Gene	Protein	Inheritance	Disease Association
DES	Desmin	AD, AR	DCM, ARVC, myofibrillar myopathy, RCM with AV block, neurogenic scapuloperoneal syndrome Kaeser type, LGMD
DSC2	Desmocollin	AD, AR	ARVC, ARVC plus skin and hair findings
DSG2	Desmoglein	AD	ARVC
DSP	Desmoplakin	AD, AR	ARVC, DCM, Carvajal syndrome
JUP	Junction plakoglobin	AD, AR	ARVC, Naxos disease
PKP2	Plakophilin 2	AD	ARVC
RYR2	Ryanodine receptor 2	AD	ARVC, CPVT, LQTS
TMEM43	Transmembrane protein 43	AD	ARVC, EMD
TTN	Titin	AD, AR	HCM, DCM, ARVC, myopathy

Table 3. Genes included in the Arrhythmogenic Cardiomyopathy Multi-Gene Panel

Abbreviations: Hypertrophic cardiomyopathy (HCM), dilated cardiomyopathy (DCM), arrhythmogenic right ventricular cardiomyopathy (ARVC), restrictive cardiomyopathy (RCM), limb-girdle muscular dystrophy (LGMD), Emory muscular dystrophy (EMD), catecholaminergic polymorphic ventricular tachycardia (CPVT), long QT syndrome (LQTS), autosomal dominant (AD), autosomal recessive (AR)

disease-specific test. This test may also be helpful when the clinical diagnosis is not clear, or when there is more than one form of cardiomyopathy in the family history. It is important to note that the number of variants of uncertain significance detected by this panel may be higher than for the disease-specific panels, making clinical correlation more difficult. See Table 4 for details regarding the genes tested by CCMGP / Comprehensive Cardiomyopathy Multi-Gene Panel, Blood and the conditions associated with them.

Conclusion

Inherited cardiomyopathies are some of the most common genetic disorders and are a major cause of heart disease in all age groups, often with an onset in adolescence or early adult life. All of the inherited cardiomyopathies are genetically heterogeneous with multiple associated genes and several different mutations within each category. Genetic testing can play an important role in the confirmation of the diagnosis of cardiomyopathy, appropriate medical management decisions, and more informed genetic counseling for patients and families. Advances in the discovery of the genetic basis of the various forms of cardiomyopathy may serve to guide clinical practice and raise expectations for new forms of novel treatment.