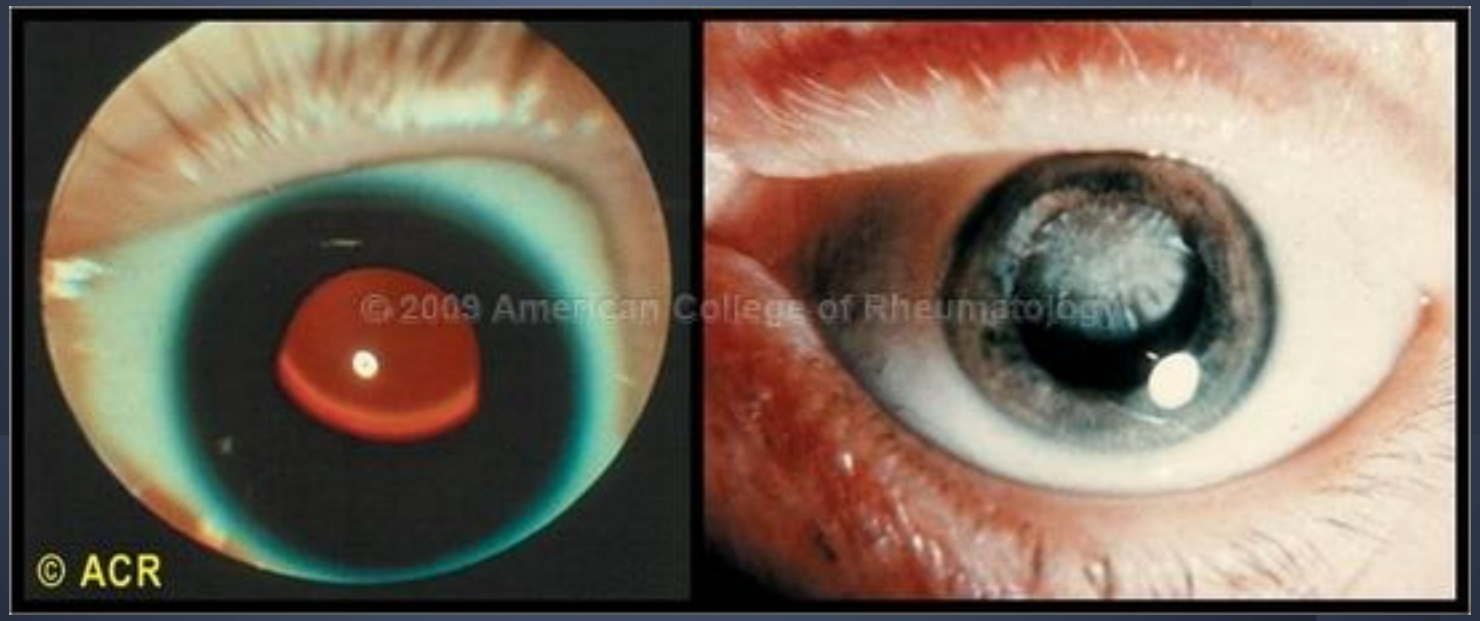
Marfan Syndrome

Diagnosis & Treatment


© ACR


© ACR
© 2009 American College of Rheumatology

What is the classical diagnosis?

Marfan Syndrome Diagnostic Criteria is a list of features doctors use to diagnose (decide if someone has) Marfan syndrome. Diagnostic criteria are sometimes called “Ghent Criteria,” named after the city in Belgium where doctors decided which features to include on the list. Some of these features are easy to see. Others need special tests to find them. These common, painless tests which are part of the clinical evaluation include:

- **Echocardiogram**
- This test looks at the heart, its valves, and the aorta (vessel that carries blood from the heart).
- **Electrocardiogram (EKG)**
- This test checks your heart rate and heart rhythm. Your doctor may to do both an EKG and an echocardiogram.
- **Slit lamp eye exam**
- This test, a part of most eye exams, helps your doctor see if the lenses in your eyes are dislocated (out of place).
- **Other tests, such as a CT scan or MRI of the lower back**
- These tests can help your doctor see if you have dural ectasia, a back problem that is very common in people who have MFS.

What is the standard treatment?

Treatments for the various problems associated with Marfan Syndrome range from surgeries to correct issues like dislocated eye lenses, to medicine to slow the heart rate and help prevent stress on the aorta.

What is the novel, gene-based diagnostic method?

Although scientists have determined that Marfan syndrome (MFS) is caused by a defect in the fibrillin 1 gene on chromosome 15, there is no simple blood test that can conclusively diagnose Marfan syndrome. Therefore, diagnosis is still made through a clinical evaluation.

Did Abraham Lincoln have Marfan Syndrome?