Invitae Genetic Health Screen Gene List



INVITAE GENETIC HEALTH SCREEN

	Gene(s)	Condition(s)
	APC	Colorectal, endocrine, gastric, nervous system/brain, and pancreatic cancer, sarcoma
	ATM	Breast, pancreatic, and prostate cancer
	AXIN2	Colorectal cancer
	BAP1	Renal/urinary tract cancer, melanoma
	BARD1	Breast cancer
	BMPR1A	Colorectal, gastric, and pancreatic cancer
	BRCA1	Breast, gynecologic, pancreatic, and prostate cancer
	BRCA2	Breast, gynecologic, pancreatic, and prostate cancer, melanoma
	BRIP1	Breast and gynecologic cancer
	CDC73	Endocrine and renal/urinary tract cancer
	CDH1	Breast, colorectal, and gastric cancer
	CDK4	Melanoma
	CDKN2A	Nervous system/brain and pancreatic cancer, melanoma
Z	CHEK2	Breast, colorectal, endocrine, gynecologic, and prostate cancer
R SCR	DICER1	Endocrine, gynecologic, nervous system/brain, and renal/urinary tract cancer, sarcoma
INVITAE CANCER SCREEN	EPCAM	Colorectal, gastric, gynecologic, nervous system/ brain, pancreatic, prostate, and renal/urinary tract cancer
Y E	FH	Renal/urinary tract cancer, sarcoma
<u>></u>	FLCN	Renal/urinary tract cancer
=	GREM1	Colorectal cancer
	HOXB13	Prostate cancer
	KIT	Gastric cancer, sarcoma
	MAX	Endocrine cancer
	MEN1	Endocrine, nervous system/brain, and pancreatic cancer
	MET	Renal/urinary tract cancer
	MITF	Melanoma
	MLH1	Colorectal, gastric, gynecologic, nervous system/ brain, pancreatic, prostate, and renal/urinary tract cancer
	MSH2	Colorectal, gastric, gynecologic, nervous system/ brain, pancreatic, prostate, and renal/urinary tract cancer
	MSH3	Colorectal cancer, includes reporting of carrier status
	MSH6	Colorectal, gastric, gynecologic, nervous system/ brain, pancreatic, prostate, and renal/urinary tract cancer
	MUTYH	Colorectal cancer

Gene(s)	Condition(s)			
NBN	Breast and prostate cancer			
NF1	Breast, endocrine, gastric, and nervous system/brain cancer			
NF2	Nervous system/brain cancer			
NTHL1	Colorectal cancer, includes reporting of carrier status			
PALB2	Breast and pancreatic cancer			
PDGFRA	Gastric cancer, sarcoma			
PMS2	Colorectal, gastric, gynecologic, nervous system/ brain, pancreatic, prostate, and renal/urinary tract cancer			
POLD1	Colorectal cancer			
POLE	Colorectal cancer			
PRKAR1A	Endocrine and nervous system/brain cancer, sarcoma			
PTCH1	Nervous system/brain and skin cancer, sarcoma			
PTEN	Breast, colorectal, endocrine, gynecologic, nervous system/brain and, renal/urinary tract cancer, melanoma			
RAD51C	Breast and gynecologic cancer			
RAD51D	Breast and gynecologic cancer			
RB1	Melanoma, retinoblastoma, sarcoma			
RET	Endocrine cancer			
SDHA	Endocrine and gastric cancer, sarcoma			
SDHAF2	Endocrine cancer			
SDHB	Endocrine, gastric, and renal/urinary tract cancer, sarcoma			
SDHC	Endocrine, gastric, and renal/urinary tract cancer, sarcoma			
SDHD	Endocrine, gastric, and renal/urinary tract cancer, sarcoma			
SMAD4	Colorectal, gastric, and pancreatic cancer			
SMARCA4	Gynecologic cancer			
SMARCB1	Nervous system/brain and renal/urinary tract cancer			
STK11	Breast, colorectal, gastric, gynecologic, and pancreatic cancer			
TMEM127	Endocrine cancer			
TP53	Breast, endocrine, gastrointestinal, genitourinary, gynecologic, hematologic, nervous system/brain, and skin cancer, sarcoma			
TSC1	Nervous system/brain, pancreatic, and renal/urinary tract cancer			
TSC2	Nervous system/brain, pancreatic, and renal/urinary tract cancer			
VHL	Endocrine, nervous system/brain, pancreatic, and renal/urinary tract cancer			
WT1	Renal/urinary tract cancer			

