https://www.ncbi.nlm.nih.gov/clinvar/docs/acmg/

ACMG Recommendations for Reporting of Secondary Findings in Clinical Exome and Genome Sequencing

The American College of Medical Genetics and Genomics has published recommendations for reporting secondary findings in clinical exome and genome sequencing.

The most recent recommendation is ACMG SF v3.2. Compared to the previous version, three genes were added - CALM1, CALM2, and CALM3. No genes were removed between the v3.2 and v3.1 lists.

The original published recommendation (PubMed 23788249) and the original PDF file are available, as well as clarifications and updates. Please note that NTRK1 was removed in an update to the original list.

NCBI adapted Table 1 of the original recommendation to facilitate access to information about the genes and diseases the table cites and to provide links to variation asserted to be pathogenic or likely pathogenic by at least one submitter to ClinVar. The content was generated from the MIM numbers reported in the ACMG recommendations, but disease names may be updated to correspond to what is used in MedGen for that MIM number. The link to ClinVar is provided only to support access; the results should not be interpreted as a statement that these alleles are universally accepted to be pathogenic or likely pathogenic.

Disease name and MIM number	MedGen	Gene via GTR	Variations that may be pathogenic
Adenomatous polyposis coli (MIM 175100)	MedGen	APC (MIM 611731)	ClinVar
Aortic aneurysm, familial thoracic 4 (MIM 132900)	MedGen	MYH11 (MIM 160745)	ClinVar
Aortic aneurysm, familial thoracic 6 (MIM 611788)	MedGen	ACTA2 (MIM 102620)	ClinVar
Arrhythmogenic right ventricular cardiomyopathy, type 5 (MIM 604400)	MedGen	TMEM43 (MIM 612048)	ClinVar

Arrhythmogenic right ventricular cardiomyopathy, type 8 (MIM 607450)	MedGen	DSP (MIM 125647)	ClinVar
Arrhythmogenic right ventricular cardiomyopathy, type 9 (MIM 609040)	MedGen	PKP2 (MIM 602861)	ClinVar
Arrhythmogenic right ventricular cardiomyopathy, type 10 (MIM 610193)	MedGen	DSG2 (MIM 125671)	ClinVar
Arrhythmogenic right ventricular cardiomyopathy, type 11 (MIM 610476)	MedGen	DSC2 (MIM 125645)	ClinVar
Biotinidase deficiency (MIM 253260)	MedGen	BTD (MIM 609019)	ClinVar
Breast-ovarian cancer, familial 1 (MIM 604370)	MedGen	BRCA1 (MIM 113705)	ClinVar
Breast-ovarian cancer, familial 2 (MIM 612555)	MedGen	BRCA2 (MIM 600185)	ClinVar
Brugada syndrome 1 (MIM 601144)	MedGen	SCN5A (MIM 600163)	ClinVar
Catecholaminergic polymorphic ventricular tachycardia 1 (MIM 604772)	MedGen	RYR2 (MIM 180902)	ClinVar
Catecholaminergic polymorphic ventricular tachycardia 2 (MIM 611938)	MedGen	CASQ2 (MIM 114251)	ClinVar
Catecholaminergic polymorphic ventricular tachycardia 4 (MIM 614916)	MedGen	CALM1 (MIM 614916)	ClinVar

Catecholaminergic polymorphic ventricular tachycardia 5 (MIM 615441)	MedGen	TRDN (MIM 603283)	ClinVar
Dilated cardiomyopathy (MIM 617047)	MedGen	FLNC (MIM 102565)	ClinVar
Dilated cardiomyopathy 1A (MIM 115200)	MedGen	LMNA (MIM 150330)	ClinVar
Dilated cardiomyopathy 1D (MIM 601494)	MedGen	TNNT2 (MIM 191045)	ClinVar
Dilated cardiomyopathy 1E (MIM 601154)	MedGen	SCN5A (MIM 600163)	ClinVar
Dilated cardiomyopathy 1I (MIM 604765)	MedGen	DES (MIM 125660)	ClinVar
Dilated cardiomyopathy 1S (MIM 613426)	MedGen	MYH7 (MIM 160760)	ClinVar
Dilated cardiomyopathy 1Z (MIM 611879)	MedGen	TNNC1 (MIM 191040)	ClinVar
Dilated cardiomyopathy 1DD (MIM 613172)	MedGen	RBM20 (MIM 613171)	ClinVar
Dilated cardiomyopathy 1HH (MIM 613881)	MedGen	BAG3 (MIM 603883)	ClinVar
Dilated cardiomyopathy (truncating variants only) (MIM 604145)	MedGen	TTN (MIM 188840)	ClinVar
Dilated cardiomyopathy with woolly hair, palmoplantar keratoderma, and tooth agenesis (MIM 615821)	MedGen	DSP (MIM 125647)	ClinVar

