PREVENTATIVE HEALTH SCREENING

Gene list and conditions covered

Eugene's preventative health panel: 184 genes across cancer risk, heart health and broader preventative health, for adult-onset conditions where early action can change outcomes.

About the preventative health screening test

A screen for adult-onset, medically actionable genetic conditions, identifying inherited risk across cancer, heart health and other areas where early detection or management can change outcomes.

This panel covers 184 genes across cancer risk, heart health and broader preventative health. Genes were selected for conditions where a positive result can guide screening, prevention or early treatment.

Variants are classified using ACMG / AMP guidelines and reported at pathogenic or likely-pathogenic levels. Individuals with a positive result are offered a genetic-counselling consultation.

This document is intended for clinicians. A negative result reduces, but does not eliminate, genetic risk. Panel composition is reviewed periodically.

Cancer	Hereditary cancer predisposition: inherited risk of cancers such as breast, ovarian, bowel and others.
Heart & circulation	Inherited conditions affecting the heart, blood vessels and circulation, including cardiomyopathy, arrhythmia, aortopathy and inherited high cholesterol.
Blood & immune	Conditions affecting the blood or the immune system.
Brain, nerve & muscle	Conditions affecting the brain, nerves or muscles.
Liver & kidney	Inherited conditions affecting the liver or kidneys, including polycystic kidney disease.
Hormones & metabolism	Conditions affecting the body's hormones and how it processes energy and nutrients.
Skin, bones & tissue	Conditions affecting the skin, bones or connective tissue.

AT A GLANCE

Total genes	184
Categories	7

CONDITIONS BY CATEGORY

Cancer

68 genes

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
CDKN1B	Prostate cancer
CDKN2A	Nervous system/brain and pancreatic cancer, melanoma
CHEK2	Breast, colorectal, endocrine, gynecologic, and prostate cancer
CTNNA1	Stomach and breast cancer
DDX41	Haematological cancer
DICER1	Endocrine, gynecologic, nervous system/brain, and renal/urinary tract cancer, sarcoma
EGFR	Lung cancer
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
LZTR1	Schwannomatosis
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
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
POT1	Melanoma, leukaemia
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
RNF43	Polyposis
RUNX1	Haematological 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
SUFU	Nervous system 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

Heart and circulation

73 genes

ACTA2	Aortopathy
ACTC1	Cardiomyopathy
ACTN2	Cardiomyopathy
ACVRL1	High blood pressure
APOA1	Cardiovascular disease
APOB	Hypercholesterolaemia
BAG3	Aortopathy, Arrhythmia
BMPR2	High blood pressure
CACNA1C	Arrhythmia, Cardiomyopathy
CALM1	Arrhythmia
CALM2	Arrhythmia
CALM3	Arrhythmia
CASQ2	Arrhythmia
CAV1	High blood pressure
CAV3	Cardiomyopathy
CRYAB	Cardiomyopathy
CSRP3	Cardiomyopathy
DES	Arrhythmia, Cardiomyopathy
DMD	Cardiomyopathy
DSC2	Arrhythmia, Cardiomyopathy
DSG2	Arrhythmia, Cardiomyopathy
DSP	Arrhythmia, Cardiomyopathy
ELN	Connective tissue, aortopathy
EMD	Arrhythmia, Cardiomyopathy
ENG	High blood pressure
FBN1	Aortopathy
FHL1	Cardiomyopathy
FLNC	Arrhythmia, Cardiomyopathy
GDF2	Arrhythmia, Cardiomyopathy
GLA	High blood pressure
HCN4	Arrhythmia
JUP	Arrhythmia, Cardiomyopathy
KCNE1	Arrhythmia
KCNH2	Arrhythmia
KCNJ2	Arrhythmia
KCNK3	Pulmonary arterial hypertension
KCNQ1	Arrhythmia
LAMP2	Cardiomyopathy
LDLR	Hypercholesterolaemia
LDLRAP1	Hypercholesterolaemia
LMNA	Arrhythmia, Cardiomyopathy
LOX	Aortopathy
MYBPC3	Cardiomyopathy
MYH11	Aortopathy
MYH7	Cardiomyopathy
MYL2	Cardiomyopathy
MYL3	Cardiomyopathy
MYLK	Cardiomyopathy
NKX2-5	Cardiomyopathy
PCSK9	Hypercholesterolaemia
PKP2	Arrhythmia, Cardiomyopathy
PLN	Arrhythmia, Cardiomyopathy
PRKAG2	Arrhythmia, Cardiomyopathy

SCN5A	Arrhythmia, Cardiomyopathy
SMAD3	Aortopathy
SMAD9	Pulmonary hypertension, primary, 2
TECRL	Arrhythmia
TGFB2	Aortopathy
TGFB3	Aortopathy
TGFBR1	Aortopathy
TGFBR2	Aortopathy
TMEM43	Cardiomyopathy
TNNC1	Cardiomyopathy
TNNI3	Cardiomyopathy
TNNT2	Cardiomyopathy
TPM1	Cardiomyopathy
TRDN	Catecholaminergic Polymorphic Ventricular Tachycardia
TTN	TTN-cardiomyopathy
TTR	TTR-cardiomyopathy - Hereditary Transthyretin Amyloidosis
VCL	Cardiomyopathy

Blood and immune health

10 genes

ANKRD26	Thrombocytopenia
ETV6	Thrombocytopenia
F11	Thrombophilia
F2	Thrombophilia
F5	Thrombophilia
F9	Haemophilia
MEFV	Familial Mediterranean Fever
PROC	Thrombophilia
PROS1	Thrombophilia
SERPINC1	High blood pressure

Brain, nerve and muscle health

4 genes

ABCD1	X-Linked Adrenoleukodystrophy
CACNA1S	Hypokalemic periodic paralysis (Malignant hyperthermia)
GCH1	Dystonia
RYR1	Central core disease (CCD) (Malignant hyperthermia)

Liver and kidney health

12 genes

ATP7B	Wilson disease
COL4A3	Alport syndrome
COL4A4	Alport syndrome
COL4A5	Alport syndrome
HAMP	Hemochromatosis type 2
HFE	Hemochromatosis
HJV	Hemochromatosis type 2A
PKD1	Polycystic kidney disease
PKD2	Polycystic kidney disease
SERPINA1	Alpha-1antitrypsin deficiency (AATD)
SLC40A1	Hemochromatosis type 4
TFR2	Hemochromatosis type 3

Hormones and metabolism

12 genes

AIP	Pituitary adenomas
BCHE	Butyrylcholinesterase deficiency
BTD	Biotinidase deficiency
CBS	Homocystinuria
G6PD	G6PD deficiency
GAA	Glycogen storage disease - Pompe disease
GCK	Maturity onset diabetes of the young
HMBS	Acute intermittent porphyria
HNF1A	Monogenic diabetes MODYIII
HNF1B	MODY5
HNF4A	Maturity onset diabetes of the young
OTC	Ornithine transcarbamylase deficiency

Skin, bones and connective tissue

5 genes

COL1A1	Vascular Ehlers-Danlos syndrome | Classical Ehlers-Danlos syndrome | Osteogenesis imperfecta
COL3A1	Vascular Ehlers-Danlos syndrome
COL5A1	Classical Ehlers-Danlos Syndrome
COL5A2	Classical Ehlers-Danlos Syndrome
CYLD	CYLD cutaneous syndrome