

The Aggressive Course of *PRKG1*-Associated Arterial Disease: Early Dissections at Near Normal Diameters

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Background

What is *PRKG1*?

- Rare cause of non-syndromic HTAD
- Dissections without aneurysms
- Single recurrent gain-of-function variant (p.Arg177Gln) in all cases

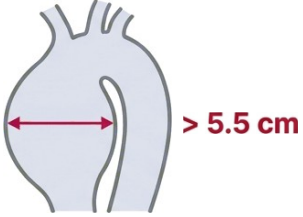
67

%

Type A
dissections
86

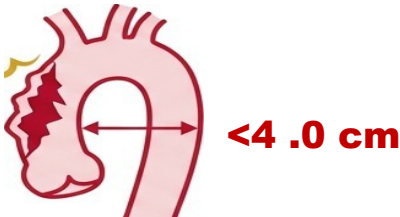
% Have

Standard Protocol



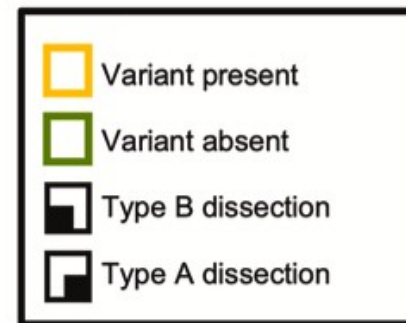
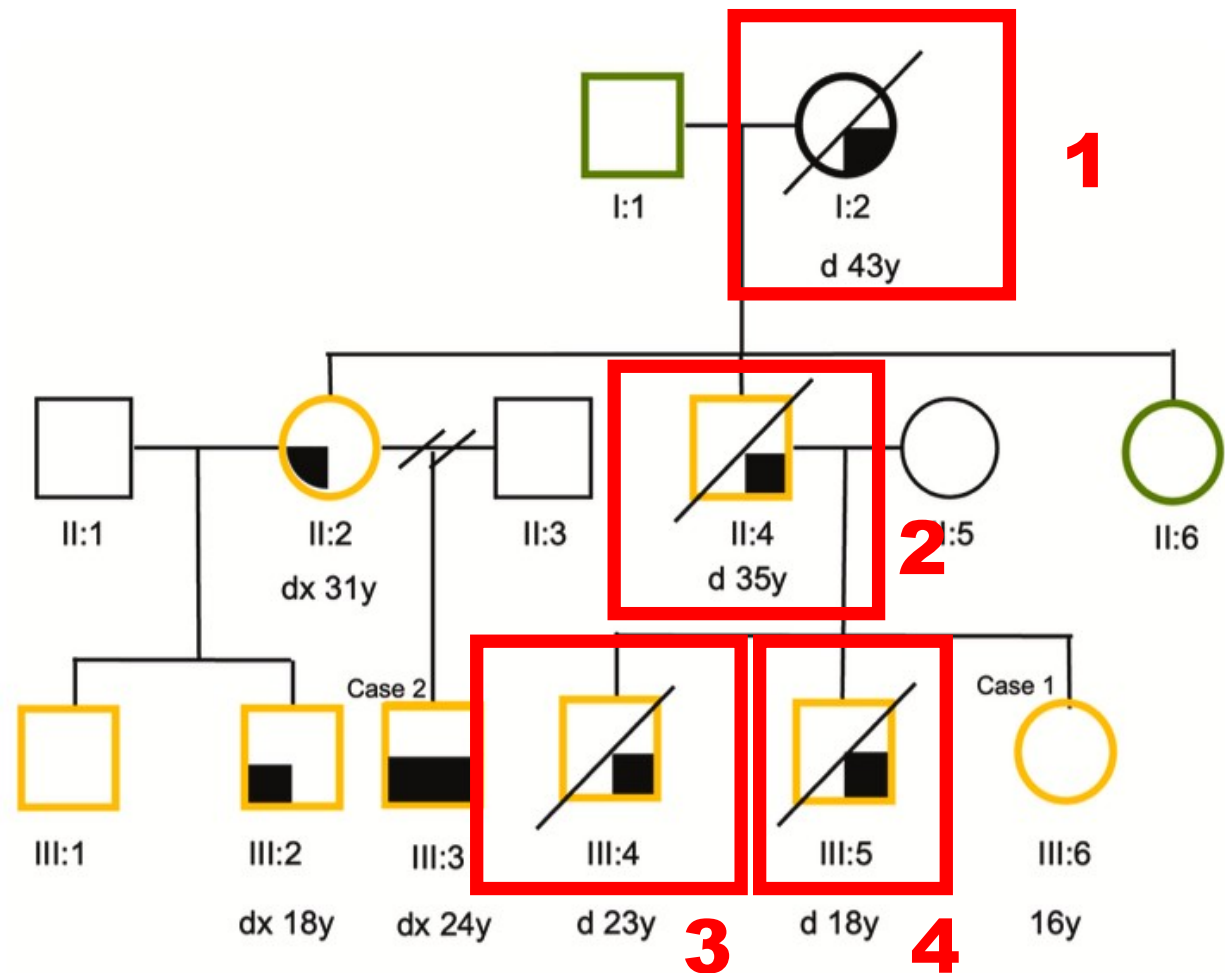
Intervention recommended only when diameter exceeds safety threshold.⁵

