From the Heart
The John Ritter Foundation for Aortic Health™

Genetic evaluation and testing in thoracic aortic aneurysms and dissections

Approximately 20% of thoracic aortic aneurysms and dissections are related to a genetic or heritable cause.

The 2022 ACC/AHA Guideline for the Diagnosis and Management of Aortic Disease includes a **Class 1 (strong) recommendation** that patients with thoracic aortic aneurysm or dissection (TAAD) who have any one of the following risk factors should be referred for genetic counseling and testing. The presence of even a single risk factor is sufficient to meet this recommendation.

- Presentation of aortic disease at <60 years.
- A family history of TAAD or peripheral/intracranial aneurysm or rupture in a first- or second-degree relative.
- A family history of unexplained sudden death at a relatively young age in a first- or second-degree relative.
- TAAD and syndromic features of Marfan syndrome, Loeys-Dietz syndrome, or Vascular Ehlers-Danlos syndrome (VEDS).

Genetic diagnosis can be lifesaving, leading to elective surgery before a dissection occurs. It is useful for guiding management, surveillance, and protecting at-risk relatives.

Many patients with genetic aortic disease lack obvious physical features (e.g., those seen in Marfan syndrome). A lack of a marfanoid phenotype or syndromic features is not a reason to forgo genetic evaluation.

If you are unsure, refer. A timely evaluation can make all the difference. Refer to a geneticist or genetic counselor with expertise in cardiovascular genetics. If you don't know of an appropriate genetics provider, The John Ritter Foundation for Aortic Health can help connect your patient to appropriate resources.

Questions or need resources?

Contact **The John Ritter Foundation for Aortic Health** at **info@johnritterfoundation.org** or **(213) 218-3329**.

This handout is intended for general informational purposes and does not reflect all current recommendations. Clinical decisions should be based on individual patient needs and professional judgment. Last reviewed: November 2025.