Germline Incidental Findings

CANCER RELATED

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

NON-CANCER RELATED

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