

ACMG Secondary Findings Gene List

Exome testing includes analysis for secondary findings in the 84 genes recommended by American College of Medical Genetics and Genomics (ACMG) guidelines. These genes are related to conditions for which medical management is available to alter the course of the disease.

Likely pathogenic and pathogenic variants in these genes will be reported unless an opt-out is selected with the order. Separate secondary finding reports are issued for each family member tested with Duo or Trio testing.

Reference: 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(8):101454. doi:10.1016/j.jim.2025.101454

Cancer Risk	
<i>APC</i>	Familial adenomatous polyposis
<i>BMPR1A</i>	Juvenile polyposis syndrome
<i>BRCA1</i>	Hereditary breast and ovarian cancer
<i>BRCA2</i>	Hereditary breast and ovarian cancer
<i>MAX</i>	Hereditary paraganglioma-pheochromocytoma syndrome
<i>MEN1</i>	Multiple endocrine neoplasia type 1
<i>MLH1</i>	Lynch syndrome
<i>MSH2</i>	Lynch syndrome
<i>MSH6</i>	Lynch syndrome
<i>MUTYH</i> [1]	MUTYH-associated polyposis
<i>NF2</i>	Neurofibromatosis type 2
<i>PALB2</i>	Hereditary breast and ovarian cancer
<i>PMS2</i>	Lynch syndrome
<i>PTEN</i>	PTEN hamartoma tumor syndrome
<i>RB1</i>	Retinoblastoma
<i>RET</i>	Multiple endocrine neoplasia type 2; Familial medullary thyroid cancer (FMTC)
<i>SDHAF2</i>	Hereditary paraganglioma-pheochromocytoma syndrome
<i>SDHB</i>	Hereditary paraganglioma-pheochromocytoma syndrome
<i>SDHC</i>	Hereditary paraganglioma-pheochromocytoma syndrome
<i>SDHD</i>	Hereditary paraganglioma-pheochromocytoma syndrome
<i>SMAD4</i>	Juvenile polyposis syndrome
<i>STK11</i>	Peutz-Jeghers syndrome
<i>TMEM127</i>	Hereditary paraganglioma-pheochromocytoma syndrome
<i>TP53</i>	Li-Fraumeni syndrome
<i>TSC1</i>	Tuberous sclerosis complex
<i>TSC2</i>	Tuberous sclerosis complex
<i>VHL</i>	von Hippel Lindau syndrome
<i>WT1</i>	WT1-related Wilms tumor
Cardiovascular Disease	
<i>ACTA2</i>	Thoracic aortic aneurysms and dissections
<i>ACTC1</i>	Cardiomyopathy
<i>APOB</i>	Familial hypercholesterolemia
<i>BAG3</i>	Cardiomyopathy
<i>CALM1</i>	Long QT syndrome
<i>CALM2</i>	Long QT syndrome, Catecholaminergic polymorphic ventricular tachycardia
<i>CALM3</i>	Long QT syndrome, Catecholaminergic polymorphic ventricular tachycardia
<i>CASQ2</i> [1]	Catecholaminergic polymorphic ventricular cardiomyopathy
<i>COL3A1</i>	Ehlers-Danlos syndrome, vascular type

<i>DES</i>	Cardiomyopathy
<i>DSC2</i>	Arrhythmogenic right ventricular cardiomyopathy
<i>DSG2</i>	Arrhythmogenic right ventricular cardiomyopathy
<i>DSP</i>	Arrhythmogenic right ventricular cardiomyopathy
<i>FBN1</i>	Marfan syndrome
<i>FLNC</i>	Cardiomyopathy
<i>KCNH2</i>	Long QT syndrome
<i>KCNQ1</i>	Long QT syndrome
<i>LDLR</i>	Familial hypercholesterolemia
<i>LMNA</i>	Cardiomyopathy
<i>MYBPC3</i>	Cardiomyopathy
<i>MYH11</i>	Thoracic aortic aneurysms and dissections
<i>MYH7</i>	Cardiomyopathy
<i>MYL2</i>	Cardiomyopathy
<i>MYL3</i>	Cardiomyopathy
<i>PCSK9</i>	Familial hypercholesterolemia
<i>PKP2</i>	Arrhythmogenic right ventricular cardiomyopathy
<i>PLN</i>	Arrhythmogenic right ventricular cardiomyopathy, dilated cardiomyopathy, and hypertrophic cardiomyopathy
<i>PRKAG2</i>	Cardiomyopathy
<i>RBM20</i>	Cardiomyopathy
<i>RYR2</i>	Catecholaminergic polymorphic ventricular tachycardia
<i>SCN5A</i>	Long QT syndrome, Brugada syndrome
<i>SMAD3</i>	Loeys-Dietz syndrome
<i>TGFBR1</i>	Loeys-Dietz syndrome
<i>TGFBR2</i>	Loeys-Dietz syndrome
<i>TMEM43</i>	Arrhythmogenic right ventricular cardiomyopathy
<i>TNNC1</i>	Cardiomyopathy
<i>TNNI3</i>	Cardiomyopathy
<i>TNNT2</i>	Cardiomyopathy
<i>TPM1</i>	Cardiomyopathy
<i>TRDN</i> [1]	Catecholaminergic polymorphic ventricular tachycardia
<i>TTN</i>	Cardiomyopathy

Inborn Errors of Metabolism

<i>ABCD1</i>	X-linked adrenoleukodystrophy
<i>BTD</i> [1]	Biotinidase deficiency
<i>CYP27A1</i> [1]	Cerebrotendinous xanthomatosis
<i>GAA</i> [1]	Pompe disease
<i>GLA</i>	Fabry disease
<i>OTC</i>	Ornithine transcarbamylase deficiency

Other Genetic Disease

<i>ACVRL1</i>	Hereditary hemorrhagic telangiectasia
<i>CACNA1S</i>	Malignant hyperthermia susceptibility
<i>ATP7B</i> [1]	Wilson disease
<i>ENG</i>	Hereditary hemorrhagic telangiectasia
<i>HFE</i> [1]	Hereditary hemochromatosis
<i>HNF1A</i> [1]	Maturity-onset diabetes of the young
<i>RPE65</i> [1]	RPE65-related retinopathy
<i>RYR1</i>	Malignant hyperthermia susceptibility
<i>TTR</i>	Hereditary transthyretin-related amyloidosis

[1] Reported only when 2 P/LP variants are identified. For *HFE* specifically, only p.C282Y homozygotes are reported.