

The Genetics of Aortic Disease

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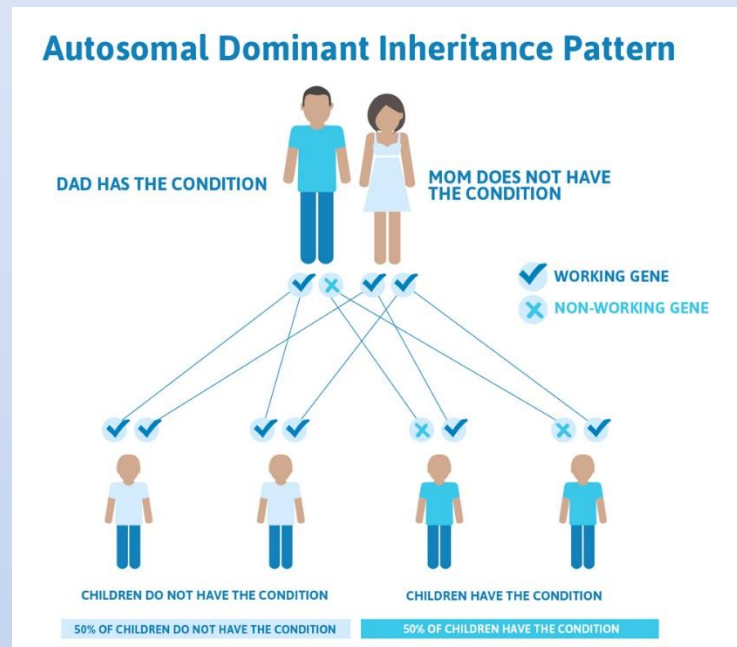
Nothing to Disclose

Overview

- Genetic of Aortic Disease
- Indications for Genetic Referral
- How to Access Genetic Services
- Risk for Family Members

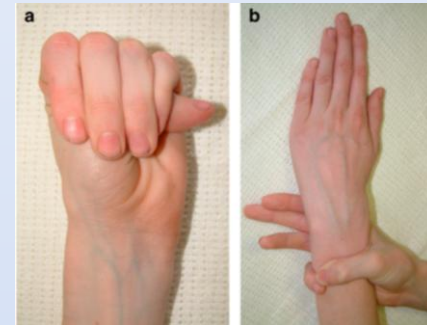
Overview

- Genetic Risk for Thoracic Aortic Disease
 - Syndromic
 - Familial
 - Sporadic
- Autosomal Dominant Inheritance

