Invitae Genetic Health Screen Gene List

INVITAE

INVITAE GENETIC HEALTH SCREEN

INVITAE CANCER 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
CHEK2	Breast, colorectal, endocrine, gynecologic, and prostate cancer
DICER1	Endocrine, gynecologic, nervous system/brain, and renal/urinary tract cancer, sarcoma
EPCAM	Colorectal, gastric, gynecologic, nervous system/brain, pancreatic, prostate, and renal/urinary tract cancer
FH	Renal/urinary tract cancer, sarcoma
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

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INVITAE GENETIC HEALTH SCREEN (continued)

INVITAE CARDIO SCREEN

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
CAV3	Arrhythmia, cardiomyopathy, neuromuscular condition
COL3A1	Aortopathy
CRYAB	Cardiomyopathy, neuromuscular condition
CSRP3	Cardiomyopathy
DES	Arrhythmia, cardiomyopathy, neuromuscular condition
DMD	Cardiomyopathy, neuromuscular condition
DSC2	Arrhythmia, cardiomyopathy
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

Gene(s)	Condition(s)
KCNH2	Arrhythmia
KCNJ2	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
MYH11	Aortopathy
MYH7	Cardiomyopathy, neuromuscular condition
MYL2	Cardiomyopathy
MYL3	Cardiomyopathy
MYLK	Aortopathy
NKX2-5	Arrhythmia, congenital heart disease
PCSK9	Familial hypercholesterolemia
PKP2	Arrhythmia, cardiomyopathy
PLN	Arrhythmia, cardiomyopathy
PRKAG2	Arrhythmia, cardiomyopathy
PRKG1	Aortopathy
PROC	Hereditary thrombophilia
PROS1	Hereditary thrombophilia
RBM20	Arrhythmia, cardiomyopathy
RYR2	Arrhythmia, cardiomyopathy
SCN5A	Arrhythmia, cardiomyopathy
SERPINC1	Hereditary thrombophilia
SGCD	Cardiomyopathy, neuromuscular condition
SMAD3	Aortopathy
SMAD4	Hereditary hemorrhagic telangiectasia
TCAP	Cardiomyopathy, neuromuscular condition
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
TPM1	Cardiomyopathy
VCL	Cardiomyopathy

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INVITAE GENETIC HEALTH SCREEN (continued)

Gene(s)	Condition(s)
ATP7B	Wilson disease, includes reporting of carrier status
CACNA1S	Hypokalemic periodic paralysis, malignant hyperthermia susceptibility
HAMP	Hereditary hemochromatosis, includes reporting of carrier status
HFE	Hereditary hemochromatosis, includes reporting of carrier status
HJV	Hereditary hemochromatosis, includes reporting of carrier status
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

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