



Barth Syndrome

Cardiovascular System

Left Ventricular Non-Compaction (LVNC)



Barth Syndrome
Foundation

Cardiomyopathy is a chronic disease of the heart muscle. It affects the heart's pumping ability. It is not uncommon for cardiomyopathy to wax (increase in size) and wane (decrease in size) with Barth syndrome. Those who have Barth syndrome may be diagnosed with dilated (thinning of heart walls) and/or hypertrophic (thickening of the heart walls) forms of cardiomyopathy. Both forms cause an enlarged heart and affect the heart's ability to pump. Cardiomyopathy results in something known as heart failure. Congestive heart failure refers to fluid build-up around the heart.

Non-compaction is often referenced as **left ventricular non-compaction cardiomyopathy (LVNC)**. As the heart develops the tissue compacts to become a network of smooth cardiac muscle fibers that allows the heart to expand (fill with blood) and contract (squeeze the blood out). LVNC occurs when the heart muscle cells within the left ventricle do not tightly bind as they should while a child is in the womb. During development, the heart muscle is a sponge-like, non-compacted network of muscle fibers. As normal development progresses, the muscle fibers bind to form the thick cardiac wall of the heart. The transformation from the heart going from having a sponge-like appearance to a smooth and solid heart wall is known as the remodeling phase. LVNC occurs when this remodeling process is interrupted. The non-compacted trabeculations are most often observed at the bottom of the heart, which is called the apex. It has also been observed elsewhere in the left ventricle and in the right ventricle of BTHS explanted hearts.

Approximately 50% of the individuals with BTHS have been reported as having LVNC. It can occur in combination with dilated or hypertrophic cardiomyopathy. It is not always apparent at birth. An EKG/ECG (electrocardiogram) is used to look at the heart's electrical impulses to assess risk of arrhythmia. Color Doppler ECHO (echocardiogram) and MRI (magnetic resonance imaging) show the flow of blood through the heart and to identify the presence of the non-compacted trabeculations.

LVNC affects the normal electrical signaling of the heart which can cause heart rhythm disturbances potentially leading to sudden cardiac arrest. It can also reduce how well the heart can pump blood which can lead to heart failure. Blood clot formation within the trabeculations lead to increased risk of thrombotic stroke. It is important for anyone with BTHS to be followed by a cardiologist with experience in caring for individuals with cardiomyopathy.

Other Cardiovascular Risks of Barth Syndrome

- **Cardiomyopathy:** Chronic disease of the heart muscle in various forms
 - Dilated cardiomyopathy: Enlarged heart with thinning and stretched heart muscle
 - Hypertrophic cardiomyopathy: Stiff and thick heart muscle
 - Endocardial fibroelastosis: Thick heart muscle due to an increase of supporting tissue and elastic fibers
- **Thrombosis:** Blood clots
- **Heart Failure:** Severe failure of the heart to function properly
- **Arrhythmia:** Irregular Heartbeats
- **Repolarization Abnormalities:** Irregular electrical signaling when the heart is relaxing following the heart's contraction.
 - ST flattening, T-wave inversion
- **Autonomic Instability**
- **Acute Decompensation**