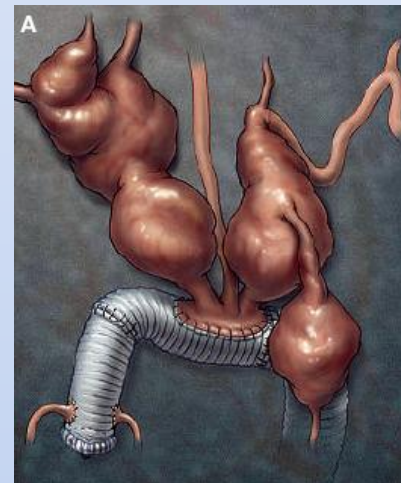


Syndromic vs. Familial

- Marfan Syndrome



- Loeys Dietz Syndrome

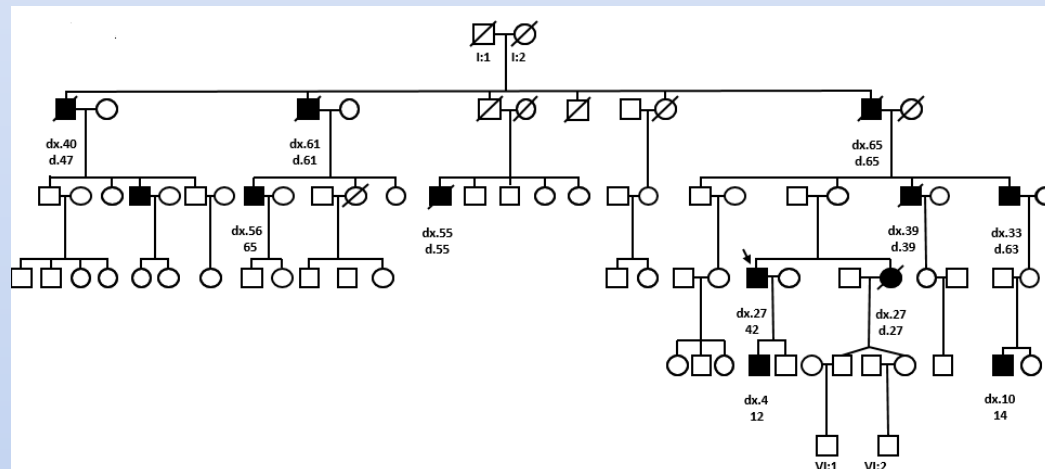


Syndromic vs. Familial

- Vascular Ehlers-Danlos

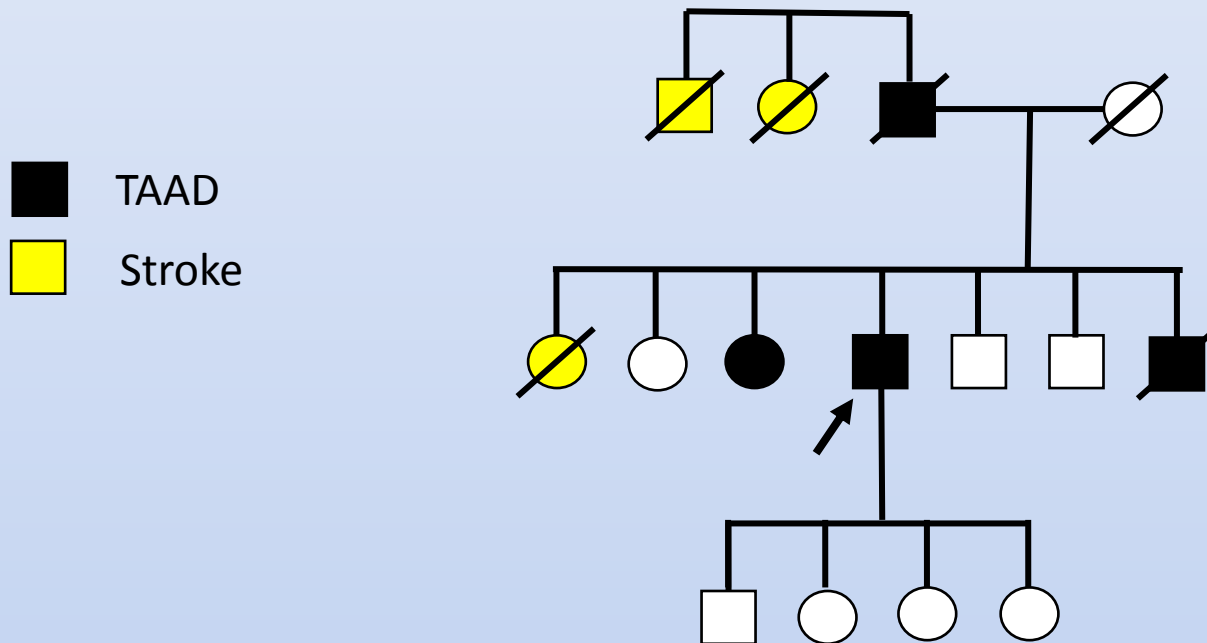


- Familial



Genetics: Why is it important?

Patient: 44 year old with history of a Type A dissection at 36. In hospital for a TAAA repair. History of scoliosis, pectus excavatum and arachnodactyly.



Indications for Genetic Referral

- Features of a possible genetic syndrome (eye, skeletal, joint, other arterial dissections/aneurysms)
- Early onset dissection: <50 years (Stanford Type A or B), including pregnancy
- Family history of aortic disease, arterial dissections and/or sudden cardiac death

How to Access Genetic Services

- Genetics clinics can often facilitate testing for your patients and their families
- Marfan.org clinical directory

Texas

Texas Children's Hospital
Cardiovascular Genetics Clinic
Genetic Aorta Disorders
6621 Fannin Street
Houston, TX 77030

Medical Director:
Shaine A. Morris, MD, MPH

University of Texas Physicians
Medical Genetics and Multidisciplinary Aortic
and Vascular Disease Clinic
6410 Fannin Street, Suite 600
Houston, TX 77030

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* Member, The Marfan Foundation Professional Advisory Board

How to Access Genetic Services

- National Society of Genetic Counselors: Find a Genetic Counselor: nsgc.org



- If no options for a genetics professional, consider ordering testing

Risk for Family Members

- If testing is positive:
 - All first degree family members should be tested. Each have up to a 50% risk of inheriting mutation
- If testing is negative:
 - We know ~30% of the genes responsible for hereditary TAAD
 - 2010 American Heart Association Guidelines recommend screening for TAA in all first degree relatives

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