

Germline Incidental Findings

CANCER RELATED			
APC	APC-associated conditions	PALB2	Breast cancer susceptibility, Pancreatic cancer susceptibility, Ovarian cancer susceptibility, Fanconi anemia
ATM	Ataxia-Telangiectasia, Breast cancer susceptibility, Pancreatic cancer susceptibility	PDGFRA	Familial gastrointestinal stromal tumor, GIST-plus syndrome
AXIN2	Oligodontia-colorectal cancer syndrome	PHOX2B	Neuroblastoma susceptibility
BAP1	BAP1 tumor predisposition syndrome	PMS2	Lynch syndrome, Constitutional mismatch repair deficiency
BARD1	Breast cancer susceptibility	PRKAR1A	Polymerase proofreading-associated polyposis
BLM	Bloom syndrome	POLD1*	Polymerase proofreading-associated polyposis
BMPR1A	Juvenile polyposis	POLE*	Carney complex
BRCA1	Hereditary breast and ovarian cancer	PTCH1	Gorlin syndrome, Basal cell nevus syndrome
BRCA2	Hereditary breast and ovarian cancer, Fanconi anemia	PTEN	PTEN hamartoma tumor syndrome
BRIP1	Ovarian cancer susceptibility, Fanconi anemia	RAD51C	Ovarian cancer susceptibility, Breast cancer susceptibility, Fanconi anemia
CDH1	Hereditary diffuse gastric cancer, Breast cancer susceptibility	RAD51D	Ovarian cancer susceptibility, Breast cancer susceptibility
CDK4	Melanoma susceptibility	RB1	Retinoblastoma
CDKN2A	Melanoma-pancreatic cancer syndrome	RET	Multiple endocrine neoplasia type 2, Familial medullary thyroid cancer
CEBPA	Acute myeloid leukemia susceptibility	RUNX1	Acute myeloid leukemia susceptibility
CHEK2	Breast cancer susceptibility, Colon cancer susceptibility	SDHA	Hereditary paraganglioma-pheochromocytoma syndrome
DICER1	DICER1 tumor predisposition syndrome	SDHAF2	Hereditary paraganglioma-pheochromocytoma syndrome
EGFR*	Lung cancer susceptibility, TKI resistance	SDHB	Hereditary paraganglioma-pheochromocytoma syndrome
EPCAM*	Lynch syndrome	SDHC	Hereditary paraganglioma-pheochromocytoma syndrome
ETV6	Leukemia susceptibility, thrombocytopenia susceptibility	SDHD	Hereditary paraganglioma-pheochromocytoma syndrome
FH	Hereditary leiomyomatosis and renal cell cancer	SMAD4	Juvenile polyposis, Hereditary hemorrhagic telangiectasia
FLCN	Birt-Hogg-Dube syndrome	SMARCA4	Rhabdoid tumor predisposition syndrome
GATA2	GATA2 deficiency with susceptibility to myeloid malignancies	SMARCB1	Rhabdoid tumor predisposition syndrome, Schwannomatosis
KIT	Familial gastrointestinal stromal tumor	STK11	Peutz-Jeghers syndrome
MAX	Hereditary paraganglioma-pheochromocytoma syndrome	SUFU	Gorlin syndrome, Basal cell nevus syndrome
MEN1	Multiple endocrine neoplasia type 1	TMEM127	Hereditary paraganglioma-pheochromocytoma syndrome
MET	Hereditary papillary renal cell carcinoma	TP53	Li-Fraumeni syndrome
MLH1	Lynch syndrome, Constitutional mismatch repair deficiency	TSC1	Tuberous sclerosis complex
MSH2	Lynch syndrome, Constitutional mismatch repair deficiency	TSC2	Tuberous sclerosis complex
MSH3	MSH3-associated polyposis	VHL	Von Hippel-Lindau syndrome
MSH6	Lynch syndrome, Constitutional mismatch repair deficiency	WT1	WT1-related Wilms tumor
MUTYH	MUTYH-associated polyposis		
NBN	Nijmegen breakage syndrome, Breast cancer susceptibility		
NF1	Neurofibromatosis type 1		
NF2	Neurofibromatosis type 2		
NTHL1	NTHL1 tumor syndrome, NTHL1-associated polyposis		

