LMNA Cardiac Disease Letter for Patients and Treating Physicians

Written by Alfredo Hernandez 2015 - Updated 2026 - An LMNA+ Patient & Heart Transplant Recipient.

Attention: Family Members and Treating Physicians

Below is some information that I have written for many family members to share with their treating physicians. It has helped bring some understanding regarding the inherited genetic mutation, **LMNA**, that has been causing cardiac disease in many of our family members for generations.

Over the last several years, we have learned a great deal about LMNA. Unfortunately, until recently, little was known about what was causing several of our loved ones to suffer from very serious cardiac problems and early mortalities.

These conditions have included chronic atrial arrhythmias, life-threatening ventricular arrhythmias, progressive non-ischemic cardiomyopathy, congestive heart failure, strokes, the need for multiple cardiac procedures, and tragically premature deaths from sudden cardiac death (SCD) and advanced heart failure requiring heart transplantation,

Understanding LMNA Cardiac Disease

LMNA-related cardiomyopathy is a genetic condition caused by mutations in the LMNA gene, which encodes the lamin A/C proteins. These proteins help maintain the structure and function of cell nuclei. LMNA-related cardiac disease usually presents in early to mid-adulthood with symptomatic conduction system disease leading to arrhythmias, heart failure, non-ischemic cardiomyopathy, stroke, and in some individuals, the need for heart transplantation or sudden cardiac death which in some instances can be the presenting manifestation.

LMNA gene mutations can manifest disease in many different ways, including Lipodystrophies, Progeroid syndromes, Muscular Dystrophies, Neuropathies, Cardiac Conduction Disorders, and Cardiomyopathies. These conditions are collectively referred to as laminopathies.

Why Awareness Matters

Many patients initially present with seemingly isolated findings such as palpitations, atrial arrhythmias, dizziness, fainting episodes, persistant conduction abnormalities or unexplained cardiomyopathy. Often, there is also a strong family history of pacemaker implants and cardiac-related early mortalities. **Because LMNA disease is relatively rare, diagnosis is often delayed, allowing progression of cardiac disease before appropriate surveillance and treatment strategies are implemented. Under these circumstances, strong consideration should be given to genetic testing of known inherited cardiac diseases.**

Clinical Features Commonly Seen in LMNA Disease

Patients may develop SA/AV node dysfunction, bradycardia, chronic atrial arrhythmias, complete AV block, ventricular arrhythmias, progressive heart failure, dilated cardiomyopathy (DCM), and an increased risk of thromboembolic events, including stroke. Disease severity and rate of progression vary considerably among affected individuals and families.

Approximately 20% of LMNA+ patients may require heart transplantation from LMNA-related Dilated Cardiomyopathy due to the rapid progression of end-stage heart failure.

Considerations for Treating Physicians

Published evidence and clinical experience suggest that patients with LMNA-Related Cardiomyopathy often require close monitoring, including serial ECGs, ambulatory rhythm monitoring, annual echocardiography, and when appropriate, cardiac MRI. Due to the progressive nature of the disease, standard pacing therapies may not provide adequate long-term protection against disease progression and associated complications. **Early consideration of implantable cardioverter-defibrillator (ICD) therapy has been recommended for all indications and advanced heart failure management may be appropriate in selected patients based on individual risk factors and current clinical guidelines.**

Current Expert Recommendations

Based on what is currently known about LMNA Cardiac Disease, in appropriately selected patients with LMNA-associated cardiac disease and progressive cardiac involvement, CRT-D implantation should be strongly considered. When combined with guideline-directed medical therapy, a CRT-D has shown to improve cardiac function, reduce heart failure symptoms, and of course, provide protection against life-threatening ventricular arrhythmias through ICD therapy.

"Risk of sudden cardiac death may be higher in LNMA gene mutation carriers as compared to other genetic cardiomyopathies, and if pacemakers are being considered clinically, then there are data that ICD-Pacers may be superior to standard pacemakers alone".

Matthew Taylor MD, PhD. Medical Geneticist, University of Colorado Anschutz Medical Campus.

To date, there is no cure for LMNA Cardiac Disease. While these therapies can improve clinical outcomes and quality of life, many patients continue to experience ongoing symptoms as the disease progresses. When combined with appropriate medication therapy, this approach may help reduce the risk of sudden cardiac death, improve the patient's quality of life and longevity, reduce hospitalizations, decrease the need for additional cardiac interventions, and help stabilize cardiac function. In some patients, this may delay the need for advanced heart failure therapies, including heart transplantation.

Cardioversions and Ablation Procedures

While these procedures have proven to be effective in treating many abnormal heart rhythms and cardiac conduction disorders, the durability in LMNA-associated cardiac disease may be limited by the progressive nature of the disease process. Recurrence of arrhythmias is common, and the potential benefits of repeat ablation procedures should be weighed against the risk of additional atrial scarring and procedural burden.

Family Screening

Since LMNA disease is typically inherited in an autosomal dominant pattern, family screening and genetic counseling are strongly encouraged. Identification of affected relatives may allow earlier surveillance and intervention before significant symptoms develop.

