that holds joints together) become stretchy—like loose rubber bands.

Only about one-third of people with Marfan syndrome have skeletal features so severe that they require treatment.



There are several skeletal features associated with Marfan syndrome. Many people with Marfan syndrome have more than one skeletal feature, but very few people have them all. While it is important for the skeletal features to be evaluated by an orthopedist (bone and joint doctor), only about one-third of people with Marfan syndrome have skeletal features so severe that they require treatment.

What are the common types of bone and joint problems in people with Marfan syndrome?

Here are some facts about common types of bone and joint problems in people with Marfan syndrome:

General Body Type

A person with Marfan syndrome will usually—but not always—be tall, slender, and somewhat loose-jointed or limber. The arms, legs, fingers, and toes may be disproportionately long when compared with the trunk. In some cases, they may not appear tall compared to the general public, but instead be tall for their family. (See Figure 1)

The face may appear long and narrow, in keeping with the general body shape. Infants often have deeply set eyes and appear older than their unaffected brothers or sisters at the same age. The roof of the mouth (palate) is often high and narrowly arched and the teeth are crowded. The lower jaw (mandible) often recedes, which may accentuate a common overbite (malocclusion).

Curvature of the spine (scoliosis)

Scoliosis is a curving of the spine to one side in which the vertebrae (bones in the spine) twist, usually into an S-shape or spiral shape. (See Figure 2) Scoliosis can be in any part of the spine, but is most common in the upper spine. About 6 in 10 people with Marfan syndrome have scoliosis, but only 1 in 3 need medical treatments for it.

Scoliosis is caused when ligaments in the spine are so loose they cannot firmly hold the vertebrae in place. Rapid growth in a child with Marfan syndrome can also make scoliosis increase. This often happens with the growth spurt during adolescence (teen-age years).

Scoliosis can be mild to severe, based on the size of the curve. Curves are measured by the angle between the vertebrae (as seen on the x-ray on the following page), given as a number or degrees. A small curve (less than 20 degrees) in an adolescent has a low chance of worsening. A moderate curve (20–40 degrees) has a greater chance of worsening in a child or adolescent. A large curve (more than 40 degrees) almost certainly will worsen, in either a child or an adult. This is because there is already so much imbalance in the spine that gravity will continue to worsen it.

All children are usually checked for scoliosis in school in the fifth grade. However, children with Marfan syndrome should be checked for scoliosis by their pediatrician even before fifth grade, then at each annual physical exam. This is done by having the child bend forward while a doctor or nurse examines the back. If a child does not show signs of scoliosis by the start of middle school, he or she probably will not develop it to a significant degree later in life. Scoliosis rarely begins in adulthood. Adults with scoliosis should be checked every 1 to 3 years.

Hunched back (kyphosis)

This is a curve best seen from the side. It is normal for people to have a slight kyphosis in the upper spine. However, increasing curves can cause deformity or back ache in some people. Kyphosis in people with Marfan syndrome may occur in the upper (thoracic) spine or in the lower (lumbar) spine.

Chest (pectus) problems

The two most common chest abnormalities related to Marfan syndrome involve the breastbone (sternum), and are caused when the ribs are too long.

- **Sunken chest (pectus excavatum)**
This develops to varying degrees in many people with Marfan syndrome, as well as in children who are not affected. In more severe cases, it may impair breathing, especially when there are other abnormalities in the spine, heart, or lungs. These breastbone abnormalities tend to become noticeable in adulthood. For some people, it may become a cosmetic concern.
- **Protruding chest (pectus carinatum)**
This develops when there is a protrusion (sticking out) of the breastbone outward. Some people with Marfan syndrome have a sunken chest on one side and a protrusion on the other. A protruding chest does not cause compression of the heart or lungs, but may still be cosmetically displeasing.

Foot problems

People with Marfan syndrome have feet that are long and slender. Extra-long bones and extra-loose ligaments can make the feet weak and less able to manage the pressure when people stand up. Some features that are common in Marfan syndrome are:

- Long, thin feet
- Flat feet (very low arch) or extra-high arch
- Long toes
- Hammer and claw toes (abnormal bending of the toes)
- Calluses (thick layers of skin) cause by too much pressure on one part of the foot
- Bunions (bone growth near the base of the big toe)
- Turned ankles (medial displacement)

Foot pain is sometimes a problem for people with Marfan syndrome. This is due to the stresses of a large frame on the flexible feet.