Invitae Genetic Health Screen Gene List



	Gene(s)	Condition(s)
	ACTA2	Aortopathy
	ACTC1	Cardiomyopathy, congenital heart disease
	ACTN2	Arrhythmia, cardiomyopathy
	ACVRL1	Hereditary hemorrhagic telangiectasia, pulmonary arterial hypertension
	APOB	Familial hypercholesterolemia, familial hypobetalipoproteinemia
	BAG3	Cardiomyopathy, neuromuscular condition
	BMPR2	Pulmonary arterial hypertension
	CACNA1C	Arrhythmia, cardiomyopathy, congenital heart disease
	CACNB2	Arrhythmia
	CALM1	Arrhythmia
	CALM2	Arrhythmia
	CALM3	Arrhythmia
	CASQ2	Arrhythmia, includes reporting of carrier status
	CAV1	Pulmonary arterial hypertension
Z	CAV3	Arrhythmia, cardiomyopathy, neuromuscular condition
RE	COL3A1	Aortopathy
50	CRYAB	Cardiomyopathy, neuromuscular condition
INVITAE CARDIO SCREEN	CSRP3	Cardiomyopathy
AR	DES	Arrhythmia, cardiomyopathy, neuromuscular condition
Щ	DMD	Cardiomyopathy, neuromuscular condition
ITA	DSC2	Arrhythmia, cardiomyopathy
Ž	DSG2	Arrhythmia, cardiomyopathy
	DSP	Arrhythmia, cardiomyopathy
	EMD	Arrhythmia, cardiomyopathy, neuromuscular condition
	ENG	Hereditary hemorrhagic telangiectasia, pulmonary arterial hypertension
	F2	Hereditary thrombophilia
	F5	Hereditary thrombophilia
	F9	Hemophilia, hereditary thrombophilia
	FBN1	Aortopathy
	FHL1	Cardiomyopathy, neuromuscular condition
	FLNC	Cardiomyopathy, neuromuscular condition
	GDF2	Hereditary hemorrhagic telangiectasia
	GLA	Cardiomyopathy, lysosomal storage disease
	GPD1L	Arrhythmia
	HCN4	Arrhythmia, cardiomyopathy
	JUP	Arrhythmia, cardiomyopathy
	KCNE1	Arrhythmia
	KCNE2	Arrhythmia

INVITAE GENETIC HEALTH SCREEN (continued)

