

Current List of genes for reporting of Secondary Variants (v 3.2, Miller et al., 2023 <https://doi.org/10.1016/j.gim.2023.100866>)

Gene	Gene MIM	Disease/Phenotype	Disorder MIM	Phenotype Category	Inheritance	SF List Version	Variants to report
ACTA2	102620	Familial thoracic aortic aneurysm	611788	Cardiovascular	AD	1.0	All P and LP
ACTC1	102540	Hypertrophic cardiomyopathy	612098	Cardiovascular	AD	1.0	All P and LP
ACVRL1	601284	Hereditary hemorrhagic telangiectasia	600376	Miscellaneous	AD	3.0	All P and LP
APC	611731	Familial adenomatous polyposis	175100	Cancer	AD	1.0	All P and LP
APOB	107730	Familial hypercholesterolemia	144010	Cardiovascular	AD	1.0	All P and LP
ATP7B	606882	Wilson disease	277900	Miscellaneous	AR	2.0	P and LP (2 variants)
BAG3	603883	Dilated cardiomyopathy	613881	Cardiovascular	AD	3.1	All P and LP
		Myofibrillar myopathy	612954	Cardiovascular	AD	3.1	All P and LP
BMPRIA	601299	Juvenile polyposis syndrome	174900	Cancer	AD	1.0	All P and LP
BRCA1	113705	Hereditary breast and ovarian cancer	604370	Cancer	AD	1.0	All P and LP
BRCA2	600185	Hereditary breast and ovarian cancer	612555	Cancer	AD	1.0	All P and LP
BTD	609019	Biotinidase deficiency	253260	Metabolic	AR	3.0	P and LP (2 variants)
CACNA1S	114208	Malignant hyperthermia	601887	Miscellaneous	AD	1.0	All P and LP
		Long-QT syndrome type 14	616247	Cardiovascular	AD	3.2	All P and LP
CALM1	114180	Catecholaminergic polymorphic ventricular tachycardia	614916	Cardiovascular	AD	3.2	All P and LP
		Long-QT syndrome type 15	616249	Cardiovascular	AD	3.2	All P and LP
CALM2	114182	Catecholaminergic polymorphic ventricular tachycardia	616249	Cardiovascular	AD	3.2	All P and LP
		Long-QT syndrome type 16	618782	Cardiovascular	AD	3.2	All P and LP
CALM3	114183	Catecholaminergic polymorphic ventricular tachycardia	618782	Cardiovascular	AD	3.2	All P and LP
CASQ2	114251	Catecholaminergic polymorphic ventricular tachycardia	611938	Cardiovascular	AR	3.0	P and LP (2 variants)
COL3A1	120180	Ehlers-Danlos syndrome, vascular type	130050	Cardiovascular	AD	1.0	All P and LP
DES	125660	Dilated cardiomyopathy	604765	Cardiovascular	AD	3.1	All P and LP
		Myofibrillar myopathy	601419	Cardiovascular	AD	3.1	All P and LP
DSC2	125645	Arrhythmogenic right ventricular cardiomyopathy	610476	Cardiovascular	AD	1.0	All P and LP
DSG2	125671	Arrhythmogenic right ventricular cardiomyopathy	610193	Cardiovascular	AD	1.0	All P and LP
DSP	125647	Arrhythmogenic right ventricular cardiomyopathy	607450	Cardiovascular	AD	1.0	All P and LP
		Dilated cardiomyopathy	615821	Cardiovascular	AD	1.0	All P and LP
ENG	131195	Hereditary hemorrhagic telangiectasia	187300	Miscellaneous	AD	3.0	All P and LP
FBN1	134797	Marfan syndrome	154700	Cardiovascular	AD	1.0	All P and LP
		Dilated cardiomyopathy	n/a	Cardiovascular	AD	3.0	All P and LP
