Several Genetic Mutations

Associated with Peripartum Cardiomyopathy (PPCM), many of which overlap with those found in other forms of dilated cardiomyopathy (DCM). Research indicates that around 10-20% of PPCM cases have an identifiable genetic component. Some of the key mutations include:

Known Genetic Mutations in PPCM:

1 TTN [Titin gene]

One of the most common mutations found in both PPCM and DCM. Titin is crucial for heart muscle function, and mutations can lead to weakened cardiac muscle.

2 LMNA [Lamin A/C]

Linked to both PPCM and severe forms of cardiomyopathy, often leading to conduction system disease and arrhythmias.

3 MYH7 [Myosin Heavy Chain 7]

Plays a role in heart muscle contraction. Mutations can impair force generation in the heart.

4 PLN [Phospholamban]

Involved in calcium regulation in heart cells. Mutations can cause arrhythmias and worsening heart function.

5 BAG3 [BCL2 Associated Athanogene 3]

Important for maintaining heart muscle cell function under stress. Mutations are linked to heart failure progression.

6 RBM20 [RNA Binding Motif Protein 20]

Regulates splicing of heart muscle proteins. Mutations can contribute to a more severe disease course.

7 DSP [Desmoplakin]

This is the gel-like substance inside the cell where most of the cell's chemical reactions take place.

8 SCN5A [Sodium Voltage-Gated Channel Alpha Subunit 5]

Affects electrical signaling in the heart, increasing the risk of arrhythmias in PPCM patients.