



Our Community's Top Questions Answered
HELP & RESOURCE CENTER FAQs

Started in 1992, the Foundation's Help & Resource Center reaches more than 5,000 people around the world via our websites, emails, and one-on-one phone calls. Our registered nurses, Director Stephanie Amdur-Clark, MSN, RN, NP, and Janice Petrella Lynch, MSN, RN, answer a variety of medical questions from the community. Help Center Manager Kathleen Bolton offers Spanish-language services to our community. The team put together the most common questions they receive at the Help & Resource Center.

1 How do I know if/when I should get evaluated for Marfan syndrome or a related condition? I believe that I may have some clinical features, and my sibling seems to have some features as well.

If you or a family member show signs of Marfan syndrome or another connective tissue condition, it is important to seek an evaluation, even if the symptoms seem mild. Key indicators like long limbs, flexible joints, eye issues (such as lens dislocation), or cardiovascular problems can point to Marfan syndrome or a related condition. Given the potential for serious complications, especially with the heart and aorta, early detection is crucial for proactive management. Schedule a consultation with a cardiologist or geneticist familiar with connective tissue conditions for a proper evaluation.

2 What specialists should I see to be evaluated for a genetic aortic and vascular condition?

A thorough evaluation for a connective tissue condition requires a multidisciplinary approach. Start with a cardiologist for initial screening and possible imaging to assess heart and aortic health. Then referrals to a geneticist or a genetic counselor, an ophthalmologist for eye-related concerns, and an orthopedist for musculoskeletal issues can be made. Depending on your symptoms, a pulmonologist may also be important if lung function is affected. It's important to find specialists who have experience with these conditions.

3 My family tells me that I should get checked and evaluated for a connective tissue condition. There's nothing that can be done for me, so why do I need to do this? Anyway, I would rather not know.

While it's understandable to feel apprehensive, an evaluation can open doors to vital treatments that significantly improve quality of life and help to prevent complications. Even if you feel fine now, connective tissue conditions can lead to life-threatening issues like genetic aortic and vascular aneurysms (an enlargement of the major blood vessel to the heart). Early detection allows for regular imaging, medications, and surgical interventions that can increase longevity and save lives in some cases. Knowledge truly is power—so getting evaluated isn't just about a diagnosis; it's about safeguarding your future.

4 For years I have lived with various medical issues that no one has been able to figure out. Now I am wondering if I have Loeys-Dietz, Marfan, VEDS, or a related condition. I'm in my 40s. Is it too late to seek a diagnosis and care?

It's never too late to seek a diagnosis. Many people are diagnosed later in life, especially when they've had unexplained symptoms for years. While earlier detection may have helped prevent some issues, getting a diagnosis now is still crucial for managing your health going forward. With proper care, you can minimize future risks, especially concerning your heart, spine, and eyes, and improve your quality of life through targeted treatments and lifestyle adjustments.

5 Do I need to get genetic testing? Where do I go to have the genetic testing done? I hear that it is very expensive.

Genetic testing can help confirm a diagnosis of Marfan syndrome or related conditions, though a clinical diagnosis is sometimes sufficient. Testing can provide clarity for you and your family, especially when considering the hereditary nature of these conditions. Testing is typically done through a geneticist or a specialized clinic. Costs can vary, but many insurance plans cover genetic testing if medically necessary. Additionally, it is good to get long-term health insurance before genetic testing as it may be denied after genetic testing.

6 I have Loeys-Dietz syndrome, and I am being told contradictory information by my friends and family. Are there any physical activities I should avoid and why?

With Loeys-Dietz syndrome (LDS), avoiding certain physical activities is essential to reduce the risk of vascular complications, especially involving the aorta. High-intensity sports, heavy lifting, and activities that involve sudden, high-pressure strain on the body (like weightlifting or contact sports) should be avoided. Low-impact activities, such as walking, swimming, or light cycling, are generally safe, but it's important to consult with your cardiologist or a connective tissue specialist to develop a plan that minimizes risks.

7 I have Marfan syndrome, and my spouse and I are hoping to have a baby. I would like to know what we should do to prepare for the pregnancy and the delivery.

Planning a pregnancy with Marfan syndrome requires careful preparation and coordination with your healthcare team. Cardiovascular risks, particularly involving the aorta, must be closely monitored before and during pregnancy. You should work with a cardiologist, obstetrician, and genetic counselor who are experienced in managing Marfan syndrome pregnancies. A detailed birth plan that includes potential surgical or intervention options should be discussed. Genetic counseling is also recommended to understand the chances of passing Marfan syndrome to your child.

8 I feel like my care is fragmented and disorganized. How do I "build a team" of specialists that can help me and communicate with one another?

Building a cohesive care team requires taking a proactive approach. Start by identifying a primary specialist, such as a cardiologist or geneticist, to coordinate your care. Ensure that each specialist you see (e.g., orthopedist, ophthalmologist) is aware of your condition. Ask your doctors to communicate with each other by sharing your medical records and discussing your treatment plan. Using a patient portal or care management app can also help track appointments and share information among your team.

9 Pain is a daily interference in my life. What can I do about it so I can try to live a more pain-free life?

Chronic pain is a common challenge with connective tissue conditions. Managing pain requires a holistic approach, which may include medications, physical therapy, and alternative treatments like acupuncture or massage. Working with a pain management specialist can help tailor a regimen that's effective for you. Additionally, counseling or support groups can offer emotional coping strategies. It's important to listen to your body, avoid activities that exacerbate the pain, and keep communication open with your healthcare team to adjust your care as needed.

10 Please explain what the recovery after heart surgery will be like. I need to plan for it since I live alone.

Recovery after heart surgery can be a gradual process that varies depending on the type of surgery and your overall health. You'll likely spend a few days in the hospital, followed by several weeks or months of at-home recovery. It's important to arrange for support during the first few weeks—whether through friends, family, or hiring a paid caregiver—since you may need assistance with daily tasks. You'll also need to attend follow-up appointments, take medications, and potentially participate in cardiac rehabilitation. Planning for rest, nutrition, and emotional support will help make your recovery smoother.

Whether you're seeking a knowledgeable doctor, a supportive community, or more information about your condition, we're here to help. Just visit [Marfan.org/resources](https://marfan.org/resources) or reach out to the Help & Resource Center team at [Marfan.org/ask](https://marfan.org/ask) or call 800.8.MARFAN. A team member will be in touch with you as soon as possible. We appreciate your patience and look forward to assisting you soon.

If you have a medical question, get connected with our Help & Resource Center team at:

MARFAN.ORG/ASK

800.8.MARFAN



Stephanie Amdur-Clark, MSN, RN, NP
 Director of Help & Resource Center



Janice Petrella Lynch, MSN, RN
 Nurse, Help & Resource Center



Kathleen Bolton
 Help Center Manager