The *PRKG1* Reality

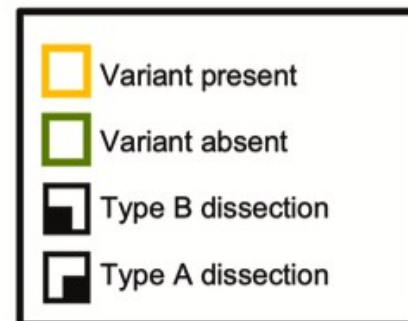
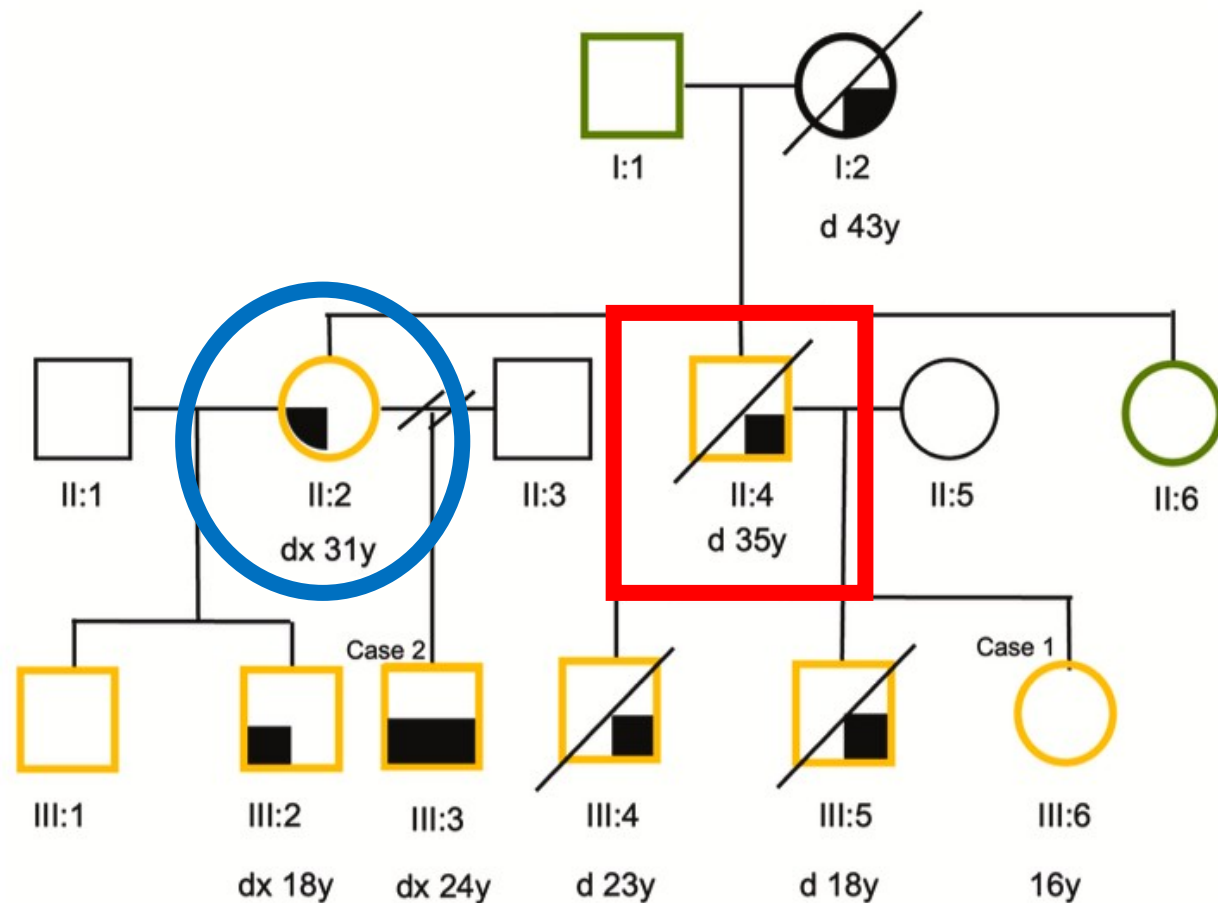


