

**ACMG SF v2.0 (September, 2016) (Kalia et al., 2016):**

Gene	Disease	Mode of Inheritance	MIM-Gene
ACTA2	Marfan Syndrome; Loeys-Dietz Syndromes; TAAD	Autosomal Dominant	102620
ACTC1	Hypertrophic cardiomyopathy; Dilated cardiomyopathy	Autosomal Dominant	102540
APC	Familial adenomatous polyposis	Autosomal Dominant	611731
APOB	Familial hypercholesterolemia	Autosomal Dominant	107730
ATP7B	Wilson disease	Autosomal Recessive	606882
BMPR1A	Juvenile polyposis	Autosomal Dominant	601299
BRCA1	Hereditary Breast and Ovarian Cancer	Autosomal Dominant	113705
BRCA2	Hereditary Breast and Ovarian Cancer	Autosomal Dominant	600185
CACNA1S	Malignant hyperthermia susceptibility	Autosomal Dominant	114208
COL3A1	Ehlers-Danlos syndrome – vascular type	Autosomal Dominant	120180
DSC2	Arrhythmogenic right ventricular cardiomyopathy	Autosomal Dominant	125645
DSG2	Arrhythmogenic right ventricular cardiomyopathy	Autosomal Dominant	125671
DSP	Arrhythmogenic right ventricular cardiomyopathy	Autosomal Dominant	125647
FBN1	Marfan Syndrome; Loeys-Dietz Syndromes; TAAD	Autosomal Dominant	134797
GLA	Hypertrophic cardiomyopathy	X-linked	300644
KCNH2	Long QT syndrome	Autosomal Dominant	152427
KCNQ1	Long QT syndrome	Autosomal Dominant	607542
LDLR	Familial hypercholesterolemia	Autosomal Dominant	606945
LMNA	Dilated cardiomyopathy; Arrhythmogenic right ventricular cardiomyopathy	Autosomal Dominant	150330
MEN1	Multiple Endocrine Neoplasia Type 1	Autosomal Dominant	613733
MLH1	Lynch Syndrome	Autosomal Dominant	120436
MSH2	Lynch Syndrome	Autosomal Dominant	609309
MSH6	Lynch Syndrome	Autosomal Dominant	600678
MUTYH	MYH-Associated Polyposis	Autosomal Recessive	604933
MYBPC3	Hypertrophic cardiomyopathy; Dilated cardiomyopathy	Autosomal Dominant	600958
MYL2	Hypertrophic cardiomyopathy	Autosomal Dominant	160781
MYL3	Hypertrophic cardiomyopathy	Autosomal Dominant	160790
MYH7	Hypertrophic cardiomyopathy; Dilated cardiomyopathy	Autosomal Dominant	160760
MYH11	Marfan Syndrome; Loeys-Dietz Syndromes; TAAD	Autosomal Dominant	160745
NF2	Neurofibromatosis type 2	Autosomal Dominant	607379
OTC	Ornithine transcarbamylase deficiency	X-Linked	300461
PCSK9	Familial hypercholesterolemia	Autosomal Dominant	607786
PKP2	Arrhythmogenic right ventricular cardiomyopathy	Autosomal Dominant	602861
PMS2	Lynch Syndrome	Autosomal Dominant	600259
PRKAG2	Hypertrophic cardiomyopathy; Dilated cardiomyopathy	Autosomal Dominant	602743
PTEN	PTEN Hamartoma Tumor Syndrome	Autosomal Dominant	601728
RB1	Retinoblastoma	Autosomal Dominant	614041
RET	Multiple Endocrine Neoplasia type 2; Familial Medullary Thyroid Cancer	Autosomal Dominant	164761
RYR1	Malignant hyperthermia susceptibility	Autosomal Dominant	180901
RYR2	Catecholaminergic polymorphic ventricular tachycardia	Autosomal Dominant	180902
SCN5A	Long QT syndrome; Brugada syndrome; Dilated Cardiomyopathy	Autosomal Dominant	600163
SDHAF2	Hereditary Paraganglioma-Pheochromocytoma Syndrome	Autosomal Dominant	613019
SDHB	Hereditary Paraganglioma-Pheochromocytoma Syndrome	Autosomal Dominant	185470
SDHC	Hereditary Paraganglioma-Pheochromocytoma Syndrome	Autosomal Dominant	602413
SDHD	Hereditary Paraganglioma-Pheochromocytoma Syndrome	Autosomal Dominant	602690
SMAD3	Marfan Syndrome; Loeys-Dietz Syndromes; TAAD	Autosomal Dominant	603109
SMAD4	Juvenile polyposis	Autosomal Dominant	600993
STK11	Peutz-Jeghers syndrome	Autosomal Dominant	602216
TGFBR1	Marfan Syndrome; Loeys-Dietz Syndromes; TAAD	Autosomal Dominant	190181
TGFBR2	Marfan Syndrome; Loeys-Dietz Syndromes; TAAD	Autosomal Dominant	190182
TMEM43	Arrhythmogenic right ventricular cardiomyopathy	Autosomal Dominant	612048
TNNI3	Hypertrophic cardiomyopathy; Dilated cardiomyopathy	Autosomal Dominant	191044
TNNT2	Hypertrophic cardiomyopathy; Dilated cardiomyopathy	Autosomal Dominant	191045
TP53	Li-Fraumeni Syndrome	Autosomal Dominant	191170
TPM1	Hypertrophic cardiomyopathy; Dilated cardiomyopathy	Autosomal Dominant	191010
TSC1	Tuberous Sclerosis Complex	Autosomal Dominant	605284
TSC2	Tuberous Sclerosis Complex	Autosomal Dominant	191092
VHL	Von Hippel Lindau syndrome	Autosomal Dominant	608537
WT1	WT1-related Wilms tumor	Autosomal Dominant	607102