ATLAS Genetic Screening Program - Genes and Associated Conditions

The list of genes is based on the American College of Medical Genetics Secondary Findings Gene List (v3.1). There are 38 conditions and 78 genes currently being analyzed under this program:

Cancer Related Condition/Risks	Brief Description	Associated Gene(s)
Increased risk for breast, ovarian, and/or pancreatic cancer		
Hereditary breast and ovarian cancer (HBOC) syndrome	Increased chance of developing breast, ovarian, pancreatic, and/or prostate cancer.	BRCA1, BRCA2
Susceptibility to breast, ovarian, and/or pancreatic cancer		PALB2
Li-Fraumeni syndrome	Increased chance of developing breast, ovarian, pancreatic, prostate cancer and/or other types of cancers and symptoms.	TP53
Peutz-Jeghers syndrome		STK11
PTEN hamartoma tumor syndrome		PTEN
Increased risk of colon cancers		
Lynch syndrome	Increased chance of developing colon, intestinal, uterine, and ovarian cancers.	MLH1, MSH2, MSH6, PMS2
Juvenile polyposis syndrome (JPS)	Increased chance of developing polyps (non-cancerous growths) in the stomach/intestinal tract and/or cancer.	BMPR1A, SMAD4
Familial adenomatous polyposis (FAP)		APC
MUTYH-associated polyposis		MUTYH
Increased risk for other cancers		
Hereditary paraganglioma–pheochromocytoma syndrome (PGL-PCC)	Increased chance of developing tumors and/or cancers of the endocrine system.	SDHD, SDHAF2, SDHC, SDHB, MAX, TMEM127
Multiple endocrine neoplasia (MEN) syndromes and/or familial medullary thyroid cancer		MEN1, RET
Von Hippel–Lindau syndrome	Increased chance to develop non-cancerous tumors, as well as kidney cancer.	VHL
WT1-related Wilms tumor	Increased chance of developing a type of childhood kidney cancer.	WT1
Neurofibromatosis type 2	Conditions involving the nervous system and skin (neurocutaneous).	NF2
Tuberous sclerosis complex (TSC)		TSC1, TSC2
Retinoblastomas	Cancer that forms in the retina of the eye, usually during childhood.	RB1

Cardiovascular Related Conditions/Risks	Brief Description	Associated Gene(s)
Connective tissue related conditions		
Marfan syndrome	Multi-system connective tissue condition, including cardiovascular problems.	FBN1
Loeys - Dietz syndrome		TGFBR1, TGFBR2, SMAD3

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Ehlers - Danlos syndrome (vascular type)		COL3A1
Familial thoracic aortic aneurysm	Increased risks for problems with the aorta (large blood vessel).	ACTA2, MYH11
Increased risk of arrhythmias and/or cardiomyopathy		
Familial arrhythmias (CPVT, Brugada syndrome, Long QT syndrome)	Conditions related to an abnormal heartbeat.	KCNH2, SCN5A, RYR2, CASQ2, TRDN, KCNQ1
Familial cardiomyopathy (dilated cardiomyopathy, hypertrophic cardiomyopathy, arrhythmogenic cardiomyopathy)	Conditions related to problem with the structure of the heart and the way the heart pumps blood.	PKP2, DSC2, TMEM43, DSG2, DSP, FLNC, TTN (truncating variants only), LMNA, TNNT2, MYH7, MYBPC3, TNNI3, TPM1, MYL3, ACTC1, MYL2, PRKAG2, RBM20, TNNC1, DES, BAG3
Other cardiovascular related conditions		
Familial hypercholesterolemia	High levels of cholesterol.	LDLR, APOB, PCSK9

Other Conditions	Brief Description	Associated Gene(s)
Increased risk for other conditions/risks		
Hereditary transthyretin-related amyloidosis	Buildup of abnormal deposits of a protein called amyloid, that may cause health problems.	TTR
Biotinidase deficiency (BTD)	Problem with the body cannot recycle a vitamin called biotin.	BTD
Fabry disease	Buildup of a fatty molecule that affects the organs in the body.	GLA
Ornithine transcarbamylase (OTC) deficiency	High levels of ammonia that may cause health problems.	OTC
Pompe disease	Buildup of a complex sugar called glycogen, that may cause weakness in heart and skeletal muscles.	GAA
Hereditary hemochromatosis	High levels of iron which may lead to liver disease.	HFE (c.845G>A; p.C282Y homozygotes only)
Hereditary hemorrhagic telangiectasia (HHT)	Some blood vessels do not develop properly.	ACVRL1, ENG
Malignant hyperthermia	Severe and potentially life-threatening reaction to certain medications used during surgery (anesthetics and muscle relaxants).	RYR1, CACNA1S
Maturity-onset diabetes of the young	Early onset diabetes (usually under age 35 years).	HNF1A
RPE65-related retinopathy	Severe visual impairment.	RPE65
Wilson disease	Build-up of copper in organs that will cause health problems.	ATP7B

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