This table lists the 160 genes included on the ASPIRE genetic screening panel performed by Inviate laboratory

Gene	Relevant condition's name and their characteristics
Genes Related to Cancer Predisposition	
APC	APC-associated polyposis; Colorectal, endocrine, gastric, nervous system/brain, and pancreatic cancer, sarcoma
ATM	ATM-associated cancer/Ataxia–telangiectasia; Breast, pancreatic, and prostate cancer
AXIN2	AXIN2-related oligodontia–colorectal cancer syndrome; Colorectal cancer
BAP1	BAP1 tumor predisposition syndrome; Renal/urinary tract cancer, melanoma
BARD1	BARD1-associated hereditary breast cancer; Breast Cancer
BMPR1A	Juvenile polyposis syndrome; Colorectal, gastric, and pancreatic cancer
BRCA1	Hereditary breast and/or ovarian cancer; Breast, gynecologic, pancreatic, and prostate cancer
BRCA2	Hereditary breast and/or ovarian cancer; Breast, gynecologic, pancreatic, and prostate cancer, melanoma
BRIP1	BRIP1-associated hereditary ovarian cancer risk; Breast and gynecologic cancer
CDC73	Hyperparathyroidism–jaw tumor syndrome; Endocrine and renal/urinary tract cancer
CDH1	Hereditary diffuse gastric cancer; Breast, colorectal, and gastric cancer
CDK4	Familial melanoma; Melanoma
CDKN1B	Multiple endocrine neoplasia type 4; Endocrine cancer
CDKN2A	Familial atypical multiple mole melanoma; Nervous system/brain and pancreatic cancer, melanoma
CHEK2	CHEK2-associated hereditary cancer predisposition; Breast, colorectal, endocrine, gynecologic, and prostate cancer
DICER1	DICER1 tumor predisposition syndrome; Endocrine, gynecologic, nervous system/brain, and renal/ urinary tract cancer, sarcoma
EGFR	EGFR-related familial lung adenocarcinoma susceptibility; Lung cancer
EPCAM	Lynch syndrome; Colorectal, gastric, gynecologic, nervous system/brain, pancreatic, prostate, and renal/urinary tract cancer
FH	Hereditary leiomyomatosis and renal cell carcinoma; Renal/urinary tract cancer, sarcoma
FLCN	Birt–Hogg–Dubé syndrome; Renal/urinary tract cancer
GREM1	Hereditary mixed polyposis syndrome; Colorectal cancer
HOXB13	Hereditary prostate cancer; Prostate cancer
KIT	Familial gastrointestinal stromal tumor; Gastric cancer, sarcoma
LZTR1	Schwannomatosis (LZTR1-related); Nervous system/brain cancer
MAX	Hereditary paraganglioma-pheochromocytoma syndrome; Endocrine cancer
MEN1	Multiple endocrine neoplasia type 1; Endocrine, nervous system/brain, and pancreatic cancer
MET	Hereditary papillary renal cell carcinoma; Renal/urinary tract cancer
MITF	MITF-associated melanoma susceptibility; Melanoma
MLH1	Lynch syndrome; Colorectal, gastric, gynecologic, nervous system/brain, pancreatic, prostate, and renal/urinary tract cancer
MSH2	Lynch syndrome; Colorectal, gastric, gynecologic, nervous system/brain, pancreatic, prostate, and renal/urinary tract cancer
MSH3	MSH3-associated polyposis; Colorectal cancer
MSH6	Lynch syndrome; Colorectal, gastric, gynecologic, nervous system/brain, pancreatic, prostate, and renal/urinary tract cancer
MUTYH	MUTYH-associated polyposis; Colorectal cancer
NF1	Neurofibromatosis type 1; Breast, endocrine, gastric, and nervous system/brain cancer
NF2	Neurofibromatosis type 2-related schwannomatosis; Nervous system/brain cancer
