



**Guidance for Establishing and Applying for Institutional Directory for Marfan, VEDS, Loeys-Dietz Syndromes and Related Genetic Aortic and Vascular Conditions**

The diagnosis, evaluation, and management of Marfan, vascular Ehlers Danlos (VEDS), Loeys-Dietz (LDS), familial aortic aneurysm, Beals (CCA), Shprintzen Goldberg and Sticklers syndrome as well as other related genetic aortic and vascular conditions, require health practitioners from multiple disciplines with specialized knowledge, skills, and experience in heritable disorders of connective tissue. Patients and families are best served in clinics with demonstrated interest and expertise in the field.

To develop the expertise and commitment necessary for a clinic specializing in Marfan and related connective tissue and genetic aortic conditions, a site is encouraged to meet the following criteria:

1. The clinic should offer coordinated, multidisciplinary evaluation and management of Marfan syndrome and related conditions. The cardiologist, cardiovascular surgeon, medical geneticist, ophthalmologist, orthopedist, vascular surgeon and genetic counselor should be knowledgeable about these conditions, their variable manifestations, the utility of molecular genetic testing, and the current management options. Other physicians who should have established connections to the clinic include, but are not limited to, specialists in thoracic surgery, vascular surgery, neurosurgery, general surgery, radiological imaging, echocardiography, pulmonology, obstetrics and gynecology, pain management, and social work. Extensive clinical experience with Marfan and related conditions is not necessary for other specialists; however, it is necessary for them to have an established relationship with the clinic director and coordinator. Clinics should also provide access to ancillary services such as lab services, physical therapy, occupational therapy, rehabilitation, and psychiatry. When a patient requires specialty care not available at the clinic, referral to appropriate consultants should be arranged.
2. The clinic should be directed by a health professional, typically a physician, who has a demonstrated interest and expertise in some aspect of Marfan syndrome, VEDS, LDS and related conditions as evidenced by extensive clinical and administrative experience as well as publications. Although the precise area of medical specialty is not of primary importance, the physician must be willing to assume administrative responsibilities for the clinic and should be given appropriate authority by the medical institution in which the clinic exists.
3. The medical director or other physician should act as a liaison with a patient's primary care doctor or local cardiologist/surgeon, if needed.
4. A clinic should have a coordinator, who may be a genetic counselor, nurse, nurse practitioner, social worker, or secretary. This coordinator should perform the following important functions:
  - a. Facilitate scheduling of all appointments, with particular regard to patients who find it difficult to make multiple trips to a clinic.
  - b. Educate the patient or family in detail and in advance about their upcoming visit, explain billing and insurance issues, and determine which medical records and imaging studies the patients need to bring to the visit or forwarded in advance.



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- c. Ensure that the patient follows through on the scheduled appointments.
  - d. Conduct an exit interview/counseling session with patients and make sure all questions are answered.
  - e. Ensure that the family and referring physician or agency receive correspondence summarizing the visit.
  - f. For diagnosis and management of patients with financial or insurance hardships, the coordinator should assist patients and families in obtaining appropriate and adequate insurance or funding from other sources to cover the costs of multidisciplinary care. Consideration should be given to innovative approaches to billing and collection within institutional guidelines, such as a sliding fee scale based on income; a single all-encompassing clinic charge; use of clinical research center funds if clinical investigation is being performed; or charge reductions when more than one family member is evaluated on the same day.
5. Clinics should begin to maintain a database of patients with Marfan and related conditions and services (if they have not done so already). In the future, verification may be requested.
6. For surgeons affiliated with the clinic, statistics for the previous full one-year and 5-year period surgeries for cardiovascular composite graft and valve-sparing operations in patients with and without Marfan or a related condition will be requested.
7. We recommend Institutions maintain a website so that patients have easy access to information and support. The Foundation will be happy to provide guidance and input and will review all websites.
8. Because of the tragedies resulting from misdiagnosis of dissection and other emergencies commonly associated with patients with genetic aortic conditions, we are asking for each site to institute an emergency protocol for these patients, if there is not one already established. For example:
  - a. Is there an on-call resident at all times, that can help a patient make a decision about going to the emergency department or not?
  - b. Is there an on-call resident that can talk to an outside physician?
  - c. Is there an on-call resident that can help coordinate physician to physician dialogue for urgent consultation between vascular and cardiac surgeons or other specialists?
  - d. Has the emergency department at your institution been made aware that you have patients that can have life threatening dissections and know about emergency department procedure and concerns for this population.
  - e. Does your clinic regularly note on the electronic medical record a highlighted patient coordination care note that provides instructions for emergency situations? For EPIC instructions, [click here](#). In other systems, it may be called an 'FYI' alert and this can vary according to institution electronic medical record version and vendor.
9. The transition from medical management in childhood to adulthood is especially important for pediatric clinics and a clear plan should be in place for all patients.



10. Each institution must utilize the 2010 Diagnostic Criteria for Marfan Syndrome, and the 2017 International Classification of the Ehlers-Danlos Syndromes Diagnostic Criteria for VEDS.
11. Each institution should adhere to the Foundation's recommendations for medical treatment for children and adults with Marfan syndrome and related conditions.
12. Each clinic must inform patients about The Marfan Foundation and offer its resources to patients and families as well as to the local care team if the family lives outside of commuting range of the clinic.
13. Each institution should inform patients of the Foundation's Marfan and Related Conditions International Patient Registry with Backpack Health.
14. The health professionals of a clinic are encouraged to utilize the staff at The Marfan Foundation and Professional Advisory Board for advice about diagnosis and management.
15. The clinical staff should be willing to act as an educational resource for the medical profession, The Marfan Foundation, the media, and the lay public by engaging in the many community and educational events hosted by the Foundation and its members.
16. Although the primary functions of the clinic are patient evaluation, treatment, counseling, and education, the professional staff should be willing to advance understanding of Marfan, VEDS, LDS and other related conditions through basic, clinical, and translational investigations. While independently designed and conducted studies are encouraged, cooperation in collaborative investigations with The Marfan Foundation through its Professional Advisory Board is encouraged.

**Recommended Services:**

1. The clinic should conduct a regular outpatient clinic dedicated to Marfan, VEDS, LDS and related conditions, if possible, and it should be noted on the website when available.
2. The turnaround time for echocardiography, MRA, CT, and ultrasound reports should be limited to 24 hours to ensure that patients receive results during their cardiology visit. Alternatively, physicians must be able to access images for discussion during patient visit.

**Application submission**

1. Applications and CV's must be submitted electronically with complete information through our website.
2. A 60 min phone interview with Foundation staff will be required after application submission.
3. Yearly application updates will be required.