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**MENDED HEARTS®** MISSION IS “TO INSPIRE HOPE AND IMPROVE THE QUALITY OF LIFE OF HEART PATIENTS AND THEIR FAMILIES THROUGH ONGOING PEER-TO-PEER SUPPORT, EDUCATION AND ADVOCACY.”
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Mended Hearts: Living With A Genetic Cardiomyopathy

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9/21/2022

Introduction to Cardiomyopathy
Cardiomyopathies

Heart Muscle Disorders

- Normal
- Hypertrophic (HCM)
- Dilated (DCM)
- LV noncompaction (LVNC)
- Arrhythmogenic Cardiomyopathy (AC)

- Age of diagnosis is highly variable
- All associated with heart muscle changes
- Can cause rhythm abnormalities
- Most cases are genetic
Genetic inheritance of cardiomyopathies

- Variants (changes in the DNA) for cardiomyopathies are present on 1 of 2 copies of a particular gene.

- Most variants causing cardiomyopathies have a 50% chance of being passed from parent to child.

- Carrying the variant does not equal developing the disease.

- An identifiable disease-causing gene variant is found in ~50% of patients with a cardiomyopathy.
Yield of Genetic Testing in Cardiomyopathies

- **Hypertrophic (HCM)**: 50-70%
- **Dilated (DCM)**: 40-50%
- **LV noncompaction (LVNC)**: 20-30%
- **Arrhythmogenic Cardiomyopathy (AC)**: 50%
Hypertrophic Cardiomyopathy (HCM)

- Heart muscle thickening (hypertrophy)
- Stiffening and scar tissue formation leads to abnormal relaxation properties of the muscle
- Squeezing force of the left ventricle is normal or greater than normal
- Obstruction to outflow of blood
- Arrhythmias, both atrial and ventricular, can occur
- Heart failure is a late complication (typically decades after diagnosis) for some patients
- Gene variants in the cardiac sarcomere (the heart’s motor)

Most common 1:500 people
Dilated Cardiomyopathy (DCM)

- Enlargement of the left (and sometimes right) ventricular cavity
- Abnormal squeeze and relaxation function of the left ventricle
- Heart failure is a known and frequent complication
- Arrhythmias, both atrial and ventricular, can occur
- Gene variants in a large number of different genes – sarcomere, structural components, cell membrane

1:2,500 people (may be underestimated)
Left Ventricular Noncompaction (LVNC)

- “Spongy” heart muscle more predominant than compact muscle in the mid to tip of the left ventricle than

- Projections of heart muscle into the cavity, in between are deep recesses

- Occurs in isolation or can be associated with DCM, sometimes HCM

- Arrhythmias and heart failure occur in some

- Most genetic variants in same genes as HCM and DCM

? prevalence
Arrhythmogenic Cardiomyopathy (AC)

- Affects right ventricle more than the left ventricle but both can be involved

- Ventricular arrhythmias are more common in AC than in other cardiomyopathies and often out of proportion to the degree of dysfunction of the heart muscle

- Heart failure can occur, particularly with involvement of the left ventricle

- Gene variants in the desmosome – structure that connects heart muscle cells physically and electrically
Diagnosis of Cardiomyopathy

- Many patients are **asymptomatic** at time of diagnosis (e.g. murmur, incidental finding, screening)

![Pie chart showing different methods of diagnosis](chart.png)

Symptoms

- Shortness of breath
- Chest pain
- Syncope/Lightheadedness
- Fatigue
- Palpitations

Routine exam

Family screening

Acute event
Diagnosis of Cardiomyopathy

Echocardiography – mainstay of diagnosis

MRI – high resolution imaging
Goals of Treatment for Cardiomyopathy

- Treat symptoms
- Improve quality of life
- Prevent the condition from progressing
- Prevent sudden death
- Prevent hospitalizations

- Medications
- Surgery, including heart transplant
- Internal defibrillators (ICD)
What Every Patient Should Ask

• What is my ejection fraction?

• Am I on all the medications that have been shown to improve ejection fraction?

• Do I have obstruction to blood flow out of my left ventricle (in the case of HCM)? How can that be improved?

• Do I need to be on blood thinners (for those with atrial fibrillation)?
Summary

• Genetic basis of inherited cardiomyopathy is now largely known

• Genetic testing, combined with clinical evaluation, can be used to identify inheritance patterns and risk in families

• Inherited cardiomyopathies are poised for major scientific advancement
Hypertrophic Cardiomyopathy
Treatment Options for Hypertrophic Cardiomyopathy

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Sept. 27
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Webinar Series

For additional questions, please email: Andrea.baer@mendedhearts.org