



**Marfan Syndrome Diagnosis Survey**  
**Survey Date: February 6-17, 2012**  
**1369 Respondents**

**Diagnosis**

- Nearly 60% of respondents were the first in their family to be diagnosed with Marfan syndrome
- Nearly one-third (28%) were diagnosed after another family member was diagnosed
- 12% were diagnosed after the death of a family member from Marfan syndrome
- Nearly three-quarters (69%) were diagnosed before the age of 20

**Inheritance**

- Nearly half (44%) inherited Marfan syndrome from a parent
- 32% are the only one affected in their family
- One fourth (25%) don't know if they inherited it or not.

**Who First Suspected Marfan Syndrome**

- Cardiologist was the first to suspect Marfan syndrome in 18% of cases.
- Generalist was first to suspect Marfan syndrome in one fourth of cases
  - family doctor 14%
  - pediatrician 11%
- Eye care specialists were the first to suspect Marfan syndrome in nearly 20% of cases
  - ophthalmologists 15%
  - optometrists 4%
- 4% of respondents said that an emergency department doctor was the first to suspect Marfan syndrome
- 8% of respondents said that a nonmedical person was the first to suspect Marfan syndrome

**Which Marfan Features Raised the Suspicion of Marfan Syndrome (respondents were asked to indicate all that apply)**

- Orthopedic characteristics
  - Long limbs – 73%
  - Long flexible fingers – 68%
  - Height – 64%
  - Indented or protruding chest bone – 45%
  - Scoliosis – 33 %
- One-third said that dislocated lens in the eye raised suspicion
- In 18% of people, the first sign that indicated suspicion of Marfan syndrome was aortic dissection