FLNC	102565	Hypertrophic cardiomyopathy	617047	Cardiovascular	AD	3.0	All P and LP
		Myofibrillar myopathy	609524	Cardiovascular	AD	3.0	All P and LP
GAA	606800	Pompe disease	232300	Metabolic	AR	3.0	P and LP (2 variants)
GLA	300644	Fabry disease	301500	Metabolic	XL	1.0	All hemi, het, homozygous P and LP
HFE	613609	Hereditary hemochromatosis (c.845G>A; p.C282Y homozygotes only)	235200	Miscellaneous	AR	3.0	p.C282Y homozygotes only
HNF1A	142410	Maturity-Onset of Diabetes of the Young	600496	Miscellaneous	AD	3.0	All P and LP
KCNH2	152427	Long-QT syndrome type 2	613688	Cardiovascular	AD	1.0	All P and LP
KCNQ1	607542	Long-QT syndrome type 1	192500	Cardiovascular	AD	1.0	All P and LP
LDLR	606945	Familial hypercholesterolemia	143890	Cardiovascular	AD	1.0	All P and LP
LMNA	150330	Dilated cardiomyopathy	115200	Cardiovascular	AD	1.0	All P and LP
MAX	154950	Hereditary paraganglioma-pheochromocytoma syndrome	171300	Cancer	AD	3.0	All P and LP
MEN1	613733	Multiple endocrine neoplasia type 1	131100	Cancer	AD	1.0	All P and LP
MLH1	120436	Lynch syndrome	609310	Cancer	AD	1.0	All P and LP
MSH2	609309	Lynch syndrome	120435	Cancer	AD	1.0	All P and LP
MSH6	600678	Lynch syndrome	614350	Cancer	AD	1.0	All P and LP
MUTYH	604933	MUTYH-associated polyposis	608456	Cancer	AR	1.0	P and LP (2 variants)
MYBPC3	600958	Hypertrophic cardiomyopathy	115197	Cardiovascular	AD	1.0	All P and LP
MYH11	160745	Familial thoracic aortic aneurysm	132900	Cardiovascular	AD	1.0	All P and LP
MYH7	160760	Hypertrophic cardiomyopathy	192600	Cardiovascular	AD	1.0	All P and LP
		Dilated cardiomyopathy	613426	Cardiovascular	AD	1.0	All P and LP
MYL2	160781	Hypertrophic cardiomyopathy	608758	Cardiovascular	AD	1.0	All P and LP
MYL3	160790	Hypertrophic cardiomyopathy	608751	Cardiovascular	AD	1.0	All P and LP
NF2	607379	NF2-related schwannomatosis	101000	Cancer	AD	1.0	All P and LP
OTC	300461	Ornithine transcarbamylase deficiency	311250	Metabolic	XL	2.0	All hemi, het, homozygous P and LP
PALB2	610355	Hereditary breast cancer	114480	Cancer	AD	3.0	All P and LP
PCSK9	607786	Familial hypercholesterolemia	603776	Cardiovascular	AD	1.0	All P and LP
PKP2	602861	Arrhythmogenic right ventricular cardiomyopathy	609040	Cardiovascular	AD	1.0	All P and LP
PMS2	600259	Lynch syndrome	614337	Cancer	AD	1.0	All P and LP
PRKAG2	602743	Hypertrophic cardiomyopathy	600858	Cardiovascular Metabolic	AD	1.0	All P and LP
PTEN	601728	PTEN hamartoma tumor syndrome	158350	Cancer	AD	1.0	All P and LP
RB1	614041	Retinoblastoma	180200	Cancer	AD	1.0	All P and LP
RBM20	613171	Dilated cardiomyopathy	613172	Cardiovascular	AD	3.1	All P and LP
		Familial medullary thyroid cancer	155240	Cancer	AD	1.0	All P and LP
RET	164761	Multiple endocrine neoplasia type 2A	171400	Cancer	AD	1.0	All P and LP
		Multiple endocrine neoplasia type 2B	162300	Cancer	AD	1.0	All P and LP