Outcome: Fatalities occur because patients dissect before significant dilation

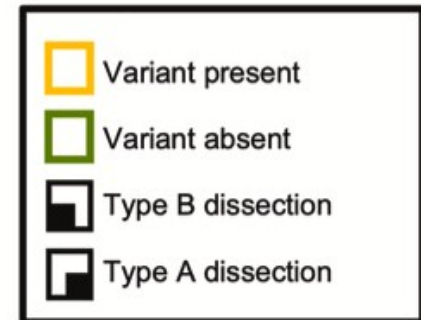
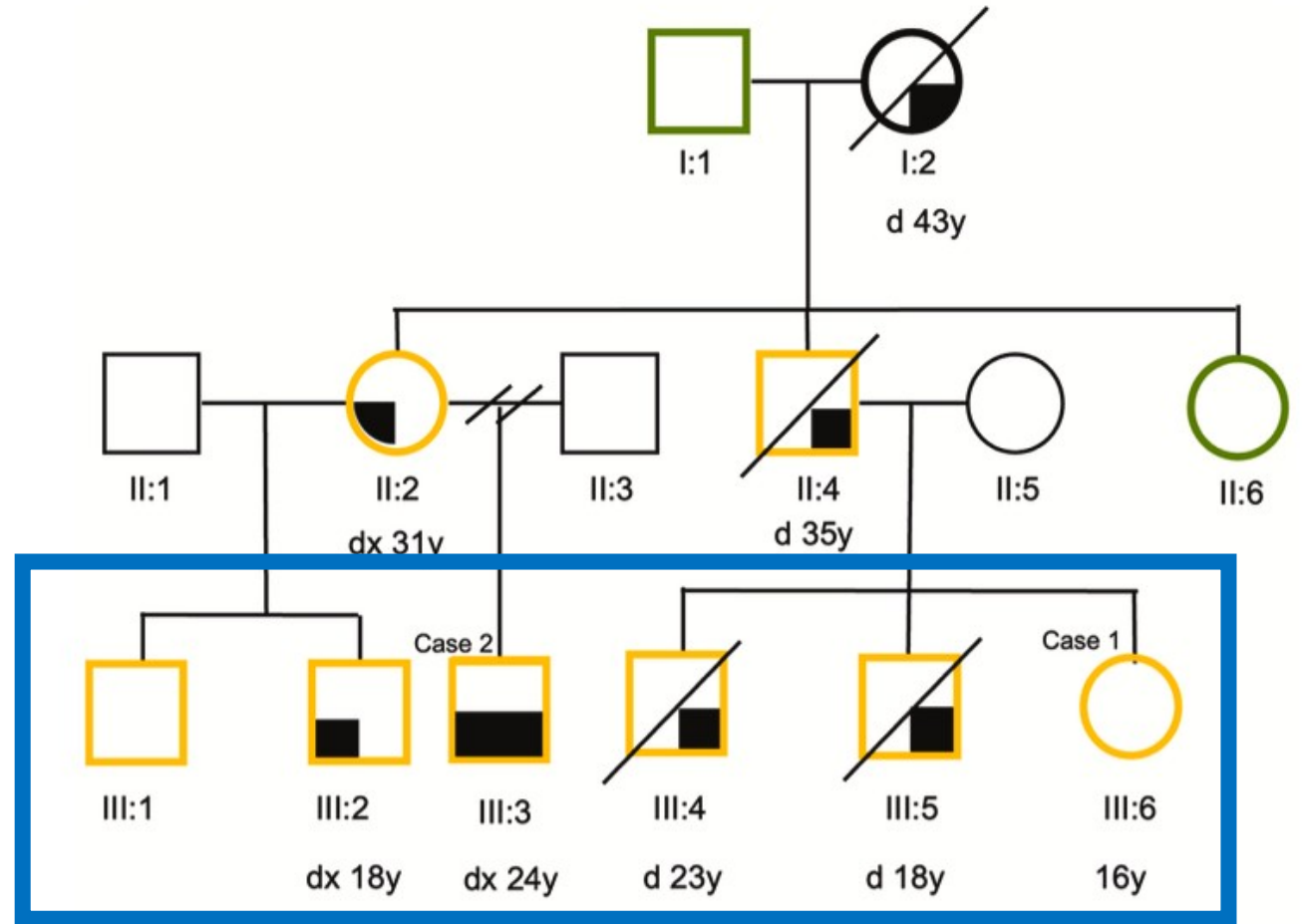
PRKG1 Family⁶



