



# ACMG Secondary Findings

## v3.1 Gene List

Gene(s)	Disorder(s)
<b>Cancer Phenotypes</b>	
<i>APC</i>	Familial adenomatous polyposis
<i>RET</i>	Familial medullary thyroid cancer
<i>BRCA1, BRCA2, PALB2</i>	Hereditary breast and/or ovarian cancer
<i>SDHD, SDHAF2, SDHC, SDHB, MAX, TMEM127</i>	Hereditary paraganglioma- pheochromocytoma syndrome
<i>BMPR1A, SMAD4</i>	Juvenile polyposis syndrome
<i>TP53</i>	Li-Fraumeni syndrome
<i>MLH1, MSH2, MSH6, PMS2</i>	Lynch syndrome
<i>MEN1</i>	Multiple endocrine neoplasia type 1
<i>MUTYH*</i>	MUTYH-associated polyposis
<i>NF2</i>	Neurofibromatosis type 2
<i>STK11</i>	Peutz-Jeghers syndrome
<i>PTEN</i>	PTEN hamartoma syndrome
<i>RB1</i>	Retinoblastoma
<i>TSC1, TSC2</i>	Tuberous sclerosis complex
<i>VHL</i>	von Hippel-Lindau syndrome
<i>WT1</i>	WT1-related Wilms tumor
<b>Cardiovascular Phenotypes</b>	
<i>FBN1, TGFB1, TGFB2, SMAD3, ACTA2, MYH11</i>	Aortopathies
<i>PKP2, DSP, DSC2, TMEM43, DSG2</i>	Arrhythmogenic right ventricular cardiomyopathy
<i>RYR2, CASQ2*, TRDN</i>	Catecholaminergic polymorphic ventricular tachycardia
<i>BAG3, DES, RBM20, TNNC1, TNNT2, LMNA, FLNC, TTN</i>	Dilated cardiomyopathy
<i>COL3A1</i>	Ehlers-Danlos syndrome, vascular type
<i>LDLR, APOB, PCSK9</i>	Familial hypercholesterolemia
<i>MYH7, MYBPC3, TNNT3, TPM1, MYL3, ACTC1, PRKAG2, MYL2</i>	Hypertrophic cardiomyopathy
<i>KCNQ1, KCNH2</i>	Long QT syndrome, types 1 and 2
<i>SCN5A</i>	Long QT syndrome, type 3; Brugada syndrome

Gene(s)	Disorder(s)
<b>Inborn Errors of Metabolism Phenotypes</b>	
<i>BTBD*</i>	Biotinidase deficiency
<i>GLA</i>	Fabry disease
<i>OTC</i>	Ornithine transcarbamylase deficiency
<i>GAA*</i>	Pompe

<b>Other Phenotypes</b>	
<i>HFE**</i>	Hereditary hemochromatosis
<i>ACVRL1, ENG</i>	Hereditary hemorrhagic telangiectasia
<i>RYR1, CACNA15</i>	Malignant hypothermia
<i>HNFB1A</i>	Maturity-onset diabetes of the young
<i>RPE65*</i>	RPE65-related retinopathy
<i>ATP7B*</i>	Wilson disease
<i>TTR</i>	Hereditary transthyretin amyloidosis

\*Will be reported only if two likely pathogenic and/or pathogenic variants are identified (homozygous or compound heterozygous state).

\*\*HFE p.Cys282Try homozygous only