NON-CANCER RELATED

ACTA2	Familial thoracic aortic aneurysms and dissections	MYH11	Familial thoracic aortic aneurysms and dissections
ACTC1	Hypertrophic cardiomyopathy, Dilated cardiomyopathy, Left ventricular noncompaction	MYH7	Hypertrophic cardiomyopathy, Dilated cardiomyopathy
ACVRL1	Hereditary hemorrhagic telangiectasia	MYL2	Hypertrophic cardiomyopathy, Dilated cardiomyopathy
APOB	Familial hypercholesterolemia	MYL3	Hypertrophic cardiomyopathy, Dilated cardiomyopathy
ATP7B	Wilson disease	OTC	Ornithine transcarbamylase deficiency
BTD	Biotinidase deficiency	PCSK9	Familial hypercholesterolemia
CACNA1S	Malignant hyperthermia susceptibility	PKP2	Arrhythmogenic right ventricular cardiomyopathy
CASQ2	Catecholaminergic polymorphic ventricular tachycardia	PRKAG2	Hypertrophic cardiomyopathy, Dilated cardiomyopathy
COL3A1	Ehlers-Danlos syndrome, vascular type	RPE65	Leber congenital amaurosis, Retinitis pigmentosa
DSC2	Arrhythmogenic right ventricular cardiomyopathy	RYR1	Malignant hyperthermia susceptibility
DSG2	Arrhythmogenic right ventricular cardiomyopathy	RYR2	Catecholaminergic polymorphic ventricular tachycardia
DSP	Arrhythmogenic right ventricular cardiomyopathy	SCN5A	Long QT syndrome, Brugada syndrome
ENG	Hereditary hemorrhagic telangiectasia	SMAD3	Loeys-Dietz syndrome, Familial thoracic aortic aneurysms and dissections
FBN1	Marfan syndrome, Loeys-Dietz syndrome, Familial thoracic aortic aneurysms and dissections	TGFBR1	Loeys-Dietz syndrome, Familial thoracic aortic aneurysms and dissections
FLNC	Dilated cardiomyopathy, Hypertrophic cardiomyopathy, Restrictive cardiomyopathy, Myofibrillar myopathy, Distal myopathy	TGFBR2	Loeys-Dietz syndrome, Familial thoracic aortic aneurysms and dissections
GAA	Pompe disease, Glycogen storage disease type II	TMEM43	Arrhythmogenic right ventricular cardiomyopathy
GLA	Hypertrophic cardiomyopathy, Dilated cardiomyopathy, Fabry disease	TNNI3	Hypertrophic cardiomyopathy, Dilated cardiomyopathy
HFE*	Hereditary hemochromatosis	TNNT2	Hypertrophic cardiomyopathy, Dilated cardiomyopathy
HNF1A	Maturity-onset diabetes of the young	TPM1	Hypertrophic cardiomyopathy, Dilated cardiomyopathy
KCNH2	Long QT syndrome, Short QT syndrome	TRDN	Catecholaminergic polymorphic ventricular tachycardia, Long QT syndrome
KCNQ1	Long QT syndrome, Short QT syndrome, Jervell and Lange-Nielsen syndrome	TTN*	Dilated cardiomyopathy
LDLR	Familial hypercholesterolemia		
LMNA	Hypertrophic cardiomyopathy, Dilated cardiomyopathy		
MYBPC3	Hypertrophic cardiomyopathy, Dilated cardiomyopathy		

This list is composed of genes selected based on recommendations from the American College of Medical Genetics (ACMG), the National Comprehensive Cancer Network (NCCN) and/or published literature. The primary focus of the xE panel is somatic reporting. Tempus also offers a separately ordered validated germline hereditary cancer panel through GeneDx.

***Special reporting** – EGFR: p.T790M, p.L792H, p.C797G, p.C797S (resistance alterations only); EPCAM: Large deletions only; HFE: p.C282Y only; POLD1: Exonuclease domain only; POLE: Exonuclease domain only; TTN: Truncating variants in A-band (exons 252-357) only.