Your doctor should always check your feet and ankles—whether you have feet problems or not. Doctors who know about feet include orthopedic surgeons, rehabilitation specialists (physiatrists), and podiatrists. Your doctor should check:

- Range of motion—how easily and far you can turn your ankles and feet
- The exact place of your pain (if any)
- The strength of your tendons
- The fit of your shoes
- Your gait (how you move) when you run, walk, and walk on your toes

If you have problems due to Marfan syndrome, your doctor is likely to ask:

- How long have you had these problems?
- Are the problems getting worse?
- How severe (bad) are your feet problems?
- Do your feet hurt most in the morning or at night?
- Do feet problems keep you from doing certain activities?
- Do your feet hurt after standing for a few hours, walking a lot, or doing other activities?

Other problems may include:

- Back and limb pain
People with Marfan syndrome may have more aches in the back and limbs than other people. This may have something to do with the mechanics of living in a large body, with all of the attendant stresses, along with the laxity of the joints.

- **Spondylolisthesis**
This refers to a slip forward of one vertebra upon the one below it. This usually happens near the lower end of the spine. It can occur in anyone, but the forward slipping is more common in people with Marfan syndrome. Someone with this condition usually has a low backache or stiffness and is unable to bend forward to touch the toes. Usually there is no neurologic damage, except in very severe cases.
- **Dural ectasia**
A unique feature of Marfan syndrome is swelling (or ectasia) of the spinal cord sac (dura). This occurs when the spinal cord sac cannot withstand the pressure of the spinal fluid. If swelling occurs, it is frequently in the lower part of the lumbar or sacral spine. Sometimes, the enlarged sac will actually press into adjacent spaces, such as the pelvis.
- **Protrusio acetabulae**
In some people with Marfan syndrome, the hip sockets become deep during growth. This is called protrusio acetabulae. The acetabulum is the socket of the hip joint. The cause of this condition is not known. It does not cause problems or symptoms in childhood. In adulthood, the deepened sockets can lead to earlier arthritic change, which is noted with aching in the hips or groin. This affects less than five percent of people with Marfan syndrome. Some people with this diagnosis need to have artificial hips in middle age or later.

Marfan syndrome is thought of as a condition of loose ligaments. Yet dislocations or sprains in the major joints (such as a shoulder, knee, or ankle) don't seem to be more common in people with Marfan syndrome than in the general population. One reason may be that people with Marfan syndrome limit themselves from intense physical activity or twisting exercises in order to avoid stress on the heart or aorta. Another probable reason is that the major ligaments in their joints are not weak; they are just a little loose.

How are the common types of bone and joint problems treated in people with Marfan syndrome?

Many of the bone and joint problems in Marfan syndrome and some related disorders cause discomfort and pain. Other problems are more of a cosmetic (appearance) issue for people. In a few circumstances, surgery is recommended to treat bone problems. Treatment options are described below.

Curvature of the spine (scoliosis)

Treatment for scoliosis is sometimes recommended because scoliosis can cause many problems, including back pain, decreased lung function, and posture and shape disturbances. If the curve is increasing and is more than 20 degrees, some treatment is usually recommended in growing children and adolescents because exercise alone will not prevent a curve from progressing.

Whether or not a person needs treatment for scoliosis depends on the size of the curve and how much more a person will grow. Children are likely to need treatment for scoliosis as they have many years to grow. The younger a child is when scoliosis begins, the more likely the curve will increase. For example, a child less than three years old with a curve of only 10 degrees may need treatment later in life while a 14-year-old with a small curve (less than 20 degrees) might not. People of all ages with large-sized curves (greater than degrees) almost always need medical treatment.

Doctors use a Risser Scale (or Risser Score) to measure how much more bone growth a child is likely to have. This is based on findings from an x-ray of the hip bones (pelvis). The Risser Scale goes from 0 to 5, with 0 being the most expected bone growth and 5 being the least. Children with a Risser Score of 0 to 2 are likely to have several more years of bone growth and are at risk for scoliosis to get worse.

Scoliosis can be treated in one or both of these ways:

- Back brace

A back brace is often recommended for children with scoliosis between 20 degrees and 40 degrees. A brace is a custom-molded, padded plastic “jacket” that fits around the trunk, from shoulders to waist, and temporarily straightens the spine and stops the curve from getting worse. However, the brace cannot permanently straighten the curve. When the brace is removed, the spine will gradually return to its original curvature. The goal of the brace is simply to keep the curve from getting worse with growth.

A back brace is usually worn 23 hours per day. The time off is for bathing/showering and dressing. In addition, the patient may remove the brace for swimming or other activity, if necessary. The brace is worn until growth is complete. For girls, this is usually until age 14 or 15; for boys, it is 16 to 17. The brace works in many, but not all, people with Marfan syndrome. If the curve is too great or getting worse, bracing does not prevent the need for surgery.