Ehlers-Danlos syndrome, type 4 (MIM 130050)	MedGen	COL3A1 (MIM 120180)	ClinVar
Fabry's disease (MIM 301500)	MedGen	GLA (MIM 300644)	ClinVar
Familial hypercholesterolemia 1 (MIM 143890)	MedGen	LDLR (MIM 606945)	ClinVar
Familial hypercholesterolemia 2 (MIM 144010)	MedGen	APOB (MIM 107730)	ClinVar
Familial hypertrophic cardiomyopathy 1 (MIM 192600)	MedGen	MYH7 (MIM 160760)	ClinVar
Familial hypertrophic cardiomyopathy 2 (MIM 115195)	MedGen	TNNT2 (MIM 191045)	ClinVar
Familial hypertrophic cardiomyopathy 3 (MIM 115196)	MedGen	TPM1 (MIM 191010)	ClinVar
Familial hypertrophic cardiomyopathy 4 (MIM 115197)	MedGen	MYBPC3 (MIM 600958)	ClinVar
Familial hypertrophic cardiomyopathy 6 (MIM 600858)	MedGen	PRKAG2 (MIM 602743)	ClinVar
Familial hypertrophic cardiomyopathy 7 (MIM 613690)	MedGen	TNNI3 (MIM 191044)	ClinVar
Familial hypertrophic cardiomyopathy 8 (MIM 608751)	MedGen	MYL3 (MIM 160790)	ClinVar
Familial hypertrophic cardiomyopathy 10 (MIM 608758)	MedGen	MYL2 (MIM 160781)	ClinVar

Familial hypertrophic cardiomyopathy 11 (MIM 612098)	MedGen	ACTC1 (MIM 102540)	ClinVar
Familial medullary thyroid carcinoma (MIM 155240)	MedGen	RET (MIM 164761)	ClinVar
Hereditary breast cancer (MIM 114480)	MedGen	PALB2 (MIM 610355)	ClinVar
Hereditary hemochromatosis (c.845G>A; p.C282Y homozygotes only) (MIM 235200)	MedGen	HFE (MIM 613609)	ClinVar
Hereditary hemorrhagic telangiectasia type 1 (MIM 187300)	MedGen	ENG (MIM 131195)	ClinVar
Hereditary hemorrhagic telangiectasia type 2 (MIM 600376)	MedGen	ACVRL1 (MIM 601284)	ClinVar
Hereditary paraganglioma-pheochromocytoma syndrome (MIM 168000, MIM 171300)	MedGen	SDHD (MIM 602690)	ClinVar
Hereditary paraganglioma-pheochromocytoma syndrome (MIM 115310, MIM 171300)	MedGen	SDHB (MIM 185470)	ClinVar
Hereditary transthyretin-related amyloidosis (MIM 105210)	MedGen	TTR (MIM 176300)	ClinVar
Hypercholesterolemia, autosomal dominant, 3 (MIM 603776)	MedGen	PCSK9 (MIM 607786)	ClinVar
Juvenile polyposis syndrome (MIM 174900)	MedGen	BMPR1A (MIM 601299)	ClinVar
Juvenile polyposis syndrome (MIM 174900)	MedGen	SMAD4 (MIM 600993)	ClinVar

Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome (MIM 175050)	MedGen	SMAD4 (MIM 600993)	ClinVar
Li-Fraumeni syndrome 1 (MIM 151623)	MedGen	TP53 (MIM 191170)	ClinVar
Loeys-Dietz syndrome type 1A (MIM 609192)	MedGen	TGFBR1 (MIM 190181)	ClinVar
Loeys-Dietz syndrome type 1B (MIM 610168)	MedGen	TGFBR2 (MIM 190182)	ClinVar
Loeys-Dietz syndrome type 3 (MIM 613795)	MedGen	SMAD3 (MIM 603109)	ClinVar
Long QT syndrome	MedGen	TRDN (MIM 603283)	ClinVar
Long QT syndrome 1 (MIM 192500)	MedGen	KCNQ1 (MIM 607542)	ClinVar
Long QT syndrome 2 (MIM 613688)	MedGen	KCNH2 (MIM 152427)	ClinVar
Long QT syndrome 3 (MIM 603830)	MedGen	SCN5A (MIM 600163)	ClinVar
Long QT syndrome 14 (MIM 616247)	MedGen	CALM1 (MIM 114180)	ClinVar
Long QT syndrome 15 (MIM 616249); also associated with catecholaminergic polymorphic ventricular tachycardia	MedGen	CALM2 (MIM 114182)	ClinVar
Long QT syndrome 16 (MIM 618782); also associated with catecholaminergic polymorphic ventricular tachycardia	MedGen	CALM3 (MIM 114183)	ClinVar

Lynch syndrome 1 (MIM 120435)	MedGen	MSH2 (MIM 609309)	ClinVar
Lynch syndrome 2 (MIM 609310)	MedGen	MLH1 (MIM 120436)	ClinVar
Lynch syndrome 4 (MIM 614337)	MedGen	PMS2 (MIM 600259)	ClinVar
Lynch syndrome 5 (MIM 614350)	MedGen	MSH6 (MIM 600678)	ClinVar
Malignant hyperthermia (MIM 145600)	MedGen	RYR1 (MIM 180901)	ClinVar
Malignant hyperthermia (MIM 601887)	MedGen	CACNA1S (MIM 114208)	ClinVar
Marfan's syndrome (MIM 154700)	MedGen	FBN1 (MIM 134797)	ClinVar
Maturity-Onset of Diabetes of the Young (MIM 600496)	MedGen	HNF1A (MIM 142410)	ClinVar
Multiple endocrine neoplasia, type 1 (MIM 131100)	MedGen	MEN1 (MIM 613733)	ClinVar
Multiple endocrine neoplasia, type 2a (MIM 171400)	MedGen	RET (MIM 164761)	ClinVar
Multiple endocrine neoplasia, type 2b (MIM 162300)	MedGen	RET (MIM 164761)	ClinVar
MYH-associated polyposis (MIM 608456)	MedGen	MUTYH (MIM 604933)	ClinVar

Myofibrillar myopathy 1 (MIM 601419)	MedGen	DES (MIM 125660)	ClinVar
Myofibrillar myopathy 5 (MIM 609524)	MedGen	FLNC (MIM 102565)	ClinVar
Myofibrillar myopathy 6 (MIM 612954)	MedGen	BAG3 (MIM 603883)	ClinVar
Neurofibromatosis, type 2 (MIM 101000)	MedGen	NF2 (MIM 607379)	ClinVar
Ornithine carbamoyltransferase deficiency (MIM 311250)	MedGen	OTC (MIM 300461)	ClinVar
Paragangliomas 2 (MIM 601650)	MedGen	SDHAF2 (MIM 613019)	ClinVar
Paragangliomas 3 (MIM 605373)	MedGen	SDHC (MIM 602413)	ClinVar
Peutz-Jeghers syndrome (MIM 175200)	MedGen	STK11 (MIM 602216)	ClinVar
Pheochromocytoma (MIM 171300)	MedGen	MAX (MIM 154950)	ClinVar
Pheochromocytoma (MIM 171300)	MedGen	TMEM127 (MIM 613403)	ClinVar
Pompe disease (MIM 232300)	MedGen	GAA (MIM 606800)	ClinVar
PTEN hamartoma tumor syndrome (MIM 158350)	MedGen	PTEN (MIM 601728)	ClinVar

Retinoblastoma (MIM 180200)	MedGen	RB1 (MIM 614041)	ClinVar
RPE65-related retinopathy (MIM 204100, MIM 613794)	MedGen	RPE65 (MIM 180069)	ClinVar
Tuberous sclerosis 1 (MIM 191100)	MedGen	TSC1 (MIM 605284)	ClinVar
Tuberous sclerosis 2 (MIM 613254)	MedGen	TSC2 (MIM 191092)	ClinVar
Von Hippel-Lindau syndrome (MIM 193300)	MedGen	VHL (MIM 608537)	ClinVar
Wilms' tumor (MIM 194070)	MedGen	WT1 (MIM 607102)	ClinVar
Wilson disease (MIM 277900)	MedGen	ATP7B (MIM 606882)	ClinVar