My Personal Experience with this Disease

At the age of 39, while training for a half marathon, I began to develop sudden and unexpected cardiac problems despite my good physical fitness and healthy lifestyle.

My symptoms initially began with the onset of atrial arrhythmias, which rapidly progressed to bradycardia and eventually complete heart block with a HR of 20 BPM. This led me to initially needing an ablation procedure followed by the emergency implantation of a standard dual-lead pacemaker. ***All occurring within a period of 10 months.***

Despite these efforts, my cardiac problems continued to progress rapidly. I underwent a heart catheterization to rule out ischemic disease, multiple cardioversions, additional ablations and various heart medications in an attempt to manage my persistent symptomatic atrial arrhythmias.

Despite these interventions, my heart function began to rapidly decline with the development of congestive heart failure from non-ischemic cardiomyopathy requiring additional cardiac procedures and medications due to ongoing complications, unresolved atrial fibrillation, and eventually life-threatening ventricular arrhythmias. My quality of life rapidly declined significantly despite all treatment efforts. ***All this occurred within 2 years from the onset of initial symptoms.***

Desperate to find answers, I began to search for someone who could help me understand what was happening to my heart and why. Ultimately, during one of my many visits to the emergency room, I was very fortunate to meet an electrophysiologist in my home town, Albuquerque, New Mexico, who would help me determine the reasons for my sudden, unexpected, and very progressive form of heart disease.

Genetic Testing and Diagnosis

My electrophysiologist decided to perform genetic testing of known inherited cardiac diseases, which would reveal that my heart problems had been due to a rare autosomal dominant LMNA gene mutation. A mutation of the Lamin A/C gene located on chromosome one.
The genetic mutation that was detected is as follows:
LMNA c.1057 C>T missense predicting a Gln353Stop (Q353X) mutation in the lamin a/c protein.

I received my CRT-D device soon after testing positive for this LMNA gene mutation. The implantation of the CRT-D device combined with the appropriate medication management did slow the progression of my heart failure and improved my quality of life. But most importantly, if it had not been for my ICD delivering lifesaving shock therapies, I would have died of sudden cardiac death from multiple episodes of ventricular fibrillation. This provided me with the lifesaving bridge I needed until ultimately receiving a heart transplant on March 31, 2015.

Family History

Within my family, this mutation has predominantly manifested with progressive cardiac conduction system disorders such as bradycardia; complete AV heart block; pacemaker implantation; chronic atrial fibrillation; non-ischemic cardiomyopathy; and, in some instances, the onset of potentially lethal ventricular arrhythmias and severe heart failure from Dilated Cardiomyopath (DCM).

Initial symptoms usually manifest with heart palpitations, fatigue, dizziness, and episodes of syncope. Upon examination we see abnormal ECGs and Holter Monitor results may indicate 1st.--2nd.deg. AV block from AV node dysfunction, which eventually lead to complete heart block.

To date, over 50% of those in my extended family have tested positive for this LMNA gene mutation, which is consistent with autosomal dominant mutations. Among these, 100% of those needing a pacemaker implant due to AV Block, have tested positive for LMNA, indicating the primary cause of heart disease in our family.

Some family members have also tested positive for other known inherited cardiac gene mutations classified as Long QT syndrome mutations, KCNE2 and SCN5A mutations, which can also lead to conduction disorders but are not suspected to be the main disease-causing mutations affecting my family members.

These findings have led many affected family members to receive standard pacemakers, which are often considered an appropriate treatment for these conditions. However, long-term observation of multiple family members, together with my personal experience, this approach has not adequately addressed the progressive nature of our LMNA-associated cardiac conduction disease.

The problem we have seen with this approach and what we now know about LMNA related cardiac conduction system disease is that we eventually see continued progression of persistent arrhythmias such as atrial flutter, atrial fibrillation, supraventricular tachycardias, thromboembolic complications, ischemic cardiac events, strokes, associated deaths and the onset of life threatening ventricular arrhythmias indicating a significant risk of sudden cardiac death (SCD).

This has required us to undergo many more cardiac procedures such as cardioversions and multiple ablations, which have demonstrated to be a very temporary solution. ***Based on our family's clinical experience, repeated ablation procedures have provided a very temporary benefit and have contributed to additional atrial scarring over time.***

Eventually, we see the progression of this disease with the onset of heart failure, non-ischemic cardiomyopathy, and in some instances the advanced stages of LMNA-related DCM requiring cardiac transplantation.

Much was learned through my experiences with LMNA-related cardiac disease, and many of my family members who have received this type of early intervention after diagnosis have demonstrated improved outcomes and quality of life. Several have received life-saving shock therapies from their CRT-D devices due to sudden episodes of ventricular fibrillation.

Closing Thoughts

Due to the aggressive expression of this LMNA gene, and what we now know about LMNA-related cardiac disease, my hope in sharing this letter is to encourage awareness, support informed clinical decision-making, and help patients receive timely evaluation and diagnosis. This approach incorporates current evidence-based treatment strategies designed to improve clinical outcomes, reduce the need for additional cardiac procedures, prolong survival, and enhance quality of life.

Sincerely,

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