KCNH2 Arrhythmia KCNQ1 Arrhythmia KCNQ1 Arrhythmia LAMP2 Cardiomyopathy, glycogen storage disease LDLR Familial hypercholesterolemia LDLRAP1 Familial hypercholesterolemia, includes reporting of carrier status LMNA Arrhythmia, cardiomyopathy, neuromuscular condition MYBPC3 Cardiomyopathy MYH1 Aortopathy MYH7 Cardiomyopathy, neuromuscular condition MYL2 Cardiomyopathy MYL8 Aortopathy MYL8 Aortopathy MYL8 Aortopathy NKX2-5 Arrhythmia, congenital heart disease PCSK9 Familial hypercholesterolemia PKP2 Arrhythmia, cardiomyopathy PLN Arrhythmia, cardiomyopathy PRKG1 Aortopathy PROC Hereditary thrombophilia RBM20 Arrhythmia, cardiomyopathy RYR2 Arrhythmia, cardiomyopathy SCN5A Arrhythmia, cardiomyopathy SCN5A Arrhythmia, cardiomyopathy SERPINC1 Hereditary thrombophilia SGCD Cardiomyopathy, neuromuscular condition SMAD3 Aortopathy SMAD4 Hereditary thrombophilia SGCD Cardiomyopathy, neuromuscular condition SMAD3 Aortopathy TGFB2 Aortopathy TGFB3 Aortopathy, multiple self-healing squamous epithelioma TGFBR2 Aortopathy TMEM43 Arrhythmia, cardiomyopathy TNNC1 Cardiomyopathy TNNC1 Cardiomyopathy TNNC1 Arrhythmia, cardiomyopathy TNNC1 Cardiomyopathy TNNC1 Cardiomyopathy TNNC1 Cardiomyopathy TNNC1 Cardiomyopathy TNNC2 Arrhythmia, cardiomyopathy TNNC1 Cardiomyopathy TNNC1 Cardiomyopathy TNNC1 Cardiomyopathy TNNC1 Cardiomyopathy TNNC1 Cardiomyopathy TNNC1 Cardiomyopathy TNNC2 Cardiomyopathy TNNC1 Cardiomyopathy TNNC2 Cardiomyopathy	Gene(s)	Condition(s)			
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TGFB2 Aortopathy TGFB3 Aortopathy, arrhythmia, cardiomyopathy TGFBR1 Aortopathy, multiple self-healing squamous epithelioma TGFBR2 Aortopathy TMEM43 Arrhythmia, cardiomyopathy TNNC1 Cardiomyopathy TNNI3 Arrhythmia, cardiomyopathy TNNT2 Arrhythmia, cardiomyopathy TNNT2 Cardiomyopathy TPM1 Cardiomyopathy	TCAP				
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TMEM43 Arrhythmia, cardiomyopathy TNNC1 Cardiomyopathy TNNI3 Arrhythmia, cardiomyopathy TNNT2 Arrhythmia, cardiomyopathy TPM1 Cardiomyopathy	TGFBR1	Aortopathy, multiple self-healing squamous			
TNNC1 Cardiomyopathy TNNI3 Arrhythmia, cardiomyopathy TNNT2 Arrhythmia, cardiomyopathy TPM1 Cardiomyopathy	TGFBR2	Aortopathy			
TNNI3 Arrhythmia, cardiomyopathy TNNT2 Arrhythmia, cardiomyopathy TPM1 Cardiomyopathy	TMEM43	Arrhythmia, cardiomyopathy			
TNNI3 Arrhythmia, cardiomyopathy TNNT2 Arrhythmia, cardiomyopathy TPM1 Cardiomyopathy	TNNC1				
TNNT2 Arrhythmia, cardiomyopathy TPM1 Cardiomyopathy	TNNI3				
TPM1 Cardiomyopathy	TNNT2				
	TPM 1				
	VCL				

Invitae Genetic Health Screen Gene List



-	Gene(s)	Condition(s)
	ATP7B	Wilson disease, includes reporting of carrier status
GENETIC HEALTH EN (continued)	CACNA1S	Hypokalemic periodic paralysis, malignant hyperthermia susceptibility
SENE N (co	HAMP	Hereditary hemochromatosis, includes reporting of carrier status
AE	HFE	Hereditary hemochromatosis, includes reporting of carrier status
INVII	HJV	Hereditary hemochromatosis, includes reporting of carrier status

Gene(s)	Condition(s)	
OTC	Ornithine transcarbamylase deficiency	
RYR1	Malignant hyperthermia susceptibility, neuromuscular condition	
SERPINA1	Alpha-1 antitrypsin deficiency, includes reporting of carrier status	
SLC40A1	Hereditary hemochromatosis	
TFR2	Hereditary hemochromatosis, includes reporting of carrier status	