NTHL1	NTHL1-associated tumor syndrome; Colorectal cancer
PALB2	Hereditary breast and/or ovarian cancer; Breast and pancreatic cancer
PDGFRA	Familial gastrointestinal stromal tumor; Gastric cancer, sarcoma

PMS2	Lynch syndrome; Colorectal, gastric, gynecologic, nervous system/brain, pancreatic, prostate, and renal/urinary tract cancer
POLD1	Polymerase proofreading-associated polyposis; Colorectal cancer
POLE	Polymerase proofreading-associated polyposis; Colorectal cancer
POT1	POT1 tumor predisposition; Melanoma/brain cancer
PRKAR1A	Carney complex; Endocrine and nervous system/brain cancer, sarcoma
PTCH1	Basal cell nevus syndrome; Nervous system/brain and skin cancer, sarcoma
PTEN	PTEN hamartoma tumor syndrome; Breast, colorectal, endocrine, gynecologic, nervous system/ brain and, renal/urinary tract cancer, melanoma
RAD51C	RAD51C-associated hereditary breast/ovarian cancer; Breast and gynecologic cancer
RAD51D	RAD51D-associated hereditary breast/ovarian cancer; Breast and gynecologic cancer
RB1	Retinoblastoma; Melanoma, retinoblastoma, sarcoma
RET	Familial medullary thyroid cancer/ Multiple endocrine neoplasia 2; Endocrine cancer
SDHA	Hereditary paraganglioma-pheochromocytoma syndrome; Endocrine and gastric cancer, sarcoma
SDHAF2	Hereditary paraganglioma-pheochromocytoma syndrome; Endocrine cancer
SDHB	Hereditary paraganglioma-pheochromocytoma syndrome; Endocrine, gastric, and renal/urinary tract cancer, sarcoma
SDHC	Hereditary paraganglioma-pheochromocytoma syndrome; Endocrine, gastric, and renal/urinary tract cancer, sarcoma
SDHD	Hereditary paraganglioma-pheochromocytoma syndrome; Endocrine, gastric, and renal/urinary tract cancer, sarcoma
SMAD4	Juvenile polyposis syndrome/hereditary hemorrhagic telangiectasia syndrome; Colorectal, gastric, and pancreatic cancer
SMARCA4	Rhabdoid tumor predisposition syndrome type 2; Gynecologic cancer
SMARCB1	Rhabdoid tumor predisposition syndrome type 1; Schwannomatosis; Nervous system/brain and renal/urinary tract cancer
STK11	Peutz-Jeghers syndrome; Breast, colorectal, gastric, gynecologic, and pancreatic cancer
TMEM127	Hereditary paraganglioma-pheochromocytoma syndrome; Endocrine cancer
TP53	Li-Fraumeni syndrome; Breast, endocrine, gastrointestinal, genitourinary, gynecologic, hematologic, nervous system/brain, and skin cancer, sarcoma
TSC1	Tuberous sclerosis complex; Nervous system/brain, pancreatic, and renal/urinary tract cancer
TSC2	Tuberous sclerosis complex; Nervous system/brain, pancreatic, and renal/urinary tract cancer
VHL	Von Hippel-Lindau syndrome; Endocrine, nervous system/brain, pancreatic, and renal/ urinary tract cancer
WT1	WT1-related Wilms tumor; Renal/urinary tract cancer
Genes Related to Cardiovascular Disease Predisposition	
ACTA2	ACTA2-related arteriopathy/Heritable thoracic aortic disease; Aortopathy
ACTC1	ACTC1-related cardiomyopathy; Cardiomyopathy, congenital heart disease
ACTN2	ACTN2-related cardiomyopathy/arrhythmia; Arrhythmia, cardiomyopathy
APOB	Familial Hypercholesterolemia/Familial hypobetalipoproteinemia; Heart disease
BAG3	BAG3-related dilated cardiomyopathy/Myofibrillar myopathy; Cardiomyopathy, neuromuscular condition