- Surgery

Doctors recommend surgery when the scoliosis curve is 40 degrees or more so that lung problems, back pain, and further deformity can be avoided. If surgery is chosen, it is usually safer and more effective to correct the curve before it progresses further.

Scoliosis surgery involves straightening the spine with metal rods and fusing it in the straightened position. The rods are placed deep under the back muscles, against the vertebrae, so they cannot be felt. While the rods are holding the spine straight, bone chips grow together to fuse the spine and permanently hold it straight.

The rods do not hurt after the surgery has healed. In children under 10 years old whose spines are still growing, an orthopedic surgeon may use special “growing rods” that can be made longer as the child grows.

Sometimes, the surgery is performed through a posterior approach, from the back. If the curve is very large or rigid, it may be performed using both an anterior approach, going to the spine from the front and removing the deformed spinal disks, and posterior approach.

Scoliosis surgery is usually successful. A blood transfusion is often needed, but the patient may be transfused his or her own blood if the blood is stored a few months prior to surgery. Complications—such as the rods coming loose, the spine not fusing, or nerve damage—may happen, but they are rare. Usually, it takes one to two months before the patient may return to school or work.

People often ask if they can have MRI studies when they have rods in their spine. MRI studies are possible no matter what kind of metal rod is used. The rods show up on the MRI as a “halo” or invisible area. Titanium (one kind of metal) rods make the smallest halos.

How Is Treating Scoliosis Different in Marfan Syndrome?

The way scoliosis is treated depends on what type it is. The most common type of scoliosis is called “idiopathic scoliosis”—meaning its cause is unknown. This is different from scoliosis caused by Marfan syndrome. Talk with your doctor about how treatment might affect you. Here are some facts it helps to know:

- The success rate of using a brace to treat Marfan scoliosis is lower than for idiopathic scoliosis. Most children with Marfan syndrome who have a curve of more than 25 degrees and a Risser score of 0 to 2 will need surgery at some point, even after using a brace.
- In 1 in 4 people with Marfan syndrome, pedicles (a part of the vertebrae) are narrow and thin. When this happens, using screws to keep the spine rods in place may not work. If so, the surgeon needs to use other methods to keep the rods in place.
- The dura (tube around the spinal column that holds the spinal fluid) is weak in people with Marfan syndrome. This increases the risk of spinal fluid leaking during surgery. The surgeon must take special care when the dura is weak.
- People with Marfan syndrome are more likely to have their scoliosis come back after surgery. This happens less often when surgeons fuse a large section of the spine.
- People with Marfan syndrome may have an unusual scallop shape on the inside of their spinal column. The surgeon needs to use CT scans to understand this shape when planning surgery.
- The spine of someone with Marfan syndrome may curve forward (kyphosis) as well as to the side (scoliosis). The surgeon may need to plan surgery to manage both the scoliosis and the kyphosis.

Hunched back (kyphosis)

The treatment for kyphosis is similar to the treatment for scoliosis. In a growing child, a brace is sometimes effective. In an older patient or in someone with more severe kyphosis, a brace does not help. Therefore, the patient should try exercises to keep the back free of pain.

If bracing doesn't relieve the pain, surgery is an option. It is done in almost the same way as scoliosis surgery, although it is less commonly needed. People with kyphosis should be careful to maintain good calcium intake and a mild exercise program to prevent osteoporosis, which may make the kyphosis worse.

Sunken chest (pectus excavatum)

There is no brace available to correct a sunken chest (pectus excavatum). The only treatment option is surgery. There are often good medical reasons, in addition to cosmetic reasons, to repair a severe pectus excavatum. If needed, surgery should be performed in mid- to late-childhood by a children's general surgeon.

Surgery consists of raising the breastbone and ribs, straightening them, and holding them with a metal bar. This surgery is called a Nuss procedure. It requires several days of hospitalization. The success rate is high. The metal bar is removed after 4-6 months in a brief outpatient procedure. Afterward, the sternum will look much improved, although there is usually slight residual abnormal chest shape that cannot be fixed completely without taking undue risks. In a few cases, the deformity may recur if it is corrected early in life.

Protruding chest (pectus carinatum)

For a protruding chest (pectus carinatum), sometimes a brace can effectively push inward on the sternum to change its outward growth pattern, but this is not used often. Surgery is another option.