		RPE65-related retinopathy	204100, 613794	Miscellaneous	AR	3.0	P and LP (2 variants)
RPE65	180069						
RYR1	180901	Malignant hyperthermia	145600	Miscellaneous	AD	1.0	All P and LP
RYR2	180902	Catecholaminergic polymorphic ventricular tachycardia	604772	Cardiovascular	AD	1.0	All P and LP
		Long QT syndrome type 3	603830	Cardiovascular	AD	1.0	All P and LP
SCN5A	600163	Brugada syndrome	601144	Cardiovascular	AD	1.0	All P and LP
		Dilated cardiomyopathy	601154	Cardiovascular	AD	1.0	All P and LP
SDHAF2	613019	Hereditary paraganglioma-pheochromocytoma syndrome	601650	Cancer	AD	1.0	All P and LP
			115310,				
SDHB	185470	Hereditary paraganglioma-pheochromocytoma syndrome	171300	Cancer	AD	1.0	All P and LP
SDHC	602413	Hereditary paraganglioma-pheochromocytoma syndrome	605373	Cancer	AD	1.0	All P and LP
			168000,				
SDHD	602690	Hereditary paraganglioma-pheochromocytoma syndrome	171300	Cancer	AD	1.0	All P and LP
SMAD3	603109	Loeys-Dietz syndrome	613795	Cardiovascular	AD	1.0	All P and LP
SMAD4	600993	Juvenile polyposis syndrome	174900	Cancer	AD	1.0	All P and LP
		Hereditary hemorrhagic telangiectasia	175050	Miscellaneous	AD	1.0	All P and LP
STK11	602216	Peutz-Jeghers syndrome	175200	Cancer	AD	1.0	All P and LP
TGFB1	190181	Loeys-Dietz syndrome	609192	Cardiovascular	AD	1.0	All P and LP
TGFB2	190182	Loeys-Dietz syndrome	610168	Cardiovascular	AD	1.0	All P and LP
TMEM127	613403	Hereditary paraganglioma-pheochromocytoma syndrome	171300	Cancer	AD	3.0	All P and LP
TMEM43	612048	Arrhythmogenic right ventricular cardiomyopathy	604400	Cardiovascular	AD	1.0	All P and LP
TNNC1	191040	Dilated cardiomyopathy	611879	Cardiovascular	AD	3.1	All P and LP
TNNI3	191044	Hypertrophic cardiomyopathy	613690	Cardiovascular	AD	1.0	All P and LP
		Dilated cardiomyopathy	601494	Cardiovascular	AD	1.0	All P and LP
TNN2	191045	Hypertrophic cardiomyopathy	115195	Cardiovascular	AD	1.0	All P and LP

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<i>TP53</i>	191170	Li-Fraumeni syndrome	151623	Cancer	AD	1.0	All P and LP
<i>TPM1</i>	191010	Hypertrophic cardiomyopathy	115196	Cardiovascular	AD	1.0	All P and LP
<i>TRDN</i>	603283	Catecholaminergic polymorphic ventricular tachycardia	615441	Cardiovascular	AR	3.0	All P and LP
		Long QT syndrome	n/a	Cardiovascular	AR	3.0	All P and LP
<i>TSC1</i>	605284	Tuberous sclerosis complex	191100	Cancer	AD	1.0	All P and LP
<i>TSC2</i>	191092	Tuberous sclerosis complex	613254	Cancer	AD	1.0	All P and LP
<i>TTN</i>	188840	Dilated cardiomyopathy (truncating variants only)	604145	Cardiovascular	AD	3.0	P and LP (truncating variants only)
<i>TTR</i>	176300	Hereditary transthyretin-related amyloidosis	105210	Miscellaneous	AD	3.1	All P and LP
<i>VHL</i>	608537	Von Hippel-Lindau syndrome	193300	Cancer	AD	1.0	All P and LP
<i>WT1</i>	607102	<i>WT1</i> -related Wilms tumor	194070	Cancer	AD	1.0	All P and LP