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Early Systolic Dysfunction and Impact of Gene Mutation Severity in Marfan Syndrome

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Background

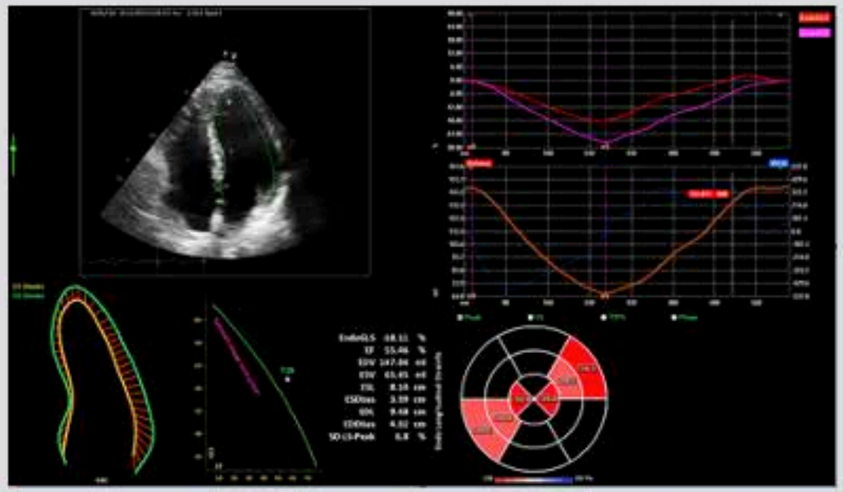
- Marfan syndrome is caused by a mutation in the fibrillin-1 gene that manifests with a variety of features including aortic root dilation.
- Recent research has identified a primary cardiomyopathy in patients with Marfan syndrome, hypothesized to be due to the presence of abnormal fibrillin-1 in the myocardium. Controversy over the nature and significance of this cardiomyopathy remains.
- There is a wide spectrum of severity of this cardiomyopathy, with many patients having no detectable dysfunction and some having pronounced systolic dysfunction without an apparent reason.
- Left ventricular ejection fraction (EF) is increasingly thought to be an insensitive marker for early systolic dysfunction, as the heart is able to preserve a normal LVEF via compensatory mechanisms with mild or early systolic dysfunction.
- Echocardiographic measurement of the first-phase of ejection (defined as the beginning of systole to peak aortic valve flow) may be more sensitive to systolic dysfunction and provide useful clinical information

Purpose

- Assess systolic dysfunction in patients with Marfan syndrome
- Describe first-phase fractional area change (FAC1) in patients with Marfan syndrome and determine how it varies between Marfan syndrome patients and a control group
- Determine if FAC1 varies between mild and severe genetic mutation groups
- Verify there is a difference in propensity for cardiomyopathy in mild vs severe gene mutation Marfan syndrome patients

Methods

- Retrospective review of all patients with Marfan syndrome followed by the Aortopathy Clinic, including clinical, echocardiographic, and genetic records
- Measure traditional echo parameters of LV systolic function as well as the novel FAC1
- Patients were excluded if they had significant mitral or aortic valve regurgitation, prior cardiovascular surgery, or poor echo image quality.



Results

- 126 patients with Marfan syndrome were identified
- 44 were excluded based on significant valve disease
- Of the 82 patients not excluded, genetic testing results are available for 59
- Echocardiographic measurements are underway.

Conclusions

- We have a sizable number of patients which should allow for meaningful results.
- Echocardiographic measurements and statistical analysis will be completed.

Disclosures

None