Laboratory of Personalized Genomic Medicine
Department of Pathology and Cell Biology

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American College of Medical Genetics and Genomics Secondary Findings List	
Disorder	**Gene**
Hereditary Breast and Ovarian Cancer	BRCA1, BRCA2
Li-Fraumeni Syndrome	TP53
Peutz-Jeghers Syndrome	STK11
Lynch Syndrome	MLH1, MSH2, MSH6, PMS2
Familial adenomatous polyposis	APC
MYH-Associated Polypsosis; Adenomas, multiple colorectal, FAP type 2; Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas	MUTYH
Juvenile Polyposis	BMPR1A, SMAD4
Von Hippel Lindau Syndrome	VHL
Multiple Endocrine Neoplasia Type 1	MEN1
Multiple Endocrine Neoplasia Type 2	RET
Familial Medullary Thyroid Cancer (FMTC)	RET
PTEN Hamartoma Tumor Syndrome	PTEN
Retinoblastoma	RB1
Hereditary Paraganlioma-Pheochromocytome Syndrome	SDHD, SDHAF2, SDHC, SDHB
Tuberous Sclerosis Complex	TSC1, TSC2
WT1-related Wilms Tumor	WT1
Neurofibromatosis Type 2	NF2
EDS-Vascular type	COL3A1
Marfan Syndrome, Loeys-Dietz Syndromes, and Familial. Thoracic Aortic Aneurysms and Dissections	FBN1, TGFBR1, TGFBR2, SMAD3, ACTA2, MYH11
Hypertrophic Cardiomyopathy, Dilated cardiomyopathy	MYBPC3, MYH7, TNNT2, TNNI3, TPM1, MYL3, ACTC1, PRKAG2, GLA, MYL2, LMNA
Catecholamineric polymorphic ventricular tachycardia	RYR2
Arryhmogenic right ventricular cardiomyopathy	PKP2, DSP, DSC2, TMEM43, DSG2
Romano-Ward Long QT Syndrome Types 1,2 and 3, Brugada Syndrome	KCNQ1, KCNH2, SCN5A
Wilsons Disease	ATP7B
Ornithine Transcarbamylase Deficiency	OTC
Familial Hypercholesterolemia	LDLR, APOB, PCSK9
Malignant Hyperthemia susceptibility	RYR1, CACNA1S