BMPR2	Heritable pulmonary arterial hypertension; Pulmonary arterial hypertension
CACNA1C	CACNA1C-related arrhythmia/cardiomyopathy/Timothy syndrome spectrum; Arrhythmia, cardiomyopathy, congenital heart disease
CALM1	Calmodulinopathy; Arrhythmia
CALM2	Calmodulinopathy; Arrhythmia
CALM3	Calmodulinopathy; Arrhythmia
CASQ2	Catecholaminergic polymorphic ventricular tachycardia type 2; Arrhythmia
CAV1	Heritable pulmonary arterial hypertension; Pulmonary arterial hypertension

CAV3	CAV3-related myopathy/Channelopathy; Arrhythmia, cardiomyopathy, neuromuscular condition
COL3A1	Vascular Ehlers-Danlos syndrome; Aortopathy
COL5A1	Classical Ehlers–Danlos syndrome; Aortopathy
COL5A2	Classical Ehlers–Danlos syndrome; Aortopathy
CRYAB	CRYAB-related myofibrillar myopathy/cardiomyopathy; Cardiomyopathy, neuromuscular condition
CSRP3	Hypertrophic cardiomyopathy; Cardiomyopathy
DES	Desmin-related myofibrillar myopathy/Cardiomyopathy; Arrhythmia, cardiomyopathy, neuromuscular condition
DMD	Duchenne/Becker muscular dystrophy/Dystrophinopathy; Cardiomyopathy, neuromuscular condition
DSC2	Arrhythmogenic right ventricular cardiomyopathy; Arrhythmia, cardiomyopathy
DSG2	Arrhythmogenic right ventricular cardiomyopathy; Arrhythmia, cardiomyopathy
DSP	Arrhythmogenic right ventricular cardiomyopathy; Arrhythmia, cardiomyopathy
EMD	Conduction disease/ Emery–Dreifuss muscular dystrophy type; Arrhythmia, cardiomyopathy, neuromuscular condition
FBN1	Marfan syndrome; Aortopathy
FHL1	FHL1-related myopathy/Emery–Dreifuss muscular dystrophy 6; Cardiomyopathy, neuromuscular condition
FLNC	Filamin C myofibrillar myopathy/Dilated/arrhythmogenic cardiomyopathy; Cardiomyopathy, neuromuscular condition
GLA	Fabry Disease; Cardiomyopathy, metabolic disorder
HCN4	HCN4-related arrhythmia/cardiomyopathy; Arrhythmia, cardiomyopathy
JUP	Arrhythmogenic cardiomyopathy; Arrhythmia, cardiomyopathy
KCNE1	Long QT syndrome type 5; Arrhythmia
KCNH2	Long QT syndrome type 2; Arrhythmia
KCNJ2	Andersen–Tawil syndrome; Arrhythmia
KCNQ1	Long QT syndrome type 1; Arrhythmia
LAMP2	Danon disease; Cardiomyopathy, metabolic disorder
LDLR	Familial Hypercholesterolemia; Heart disease
LDLRAP1	Familial Hypercholesterolemia; Heart disease
LMNA	Laminopathy; Arrhythmia, cardiomyopathy, neuromuscular condition
MYBPC3	Hypertrophic cardiomyopathy; Cardiomyopathy
MYH11	MYH11-related aortopathy; Aortopathy
MYH7	Myosin heavy chain 7-related myopathy; Cardiomyopathy, neuromuscular condition
MYL2	Hypertrophic cardiomyopathy; Cardiomyopathy
MYL3	Hypertrophic cardiomyopathy; Cardiomyopathy
MYLK	MYLK-related aortopathy; Aortopathy
PCSK9	Familial Hypercholesterolemia; Heart disease
PKP2	Arrhythmogenic right ventricular cardiomyopathy; Arrhythmia, cardiomyopathy
PLN	Arrhythmogenic cardiomyopathy/Dilated cardiomyopathy; Arrhythmia, cardiomyopathy
PRKAG2	Hypertrophic cardiomyopathy/Wolff-Parkinson-White syndrome; Arrhythmia, cardiomyopathy
PRKG1	PRKG1-related aortopathy; Aortopathy
RBM20	Dilated cardiomyopathy; Arrhythmia, cardiomyopathy
RYR2	Catecholaminergic polymorphic ventricular tachycardia type 1; Arrhythmia, cardiomyopathy
