TABLE 287-3 SELECTED GENETIC DEFECTS ASSOCIATED WITH CARDIOMYOPATHY

	Gene Product	Inheritance	Cardiac Phenotype	Isolated Cardiac Phenotype	Extracardiac Manifestations
Sarcomere	*MYH7* (β myosin heavy chain)	AD	HCM, DCM, LVNC	Yes	Skeletal myopathy
	MYBPC3 (myosin binding protein C)	AD	HCM	Yes	
	TNNT2 (cardiac troponin T)	AD	HCM, DCM, LVNC	Yes	
	TNNI3 (cardiac troponin I)	AD, AR	HCM, DCM, RCM	Yes	
	TTN (Titin)	AD	DCM	Yes	
	TPM1 (α-tropomyosin)	AD	HCM, DCM	Yes	
	TNNC1 (cardiac troponin C)	AD	DCM	Yes	
	ACTC (α-actin)	AD	HCM, DCM, (LVNC)	Yes	
	MYL2 (myosin regulatory light chain)	AD	HCM	Yes	Skeletal myopathy
	MYL3 (myosin essential light chain)	AD	HCM	Yes	
Z-disk and Cytoskeleton	*DES* (Desmin)	AD	DCM, RCM	Yes	Skeletal myopathy
	LDB3 (Cypher-ZASP)	AD	DCM, LVNC	Yes	Skeletal myopathy
	MYOZ2 (Myozenin)	AD	HCM	Yes	
	TCAP (Telethonin)	AD	DCM, HCM	Yes	
	ANKRD1 (CARP)	AD	HCM, (DCM)	Yes	
	CSRP3 (MLP)	AD	DCM, (HCM)	Yes	
	ACTN2 (α-actinin-2)	AD	DCM	Yes	
	CRYAB (αB-crystallin)	AD	DCM	Yes	
Nuclear Membrane	*LMNA* (Lamin A/C)	AD, AR	CDDC	Yes	Skeletal myopathy
	EMD (Emerin)	X-linked	CDDC	No	Skeletal myopathy, contractures
Excitation-Contraction Coupling	*PLN* (Phospholamban)	AD	DCM	Yes	
	SCN5A (NAV 1.5)	AD	CDDC	Yes	Note other mutations associated with Brugada syndrome
	RYR2 (cardiac ryanodine receptor)	AD	ARVC	Yes	
	CASQ2 (calsequestrin 2)	AR	ARVC	Yes	
Cellular Metabolism	*PRKAG2* (γ-subunit of AMP kinase)	AD	HCM+	Yes	
	LAMP2 (lysosomal associated membrane protein)	X-linked	HCM+	No[a]	Danon's disease: skeletal myopathy, cognitive impairment
	TAZ (Tafazzin)	X-linked	DCM, LVNC	No	Barth's syndrome: skeletal myopathy, cognitive impairment, neutropenia
	FXN (Frataxin)	AR	HCM	No	Friedreich's ataxia: ataxia, diabetes mellitus type 2
	TMEM43 (transmembrane protein 43)	AD	ARVC	Yes	
	GLA (α-galactosidase-A)	X-linked	HCM+	Yes	Fabry's disease: renal failure, angiokeratomas and painful neuropathy
Mitochondria	Mitochondrial DNA	Maternal transmission	DCM, HCM	No	MELAS, MERRF, Kearns-Sayre syndrome, ocular myopathy
Sarcolemmal Membrane	*DMD* (Dystrophin)	X-linked	DCM	No[a]	Duchenne's and Becker's muscular dystrophy
	DMPK (dystrophica myotonica protein kinase)	AD	DCM	No	Myotonic dystrophy type 1
	SGCD (δ-sarcoglycan)	AD	DCM	Yes	
Desmosome	*DSP* (Desmoplakin) *JUP* (Plakoglobin)	AD, AR	ARVC	Yes	Carvajal syndrome (AR), Naxos syndrome (AR), "woolly hair" and hyperkeratosis of palms and soles
	DSG2 (Desmoglein 2) *DSC2* (Desmocollin 2) *PKP2* (Plakophilin 2)	AD	ARVC	Yes	
Other Examples	*RBM20* (RNA binding motif 20)	AD	DCM	Yes	
	PSEN1 (Presenilin-1,2)	AD	DCM	Yes	Dementia
	BAG3 (BCL2-associated athanogene 3)	AD	DCM	Yes	

[a]Indicates that isolated cardiac phenotype can occur in women with the X-linked defects.

Abbreviations: AD, autosomal dominant; AR, autosomal recessive; ARVC, arrhythmogenic right ventricular cardiomyopathy; CDDC, conduction disease with dilated cardiomyopathy; DCM, dilated cardiomyopathy; HCM, hypertrophic cardiomyopathy; HCM+, HCM with preexcitation; HCMc, HCM with conduction disease; LVNC, left ventricular noncompaction; MELAS, (mitochondrial) myopathy, encephalopathy, lactic acidosis, and strokelike episodes syndrome; MERRF, myoclonic epilepsy with ragged red fibers; RCM, restrictive cardiomyopathy.