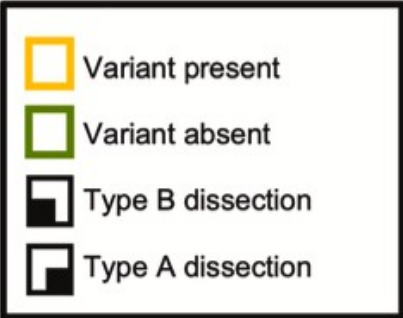
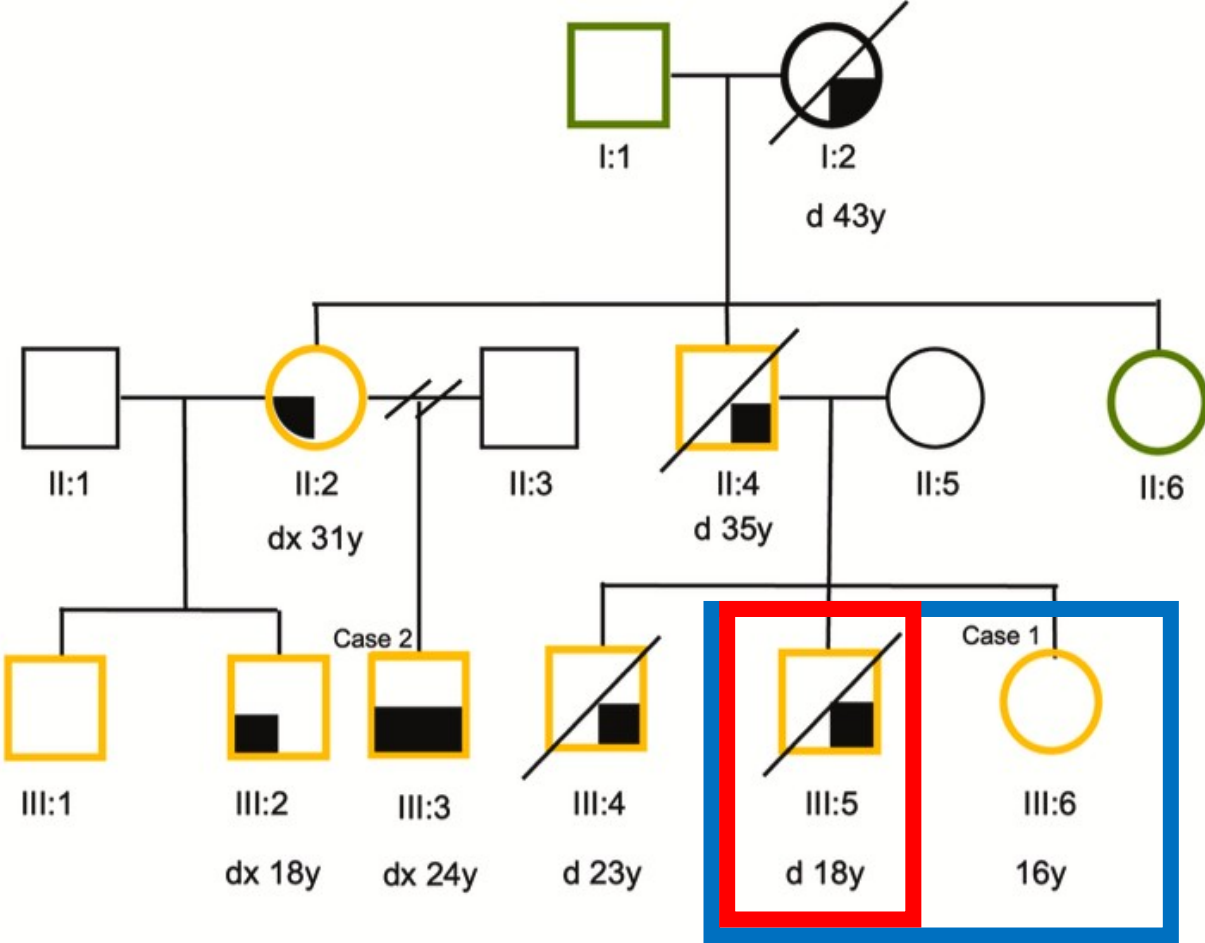
Acute Type
A dissection
at Age 35
led to death



Genetic testing confirmed the presence of the PRKG1 PV in six additional family members

