Genes annotated by NHGRI SGFS for reportable secondary variants

Adapted from Kalia et al., 2017

Gene (s)	Associated Disorder(s)
BRCA1 *BRCA2*	Hereditary breast and ovarian cancer
TP53	Li-Fraumeni syndrome
STK11	Peutz-Jeghers syndrome
APC	Familial adenomatous polyposis
MUTYH	MYH-associated polyposis, FAP type 2, colorectal adenomatous polyposis, recessive, with pilomatricomas
BMPR1A *SMAD4*	Juvenile Polyposis
VHL	Von Hippel-Lindau syndrome
MEN1	Multiple endocrine neoplasia type 1
MEN2	Multiple endocrine neoplasia type 1
RET	Familial medullary thyroid cancer
PTEN	PTEN hamartoma tumor syndrome
RB1	Retinoblastoma
SDHD *SDHAF2*	Hereditary paraganglioma-pheochromocytoma syndrome
TSC1 *TSC2*	Tuberous sclerosis complex
WT1	WT1-related Wilms tumor
NF2	Neurofibromatosis type 2
COL3A1	Ehlers-Danlos syndrome, vascular type
FBN1 *TGFBR1* *TGFBR2* *SMAD3* *ACTA2* *MYH11*	Marfan syndrome, Loeys-Dietz syndromes, and familial thoracic aortic aneurysms
MYBPC3 *MYH7* *TNNT2* *TINNI3* *TPM1* *MYL3* *ACTC1* *PRKAG2* *GLA* *MYL2* *LMNA*	Hypertrophic cardiomyopathy, dilated cardiomyopathy
RYR2	Catecholaminergic polymorphic ventricular tachycardia
PKP2 *DSP* *DSC2* *TMEM43* *DSG2*	Arrhymthmogenic right ventricular tachycardia
KCNQ1 *KCNH2* *SCN5A*	Romano-Ward long-QT syndrome types 1, 2, and 3, Brugada syndrome
LDLR *APOB* *PCSK9*	Familial hypercholesterolemia
ATP7B	Wilson disease
OTC	Ornithine transcarbamylase deficiency
RYR1 *CACNA1S*	Malignant hyperthermia susceptibility