Ambry Genetics®

ACMG Secondary Findings Gene List

Exome testing includes analysis for secondary findings in the 81 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: Miller DT, Lee K, Abul-Husn NS, et al. ACMG SF v3.2 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*. 2023;25(8):100866. doi:10.1016/j.gim.2023.100866

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

COL3A1	Ehlers-Danlos syndrome, vascular type
DES	Cardiomyopathy
DSC2	Arrhythmogenic right ventricular cardiomyopathy
DSG2	Arrhythmogenic right ventricular cardiomyopathy
DSP	Arrhythmogenic right ventricular cardiomyopathy
FBN1	Marfan syndrome
FLNC	Cardiomyopathy
KCNH2	Long QT syndrome
KCNQ1	Long QT syndrome
LDLR	Familial hypercholesterolemia
LMNA	Cardiomyopathy
MYBPC3	Cardiomyopathy
MYH11	Thoracic aortic aneurysms and dissections
MYH7	Cardiomyopathy
MYL2	Cardiomyopathy
MYL3	Cardiomyopathy
PCSK9	Familial hypercholesterolemia
PKP2	Arrhythmogenic right ventricular cardiomyopathy
PRKAG2	Cardiomyopathy
RBM20	Cardiomyopathy
RYR2	Catecholaminergic polymorphic ventricular tachycardia
SCN5A	Long QT syndrome, Brugada syndrome
SMAD3	Loeys-Dietz syndrome
TGFBR1	Loeys-Dietz syndrome
TGFBR2	Loeys-Dietz syndrome
TMEM43	Arrhythmogenic right ventricular cardiomyopathy
TNNC1	Cardiomyopathy
TNNI3	Cardiomyopathy
TNNT2	Cardiomyopathy
TPM1	Cardiomyopathy
TRDN[1]	Catecholaminergic polymorphic ventricular tachycardia
TTN	Cardiomyopathy
Inborn Errors of Metabolism	
BTD[1]	Biotinidase deficiency
GAA[1]	Pompe disease
GLA	Fabry disease
OTC	Ornithine transcarbamylase deficiency
Other Genetic Disease	
ACVRL1	Hereditary hemorrhagic telangiectasia
CACNA1S	Malignant hyperthermia susceptibility
ATP7B[1]	Wilson disease
ENG	Hereditary hemorrhagic telangiectasia
HFE[1]	Hereditary hemochromatosis
HNF1A[1]	Maturity-onset diabetes of the young
RPE65[1]	RPE65-related retinopathy
RYR1	Malignant hyperthermia susceptibility
TTR	Hereditary transthyretin-related amyloidosis

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

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MKT-SPEC-FLYR-50085-EN v1 07.29.24