



## GEN55 - Cardiomyopathy in MFS

**OBJECTIVE:** To establish the association of cardiac dysfunction in Marfan Syndrome patients.

---

### ORGANIZATION

*Lead Investigator:* Jason Cook

*Co-Investigators:* Luca Carta, Cheryl Maslen PhD, Francesco Ramirez DSc

*Funding Source:* Icahn School of Medicine at Mt. Sinai

*Samples:*

- None

*Data:*

- Organ system review
- Medication use
- Imaging

### BACKGROUND AND RATIONALE

Mutations in fibrillin-1 impair the structural integrity of multiple organ systems although a thorough understanding of primary versus secondary manifestations associated with FBN1 mutations is unavailable. Thoracic aortic aneurysm (TAA) with dissection (TAAD) and cardiac valve (CV) dysfunction are common manifestations of MFS and the main causes of morbidity and mortality in affected individuals. Although cardiac abnormalities are generally considered secondary to valve disease, the occasional finding of MFS patients with cardiomyopathy in the absence of valvular problems has suggested that mutations in fibrillin-1 may directly impair cardiomyocyte function.

### CONCLUSIONS

*Results:*

- *Results pending*

### DESIGN

*Method:*

- Establish the frequency of cardiac dysfunction in MFS patient: including EKG abnormalities.
- Characterize the association of cardiac dysfunction with other primary manifestations
- Examine for gender and age related bias in predisposition to heart disease as well as for patients having undergone aortic repair.
- Explore whether frequency of cardiac dysfunction is distinct in patients treated with losartan vs. non-treatment or propranolol.

*Inclusion criteria:*

- Subjects with MFS, FTAAD and other aneurysms <50 years of age.