Rotation of the ribs, another form of chest protrusion, is a side effect of scoliosis. The ribs on the side of the curve usually protrude farther in the back. If spine surgery is done, the ribs often straighten out. Rarely, an operation on the ribs themselves is needed.

Foot problems

Treatment for foot problems associated with Marfan syndrome and some related disorders is not usually required. An arch support cannot make a flat foot develop an arch, but it may improve the gait or lessen discomfort. Special cushions, inserts, or orthotics may help. Wearing shoes with low heels, or no heels, is recommended. The key is to try different shoe styles to find one that is comfortable.

People with Marfan syndrome may also have curled toes (claw toes or hammer toes). Wearing tight shoes can make this worse.

Foot pain can be caused by arthritis, pressure on one part of the foot, tired muscles, or calluses. Aches can be treated with heat, an over-the-counter analgesic (such as Tylenol®), and modification of activities.

Foot surgery is rarely needed and should be undertaken only after careful trials of non-operative treatments. People sometimes need surgery when it is hard to stand on tiptoes or “push off” while walking because of very flexible flat feet. There are many types of surgery. They include:

- Osteotomy: Changes (realigns) the position of bones to improve how pressure is spread in the foot.
- Fusing joints (arthrodesis): For pain caused by arthritis. This type of surgery joins the ends of the bones together so that certain joints can no longer move.
- Hammertoe repair: To keep toes straight by moving toe tendons or fusing toe joints. This surgery should only be done to reduce pain and not to change how toes look.

There is no proven success for artificial ankles or toe joints.

You cannot always prevent foot issues due to Marfan syndrome, but there are things to try before problems occur:

- Stay the right weight for your height.
- Wear shoes that fit well, add support, and have low heels. You may be able to find these shoes on the Internet, although there might be just a few styles. Of note, these shoes can cost a lot, but many people say it is worth it to them to avoid foot pain.

Spondylolisthesis

Someone with this condition usually has a low back ache or stiffness and is unable to bend forward to touch their toes. Usually there is no neurologic damage, except in very severe cases. Treatment options include exercise or braces for mild cases; if slipping is greater than 30 percent, surgery may also be considered.

Dural ectasia

Symptoms of dural ectasia include low back ache and a burning sensation, numbness, or weakness in the legs. The symptoms resemble any back ache; however, enlarged dura can be seen on magnetic resonance imaging (MRI) or computed tomography (CT).

Medication or spinal shunting (inserting a tube into the spinal sac to help relieve the pressure from the fluid that accumulates around the base of the spine) may help in severe cases of dural ectasia. It is best to leave mild cases alone.

Chiropractic management of back pain

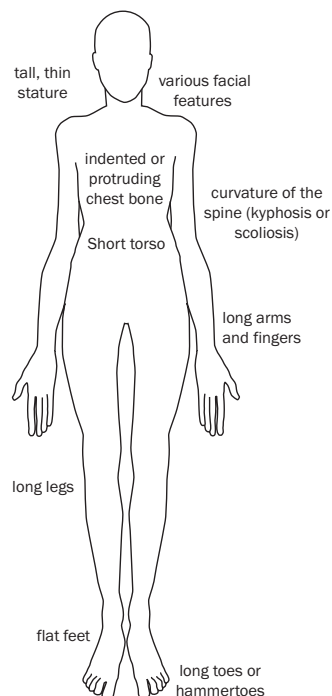
There are no specific guidelines on chiropractic care for people with Marfan syndrome, and The Marfan Foundation does not have an official stance on chiropractic care. Every case needs to be assessed individually. It is recommended to discuss the issue with your doctors who are familiar with your full medical history and physical status. Please be aware that chiropractic manipulation is not recommended for those with certain related disorders of connective tissue, such as Loeys Dietz syndrome, if they have cervical instability.

Some people with Marfan syndrome report having favorable experiences with chiropractic care; however, chiropractic care should be approached cautiously. If you are considering chiropractic care, we recommend identifying a provider who is knowledgeable about Marfan syndrome. After a patient history and physical exam to exclude contradictions for manipulation, low-stress manipulations can be tested with progress forward as tolerance dictates.

Chiropractic care remains a concern primarily with cervical manipulation, which may put people at risk of vertebral and carotid dissections. People with Marfan syndrome who see chiropractors should warn their practitioners about this potential issue.

**Figure 1
Common Skeletal
Features in
Marfan Syndrome**

Note: Not all people with Marfan syndrome have the “typical” body type.



**Figure 2
Spine X-Ray
of Person
with Marfan
Syndrome**



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 from the pages that interest you. You can also ask questions online.