Reportable Germline Genes

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
BMPR1A	Juvenile polyposis syndrome
BRCA1	Hereditary breast and ovarian cancer syndrome, Fanconi anemia
BRCA2	Hereditary breast and ovarian cancer syndrome, Fanconi anemia
BRIP1	Ovarian cancer susceptibility, Fanconi anemia
CDH1	Hereditary diffuse gastric cancer syndrome
CDK4	Melanoma susceptibility
CDKN2A	Familial atypical multiple mole-melanoma 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	MDS susceptibility, Leukemia susceptibility, Thrombocytopenia susceptibility
FH	Hereditary leiomyomatosis and renal cell cancer syndrome, Fumarate hydratase deficiency
FLCN	Birt-Hogg-Dube syndrome
GATA2	GATA2 deficiency with susceptibility to myeloid malignancies
KIT	Gastrointestinal stromal tumor susceptibility
MAX	Hereditary paraganglioma-pheochromocytoma syndrome
MEN1	Multiple endocrine neoplasia type 1, Familial isolated hyperparathyroidism
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
NF1	Neurofibromatosis type 1
NF2	NF2-related schwannomatosis
NTHL1	NTHL1 tumor syndrome
PALB2	Breast cancer susceptibility, Pancreatic cancer susceptibility, Ovarian cancer susceptibility, Fanconi anemia
PDGFRA	GIST-plus syndrome
PHOX2B	Congenital central hypoventilation syndrome, 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 susceptibility
RET	Multiple endocrine neoplasia type 2, Familial medullary thyroid cancer
RUNX1	RUNX1 familial platelet disorder, Myeloid malignancy susceptibility
SDHA	Hereditary paraganglioma-pheochromocytoma syndrome, Leigh syndrome
SDHAF2	Hereditary paraganglioma-pheochromocytoma syndrome
SDHB	Hereditary paraganglioma-pheochromocytoma syndrome, Mitochondrial complex II deficiency
SDHC	Hereditary paraganglioma-pheochromocytoma syndrome
SDHD	Hereditary paraganglioma-pheochromocytoma syndrome
SMAD4	Juvenile polyposis, Hereditary hemorrhagic telangiectasia
SMARCA4	Rhabdoid tumor predisposition syndrome, Coffin-Siris syndrome
SMARCB1	Rhabdoid tumor predisposition syndrome, Schwannomatosis, Coffin-Siris syndrome
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 Disorder

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
BAG3	Dilated cardiomyopathy, Myofibrillar myopathy
BTD	Biotinidase deficiency
CACNA1S	Malignant hyperthermia susceptibility
CALM1	Long QT syndrome
CALM2	Long QT syndrome
CALM3	Long QT syndrome
CASQ2	Catecholaminergic polymorphic ventricular tachycardia
COL3A1	Ehlers-Danlos syndrome, vascular type
DES	Dilated cardiomyopathy, Myofibrillar myopathy
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
RBM20	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
TNNC1	Hypertrophic cardiomyopathy, Dilated 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
TTR	Hereditary TTR amyloidosis

This list is composed of genes selected 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 separately ordered, validated germline hereditary cancer panels.

***Special reporting** – 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.