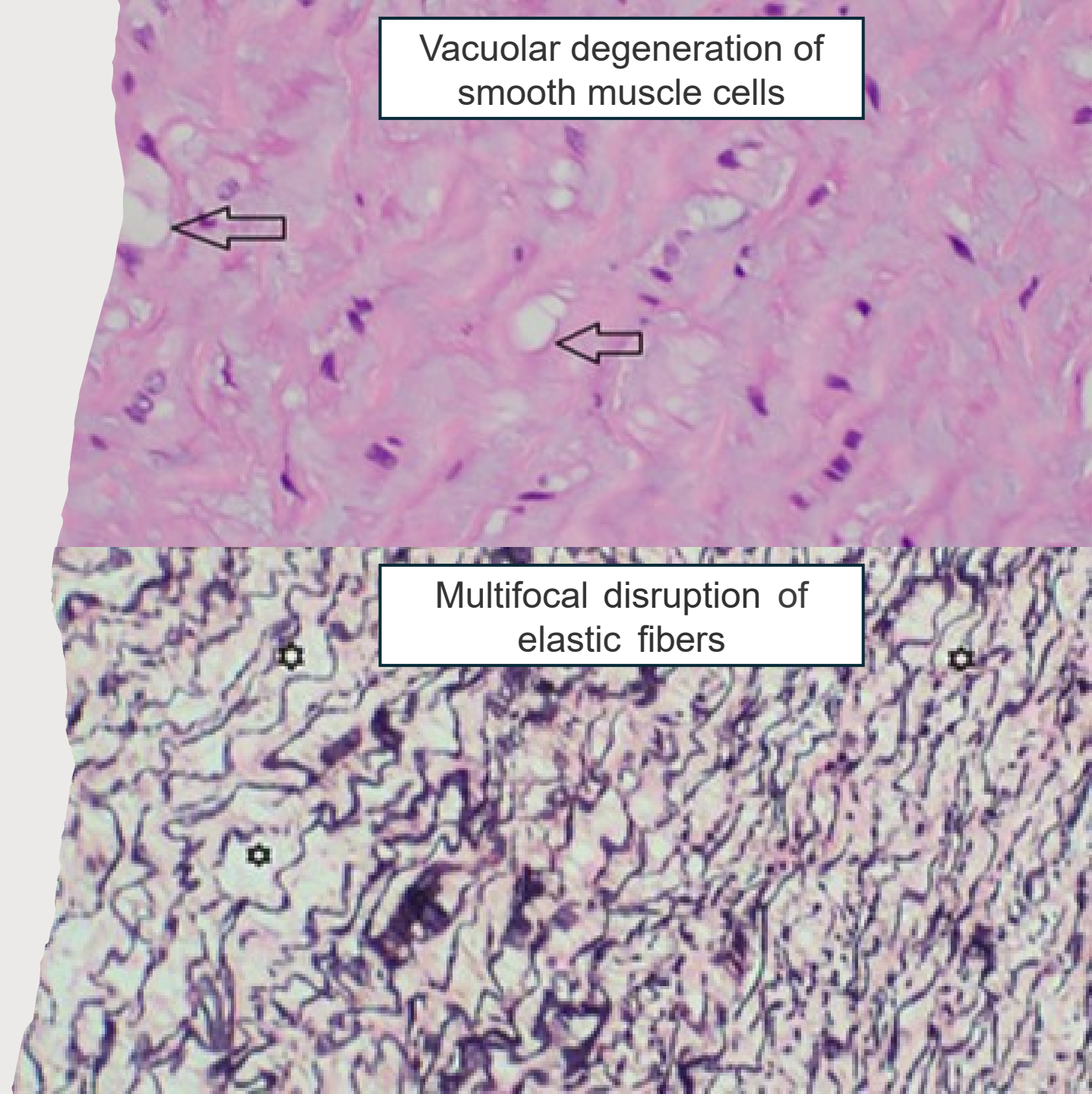


Prophylactic Beta Blocker Therapy to Reduce Aortic Dissection Risk



Proband Family

- 16-year-old female
- Mild dilation: 3.6 cm
- Underwent preventative aortic repair



Take Home Message

Cascade genetic testing & proactive surgical intervention

There is substantial variation in age of onset and severity of PRKG1 aortic disease, even in the same family.

Beta blockers recommended and Lifelong surveillance is mandatory

References

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3. Regalado ES, Morris SA, Braverman AC, et al. Comparative risks of initial aortic events associated with genetic thoracic aortic disease. *J Am Coll Cardiol*. 2022;80:857—869. (4)
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Thank you!