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ACMG SECONDARY FINDINGS GENE LIST

The American College of Medical Genetics and Genomics (ACMG)[1] recommends analysis of certain genes for pathogenic variants in individuals undergoing genome and/or exome sequencing. If there is clinical suspicion or family history of a genetic condition associated with one of the ACMG-recommended genes, additional targeted testing should be considered, as standard genome or exome sequencing will not identify all pathogenic variants in these genes. Note that single pathogenic variants in autosomal recessive ACMG-recommended genes are not reported.

Last updated: September 2025

ACMG Recommends Reporting Secondary Findings for These Genes		
Conditions		**Associated Genes**
Tumors/cancer syndromes	Familial adenomatous polyposis	*APC*
	Familial medullary thyroid cancer Multiple endocrine neoplasia type 2	*RET*
	Hereditary breast and ovarian cancer	*BRCA1*, *BRCA2*, *PALB2*
	Hereditary paraganglioma/ pheochromocytoma	*MAX*, *SDHAF2*, *SDHB*, *SDHC*, *SDHD*, *TMEM127*
	Juvenile polyposis	*BMPR1A*, *SMAD4*
	Li-Fraumeni syndrome	*TP53*
	Lynch syndrome	*MLH1*, *MSH2*, *MSH6*, *PMS2*
	Multiple endocrine neoplasia type 1	*MEN1*
	MUTYH-associated polyposis	*MUTYH*
	Neurofibromatosis type 2	*NF2*
	Peutz-Jeghers syndrome	*STK11*
	PTEN hamartoma tumor syndrome	*PTEN*
	Retinoblastoma	*RB1*
	Tuberous sclerosis complex	*TSC1*, *TSC2*
	Von Hippel-Lindau syndrome	*VHL*
	WT1-related Wilms tumor	*WT1*

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ACMG Recommends Reporting Secondary Findings for These Genes		
Conditions		**Associated Genes**
Cardiovascular conditions/ syndromes	Arrhythmogenic right-ventricular cardiomyopathy	*DSC2*, *DSG2*, *DSP*, *PKP2*, *TMEM43*
	Brugada syndrome Romano-Ward long QT syndrome, types 1–3 and 14–16	*CALM1*, *CALM2*, *CALM3*, *KCNH2*, *KCNQ1*, *SCN5A*
	Catecholaminergic polymorphic ventricular tachycardia	*CASQ2*, *RYR2*, *TRDN*
	Ehlers-Danlos syndrome, vascular type	*COL3A1*
	Familial hypercholesterolemia	*APOB*, *LDLR*, *PCSK9*
	Familial thoracic aortic aneurysms and dissections	*ACTA2*, *MYH11*, *SMAD3*
	Hypertrophic cardiomyopathy, dilated cardiomyopathy	*ACTC1*, *BAG3*, *DES*, *FLNC*, *GLA*, *LMNA*, *MYBPC3*, *MYH7*, *MYL2*, *MYL3*, *PLN*, *PRKAG2*, *RBM20*, *TNNC1*, *TNNI3*, *TNNT2*, *TPM1*, *TTN*
	Loeys-Dietz syndrome	*TGFBR1*, *TGFBR2*
	Marfan syndrome	*FBN1*
Metabolic conditions	Biotinidase deficiency	*BTD*
	Cerebrotendinous xanthomatosis	*CYP27A1*
	Fabry disease	*GLA*
	Maturity-onset diabetes of the young	*HNF1A*
	Ornithine transcarbamylase deficiency	*OTC*
	Pompe disease	*GAA*
	Wilson disease	*ATP7B*
	X-linked adrenoleukodystrophy	*ABCD1*
Other conditions	Hereditary hemochromatosis	*HFE*
	Hereditary hemorrhagic telangiectasia	*ACVRL1*, *ENG*
	Hereditary transthyretin amyloidosis	*TTR*
	Malignant hyperthermia susceptibility	*CACNA1S*, *RYR1*
	RPE65-related retinopathy	*RPE65*

REFERENCES

Lee K, Abul-Husn NS, Amendola LM, et al. ACMG SF v3.3 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of the American College of Medical Genetics and Genomics (ACMG). *Genet Med*. 2025;27:101454.