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CardioNext® Genes and Associated Diseases | April 2020

CardioNext is a targeted panel for patients with inherited cardiomyopathies, arrhythmias, and other inherited cardiovascular conditions. Given the genetic and clinical overlap between these conditions, one comprehensive inherited cardiovascular test is an effective way of identifying at-risk individuals, or confirming a diagnosis.

GENE NAME	ASSOCIATED DISEASE
ABCC9	DCM
ACTC1	Congenital Heart Defects, DCM, HCM, LVNC
ACTN2	DCM, HCM
AKAP9	LQTS
ALMS1	Alström syndrome (DCM)
ALPK3	HCM
ANK2	Arrhythmia
ANKRD1	DCM, HCM
BAG3	DCM
CACNA1C	BrS, Timothy syndrome
CACNA2D1	BrS
CACNB2	BrS
CALM1	CPVT, LQTS
CALM2	LQTS
CALM3	LQTS
CASQ2	CPVT
CAV3	HCM, LQTS
CRYAB	Cardiomyopathy
CSRP3	DCM, HCM
DES	Cardiomyopathy
DMD	Dystrophinopathy (DCM)
DOLK	DCM
DSC2	ARVC
DSG2	ARVC, DCM
DSP	ARVC, DCM
EMD	Emery-Dreifuss Muscular Dystrophy (Cardiomyopathy)
EYA4	DCM
FHL1	Emery-Dreifuss Muscular Dystrophy (HCM)
FKRP	Limb-Girdle Muscular Dystrophy (DCM)
FKTN	DCM
FLNC	DCM, HCM
GATAD1	DCM
GLA	Fabry Disease (HCM)
GPD1L	BrS
HCN4	Cardiomyopathy, BrS
JPH2	HCM
JUP	ARVC
KCND3	BrS
KCNE1	LQTS
KCNE2	LQTS
KCNE3	BrS
KCNH2	LQTS, Short QT syndrome
KCNJ2	Andersen-Tawil syndrome, Short QT syndrome
KCNJ5	Andersen-Tawil syndrome, LQTS
KCNJ8	BrS
KCNQ1	LQTS, Short QT syndrome
LAMA4	DCM

GENE NAME	ASSOCIATED DISEASE
LAMP2	Danon Disease (HCM, DCM)
LDB3	DCM, LVNC
LMNA	Arrhythmia, Cardiomyopathy, Cardiac Conduction Disease, DCM
MYBPC3	DCM, HCM, LVNC
MYH6	Congenital Heart Defects, DCM, HCM
MYH7	DCM, HCM, LVNC
MYL2	HCM
MYL3	HCM
MYOZ2	HCM
MYPN	DCM, HCM
NEXN	DCM, HCM
NKX2-5	Arrhythmia, Cardiomyopathy, Congenital Heart Defects
PKP2	ARVC
PLN	ARVC, DCM, HCM
PRKAG2	HCM, Wolff-Parkinson-White syndrome (Arrhythmia)
PTPN11	Noonan syndrome (HCM)
RAF1	Noonan syndrome (DCM, HCM)
RBM20	DCM
RIT1	Noonan syndrome (HCM)
RYR2	ARVC, CPVT
SCN1B	BrS
SCN2B	BrS
SCN3B	BrS
SCN4B	LQTS
SCN5A	ARVC, BrS, DCM, LQTS
SCN10A	Arrhythmia, BrS
SNTA1	LQTS
SOS1	Noonan syndrome (HCM)
TAZ	Barth syndrome (Cardiomyopathy)
TBX20	Congenital Heart Defects, DCM
TBX5	Holt-Oram syndrome (Congenital Heart Defects, Cardiac Conduction Disease)
TCAP	DCM, HCM
TECRL	Arrhythmia, CPVT
TGFB3	ARVC, Loeys-Dietz syndrome
TMEM43	ARVC
TNNC1	DCM, HCM
TNNI3	DCM, HCM
TNNT2	DCM, HCM, LVNC
TPM1	DCM, HCM, LVNC
TRDN	CPVT, LQTS
TRPM4	Arrhythmia, BrS
TTN	DCM
TTR	Amyloidosis (Cardiomyopathy, Arrhythmia)
TXNRD2	DCM
VCL	DCM, HCM

References available upon request.
HCM = Hypertrophic Cardiomyopathy, DCM = Dilated Cardiomyopathy, ARVC = Arrhythmogenic Right Ventricular Cardiomyopathy, LVNC = Left Ventricular Non-Compaction, CPVT = Catecholaminergic Polymorphic Ventricular Tachycardia, LQTS = Long QT Syndrome, BrS = Brugada Syndrome

Genetic Testing Impacts Medical Management

Identifying a genetic etiology for patients with a clinical diagnosis of a cardiomyopathy or arrhythmia can have direct implications for that patient's medical management. Use the guide below to learn more about professional guideline recommendations for the management of patients based on their genotype.

Genes and Treatment Considerations

TAILORED THERAPIES[1,2,5]

GLA	*KCNQ1*	*TTR*
KCNH2	*SCN5A*	

CONSIDER EARLIER ICD IMPLANTATION*[,2,3]

DES	*LMNA*	*RBM20*	*TMEM43*
FLNC	*PLN*	*SCN5A*	

MEDICAL SPECIALIST REFERRALS

ALMS1	*GLA*	*RIT1*
CACNA1C	*KCNJ2*	*SOS1*
DMD	*KCNJ5*	*TAZ*
EMD	*LAMP2*	*TBX5*
FHL1	*PTPN11*	*TGFB3*
FKRP	*RAF1*	*TTR*

LIFESTYLE MODIFICATIONS[1,3]

DES	*JUP*	*PLN*
DSC2	*KCNH2*	*RBM20*
DSG2	*KCNQ1*	*RYR2*
DSP	*LMNA*	*SCN5A*
FLNC	*PKP2*	*TMEM43*

ENHANCED FAMILY SURVEILLANCE[1,2,3,4,6]

ACTC1	*DES*	*JUP*	*MYH7*	*RBM20*	*TNNT2*
CALM1	*DSC2*	*KCNH2*	*MYL2*	*RYR2*	*TRDN*
CALM2	*DSG2*	*KCNQ1*	*MYL3*	*SCN5A*	*TPM1*
CALM3	*DSP*	*LMNA*	*PLN*	*TMEM43*	*TTN*
CASQ2	*FLNC*	*MYBPC3*	*PKP2*	*TNNI3*	*TTR*

* With presence of additional risk factors

See reverse side for CardioNext gene list and associated risks

REFERENCES

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