ACMG Secondary Findings

v3.2 Gene LIst

Gene(s)	Disorder(s)
Cancer Phenotypes	
APC	Familial adenomatous polyposis
RET	Familial medullary thyroid cancer
BRCA1, BRCA2, PALB2	Hereditary breast and/or ovarian cancer
SDHD, SDHAF2, SDHC, SDHB, MAX, TMEM127	Hereditary paraganglioma- pheochromocytoma syndrome
BMPR1A, SMAD4	Juvenile polyposis syndrome
TP53	Li-Fraumeni syndrome
MLH1, MSH2, MSH6, PMS2	Lynch syndrome
MEN1	Multiple endocrine neoplasia type 1
*MUTYH**	MUTYH-associated polyposis
NF2	Neurofibromatosis type 2
STK11	Peutz-Jeghers syndrome
PTEN	PTEN hamartoma syndrome
RB1	Retinoblastoma
TSC1, TSC2	Tuberous sclerosis complex
VHL	von Hippel-Lindau syndrome
WT1	WT1-related Wilms tumor

Cardiovascular Phenotypes	
FBN1, TGFBR1, TGFBR2, SMAD3, ACTA2, MYH11	Aortopathies
PKP2, DSP, DSC2, TMEM43. DSG2	Arrhythmogenic right ventricular cardiomyopathy
RYR2, CASQ2, TRDN*	Catecholaminergic polymorphic ventricular tachycardia
BAG3, DES, RBM20, TNNC1, TNNT2, LMNA, FLNC, TTN	Dilated cardiomyopathy
CALM1, CALM2, CALM3	Long QT syndrome types 14 and 16
COL3A1	Ehlers-Danlos syndrome, vascular type
LDLR, APOB, PCSK9	Familial hypercholesterolemia
MYH7, MYBPC3, TNNI3, TPM1, MYL3, ACTC1, PRKAG2, MYL2	Hypertrophic cardiomyopathy
KCNQ1, KCNH2	Long QT syndrome, types 1 and 2
SCN5A	Long QT syndrome, type 3; Brugada syndrome

Gene(s)	Disorder(s)
Inborn Errors of Metabolism Phenotypes	
*BTD**	Biotinidase deficiency
GLA	Fabry disease
OTC	Ornithine transcarbamylase deficiency
*GAA**	Pompe

Other Phenotypes	
*HFE***	Hereditary hemochromatosis
ACVRL1, ENG	Hereditary hemorrhagic telangiectasia
RYR1, CACNA15	Malignant hypothermia
HNF1A	Maturity-onset diabetes of the young
*RPE65**	RPE65-related retinopathy
*ATP7B**	Wilson disease
TTR	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