Ambry Genetics®

ACMG RECOMMENDED SECONDARY FINDINGS LIST

SUPPLEMENTAL TEST FORM December 2016

GENE	DISORDER
BRCA1, BRCA2	Hereditary breast and ovarian cancer
TP53	Li-Fraumeni syndrome
STK11	Peutz-Jeghers syndrome
MLH1, MSH2, MSH6, PMS2	Lynch syndrome
APC	Familial adenomatous polyposis
MUTYH	MYH-associated polyposis; Adenomas, multiple colorectal, FAP type 2; Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas
BMPR1A, SMAD4	Juvenile polyposis
VHL	von Hippel Lindau syndrome
MEN1	Multiple endocrine neoplasia type 1
RET	Multiple endocrine neoplasia type 2
RET	Familial medullary thyroid cancer (FMTC)
PTEN	PTEN hamartoma tumor syndrome
RB1	Retinoblastoma
SDHD, SDHAF2, SDHC, SDHB	Hereditary paraganglioma- pheochromocytoma syndrome
TSC1, TSC2	Tuberous sclerosis complex
WT1	*WT1*-related Wilms
NF2	Neurofibromatosis type 2
COL3A1	Ehlers-Danlos syndrome, vascular type
FBN1, TGFBR1, TGFBR2, SMAD3, ACTA2, MYH11	Marfan syndrome, Loeys-Dietz syndromes, Familial thoracic aortic aneurysms and dissections
MYBPC3, MYH7, TNNT2, TNNI3, TPM1, MYL3, ACTC1, PRKAG2, GLA, MYL2, LMNA	Hypertrophic cardiomyopathy, Dilated cardiomyopathy
RYR2	Catecholaminergic polymorphic ventricular tachycardia
PKP2, DSP, DSC2, TMEM43, DSG2	Arrhythmogenic right ventricular cardiomyopathy
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

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