Abstract Kristina Haugaa

Genetic cardiomyopathies are inherited and caused by genetic mutations. These mutations affect the structure and function of the heart muscle, leading to various types of cardiomyopathies.

The new ESC guidelines for cardiomyopathies focus on the morphologic traits to classify the cardiomyopathies.
There main types of genetic cardiomyopathies are:

1. Hypertrophic cardiomyopathy (HCM)
2. Dilated cardiomyopathy (DCM)
3. Non dilated left ventricular cardiomyopathy (NDLVC)
4. Arrhythmogenic right ventricular cardiomyopathy (ARVC)
5. Restrictive cardiomyopathy (RCM)

Genetic cardiomyopathies can be caused by mutations in various genes that are involved in the structure and function of the heart muscle. The inheritance pattern is most often autosomal dominant.
Diagnosis of genetic cardiomyopathies involves a thorough evaluation of a patient's medical history, physical examination, electrocardiogram (ECG), echocardiogram, CMR and genetic testing.
In the new 2023 ESC guidelines for cardiomyopathies, genetic testing is recommended in patients fulfilling diagnostic criteria for cardiomyopathy.
It is important for individuals with a family history of genetic cardiomyopathies to undergo genetic counseling and testing, as early detection and management can help improve outcomes and prevent complications.
Lamin A/C cardiomyopathy is a specific subtype of genetic cardiomyopathy caused by mutations in the LMNA gene. It is characterized by early onset, high penetrance (meaning a high likelihood of the mutation causing disease), an increased risk of ventricular tachycardia (VT), and a high likelihood of requiring heart transplantation. Lamin A/C cardiomyopathy is considered a malignant form of cardiomyopathy due to its potential for severe complications.
High risk genes include LMNA, FLNC-truncating variants, TMEM43, PLN, DSP, RBM20

In summary:
Genetic dilated cardiomyopathy should not be considered as a specific entity but as different genetic diseases. Genetic testing should be performed in patients fulfilling a cardiomyopathy phenotype.
Genotype specific management is required, in particular in the high risk genes:
LMNA, FLNC-truncating variants, TMEM43, PLN, DSP, RBM20
These patients require other risk stratification for primary prevention ICD than the traditional EF<35%. Primary prevention ICD should be considered already when EF < 45-50%