Join the Mission

The PLN North America Foundation has a mission to find a cure and connect affected individuals through support groups, educational resources, and advocacy efforts to push for better treatments and research funding.

TOGETHER WE CAN MAKE A DIFFERENCE

Instagram

YouTube

LinkedIn

Facebook

Facebook Support Group

PLN Cardiomyopathy Support

Zoom Support Group

First Sunday every month at 1:00pm EST
Meeting ID: 733 9348 9503
Password: 9bzXQs

WE ARE HERE FOR YOU

For more information follow our social media accounts for the latest updates, join our monthly newsletter, or email any questions to **plnfoundation.na@gmail.com**

Please visit our website at: **PLNFoundationNA.com**

What is PLN

genetic heart mutation

Phospholamban is a protein that plays an important role in regulating the heart's pumping activity (contraction and relaxation). Abnormalities in phospholamban function lead to cardiac diseases. R14 Del (PLN) is a rare genetic mutation in the phospholamban gene, that impairs its activity, and leads to conditions such as **arrhythmias** (irregular heartbeat and life threatening heart rhythm problems), **cardiomyopathy** (heart becomes weaker and less efficient), and **heart failure**. Identified in 2006, it primarily affects certain populations but has global implications

Current **Treatment Options**

Early identification and management are critical for improving outcomes in affected individuals. Regular follow-up with a cardiologist is essential. Treatment often involves managing symptoms of heart failure and arrhythmias. This may include:

Medications

to stabilize heart rhythm and function

Implantable devices

to prevent life-threatening arrhythmias such as pacemakers, defibrillators, or loop recorders

Advanced care

sometimes required for severe cases such as a heart transplant

IVF

For families with this mutation, In Vitro Fertilization (IVF) with genetic screening offers the option to prevent passing the mutation to the next generation

New therapies, including gene therapy, are being explored. These aim to treat the root cause of PLN R14Del and prevent symptoms all together.

Prognosis

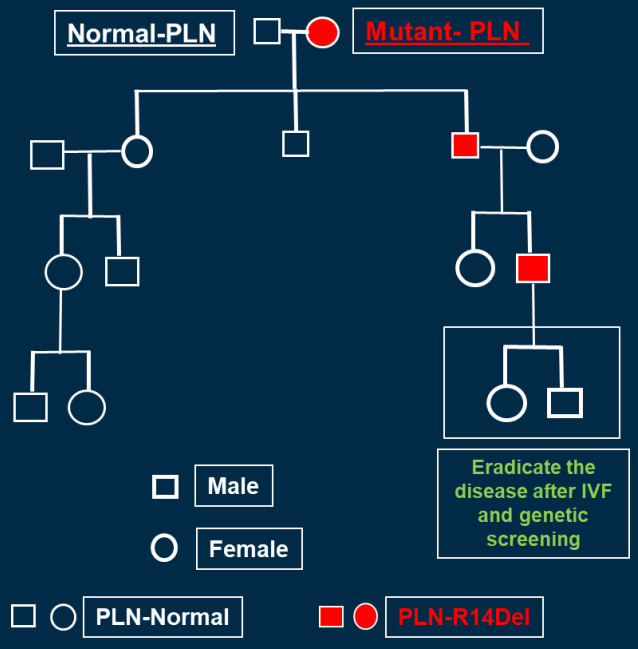
The prognosis for individuals with the PLN R14del mutation depends on several factors, including the extent of cardiac involvement, the presence of arrythmias, and the effectiveness of treatment. Some individuals may retain asymptomatic for many years, while others may develop severe cardiac dysfunction early in life.

Inheritance Risk

PLN R14Del is inherited in an autosomal dominant pattern (an individual only needs one copy of the mutated gene to potentially exhibit symptoms of the disease). A carrier has 50% chance of passing it onto their children. Genetic testing is highly encouraged for early intervention and management

How many carriers?

10% of 20,000 carriers worldwide have been identified. It is vital to identify remaining carriers and alert them to new treatments



Symptoms and Diagnosis

PLN-R14Del is confirmed through genetic testing. Early detection is key to managing health effectively. Symptoms seldomly occur during childhood and usually arise later in life. Symptoms include:

Irregular Rhythms

palpitations or fluttering (heart racing or skipping beats), ventricular tachycardia (life threatening fast/irregular heart beat) or sudden cardiac death

Shortness of Breath

reduced exercise tolerance (getting tired during physical activity)

Fatigue

weakness during activities and dizziness

Heart Failure

chest pain and swelling in the legs

FREE genetic testing resources:

SCAN ME