# PROVIDER INFORMATION SHEET: MARFAN SYNDROME

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## Overview

Those with Marfan syndrome

- Can have significant and variable signs and symptoms that affect their heart/blood vessels, bones/joints, and eyes and overlap with other connective tissue disorders
- Can have manifestations that emerge with age
- Should avoid contact and competitive sports, isometric exercise, and certain medications and substances, such as caffeine, fluoroquinolone antibiotics, and the routine use of decongestants

## When to suspect?

Clinical findings

- Aortic root enlargement (z score ≥2.0)
- Ectopia lentis; most reliably diagnosed by slit-lamp examination after maximal pupillary dilation.
- A systemic score ≥7 (Table 1)
  - Calculation of the systemic score (https://marfan.org/dx/score/)

Family history

- Consistent with autosomal dominant inheritance
- 75% of cases are inherited from an affected parent, 25% are due to new genetic variants

## Possible Next Steps

Referrals

- Cardiologist to evaluate for aortic root enlargement
- Ophthalmologist to assess for ectopia lentis
- Geneticist to determine systemic score
- Genetic counselor to review the benefits, limitations, and risk of genetic testing and to discuss genetic results and their implications
- Perinatologist for any patient who is pregnant or planning a pregnancy

## Resources

- https://marfan.org/conditions/marfan-syndrome/
- https://www.ncbi.nlm.nih.gov/books/NBK1335/#marfan.Suggestive_Findings
- https://www.cdc.gov/heart-disease/about/marfan-syndrome.html

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