SCN5A	Long QT syndrome type 3/Brugada syndrome; Arrhythmia, cardiomyopathy
SGCD	Limb-girdle muscular dystrophy type 2F; Cardiomyopathy, neuromuscular condition
SMAD3	Loeys–Dietz syndrome type 3; Aortopathy
SMAD9	Heritable pulmonary arterial hypertension; Pulmonary arterial hypertension

TCAP	Limb-girdle muscular dystrophy type 2G/Cardiomyopathy; Cardiomyopathy, neuromuscular condition
TGFB2	Loeys–Dietz syndrome type 4, Aortopathy
TGFB3	Loeys–Dietz syndrome type 5, Aortopathy, Arrhythmia, cardiomyopathy
TGFBR1	Loeys–Dietz syndrome type 1, Aortopathy
TGFBR2	Loeys–Dietz syndrome type 2, Aortopathy
TMEM43	Arrhythmogenic right ventricular cardiomyopathy; Arrhythmia, cardiomyopathy
TNNC1	Hypertrophic cardiomyopathy; Cardiomyopathy
TNNI3	Hypertrophic/restrictive cardiomyopathy; Arrhythmia, cardiomyopathy
TNNT2	Hypertrophic/dilated cardiomyopathy; Arrhythmia, cardiomyopathy
TPM1	Hypertrophic cardiomyopathy; Cardiomyopathy
TRDN	Catecholaminergic polymorphic ventricular tachycardia; Arrhythmia
TTN	TTN-related titinopathy; Cardiomyopathy, neuromuscular condition
TTR	Hereditary transthyretin amyloidosis; Cardiomyopathy, neuromuscular condition
VCL	Dilated cardiomyopathy; Cardiomyopathy
Genes Related to Metabolic Conditions	
ABCD1	X-linked adrenoleukodystrophy; Nervous system and adrenal gland disorder
ATP7B	Wilson disease; Liver, eye and brain disease.
BTD	Biotinidase deficiency; Metabolic disorder
G6PD	Glucose-6-phosphate dehydrogenase deficiency; Red blood cell disorder
GAA	Pompe disease; Metabolic and neuromuscular disorder
HAMP	Hereditary hemochromatosis; Liver, heart, and pancreatic disease
HFE	Hereditary hemochromatosis; Liver, heart, and pancreatic disease
HJV	Hereditary hemochromatosis; Liver, heart, and pancreatic disease
HMBS	Acute intermittent porphyria; Metabolic disorder
HNF1A	Maturity-onset of diabetes of the young; Metabolic disorder
HNF1B	Renal cysts and diabetes syndrome; Metabolic disorder
OTC	Ornithine transcarbamylase deficiency; Metabolic disorder
SLC40A1	Hereditary hemochromatosis; Liver, heart, and pancreatic disease
TFR2	Hereditary hemochromatosis; Liver, heart, and pancreatic disease
Genes Related to Other Disease Phenotypes	
ACVRL1	Hereditary hemorrhagic telangiectasia type 2/Heritable pulmonary arterial hypertension (hPAH); Blood vessels disorder
CACNA1S	Malignant hyperthermia; Musculoskeletal disorder
ENG	Hereditary hemorrhagic telangiectasia type 1; Blood vessels disorder
F9	Hemophilia/Hereditary thrombophilia; Blood disorder
GCH1	Dystonia; Neurological movement disorder
GDF2	Hereditary hemorrhagic telangiectasia type 5; Blood vessels disorder
MEFV	Familial Mediterranean fever; Autoinflammatory disease
PROC	Hereditary thrombophilia; Blood disorder
PROS1	Hereditary thrombophilia; Blood disorder
RPE65	RPE65-related retinopathy; Eye disease
RYR1	Malignant hyperthermia; Musculoskeletal disorder
SERPINA1	Alpha 1 antitrypsin deficiency; Lung and liver disease
SERPINC1	Hereditary thrombophilia; Blood disorder

Genes whose carrier status will be reported are **bolded**.
Genes that are included in the American College of Medical Genetics and Genomics secondary finding list are underlined.