List of Genes Analyzed:

Phenotype/Associated Condition	Gene	Description
Genes related to cancer risks		
Familial Adenomatous Polyposis	APC	A syndrome marked 20-1000's of colon polyps and an increased risk for the development of colon cancer.
Familial Medullary Thyroid Cancer	RET	A syndrome associated with a high risk of developing medullary thyroid cancer.
Hereditary Breast and Ovarian Cancer	BRCA1, BRCA2, PALB2	An increased risk for the development of breast, ovarian, pancreatic and other cancers.
Hereditary Paraganglioma-Pheochromocytoma Syndrome	SDHD, SDHAF2, SDHC, SDHB, MAX, TMEM127	Syndromes characterized by the growth of paragangliomas and pheochromocytomas (paragangliomas in the adrenal glands) which are tumors that come from neuroendocrine tissues that control blood pressure.
Juvenile Polyposis Syndrome	BMRP1, SMAD4	A disorder characterized by the development of noncancerous (benign) polyps, specifically in the gastrointestinal tract, before the age of twenty.
Li-Fraumeni Syndrome	TP53	A rare disorder that greatly increases the risk of developing several types of cancer, particularly in children and young adults.
Lynch Syndrome	MLH1, MSH2, MSH6, PMS2	A syndrome characterized by an increased risk of many types of cancers, particularly colon and uterine cancer.
Multiple Endocrine Neoplasia Type 1	MEN1	A syndrome associated with tumors of the endocrine (hormone-producing) glands. Particularly: islet cells of pancreas, pituitary and parathyroid.

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MYH-Associated Polyposis (MAP); (autosomal recessive)	MUTYH	A disorder characterized by an increased risk for colon polyps and colon cancer.
Neurofibromatosis type 2	NF2	A disorder most commonly associated with noncancerous tumors in the nervous system. These growths usually develop along the nerve that carries information from the inner ear to the brain and can lead to hearing loss.
Peutz-Jeghers Syndrome	STK11	A syndrome that causes the development of noncancerous growths, called hamartomatous polyps, in the gastrointestinal tract (stomach and intestines).
PTEN Hamartoma Tumor Syndrome	PTEN	A disorder that causes a high risk for the development of benign and malignant tumors of the thyroid, breast, colon, and uterus.
Retinoblastoma	RB1	An eye cancer that begins in the back of the eye (retina) in children.
Tuberous Sclerosis Complex	TSC1, TSC2	A multisystem disorder characterized by the growth of noncancerous (benign) tumors in many parts of the body, most commonly skin, brain, and kidneys.
Von Hippel Lindau Syndrome	VHL	A syndrome characterized by the formation of tumors and cysts in many parts of the body, including the brain, spinal cord, and retina.
WT1- related Wilms tumor	WT1	Increases risk for a specific type of kidney tumor called a Wilms tumor. Typically occurs in childhood.
Genes related to cardiac/cardiovascular risk conditions		
Aortopathies: Marfan Syndrome, Loeys-Dietz Syndromes, and Familial Thoracic Aortic Aneurysms and Dissections	FBN1, TGFBR1, TGFBR2, SMAD3, ACTA2, MYH11	Disorders characterized by the enlargement of the aorta which increases risk of aneurysm/ dissection and can include other health issues such as lens dislocation of the eye (Marfan syndrome) or aneurysms/ dissections of other arteries (Loeys Dietz syndrome).

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Arrhythmogenic right ventricular cardiomyopathy	PKP2, DSP, DSC2, TMEM43, DSG2	A disease of the heart muscle that can lead to an abnormal heart rhythm (arrhythmia), heart failure and other cardiac complications such as sudden death.
Catecholaminergic polymorphic ventricular tachycardia	RYR2, CASQ2, TRDN	A condition characterized by abnormal heart rhythm (arrhythmia) where a trigger such as physical activity can lead to an abnormally fast and irregular heartbeat.
Dilated cardiomyopathy	TNNT2, LMNA, FLNC, TTN, BAG3, DES, RMB20, TNNC1	A disease of the heart muscle characterized by an enlargement (dilation) of the heart that can lead to an abnormal heart rhythm (arrhythmia), heart failure and other cardiac complications.
Ehlers-Danlos Syndrome-vascular type	COL3A1	The most severe type of Ehlers-Danlos Syndrome that affects the connective tissues that support the skin, bones, blood vessels and joints. This form of Ehlers Danlos syndrome has an increased risk for rupturing of arteries, muscles and internal organs. Other forms of Ehlers Danlos syndrome not evaluated through this gene.
Familial hypercholesterolemia	APOB, LDLR, PCSK9,	Increased risk for high cholesterol.
Hypertrophic cardiomyopathy	MYBPC3, MYH7, TNNI3, TPM1, MYL3, ACTC1, PRKAG2, MYL2	A disease of the heart muscle characterized by a thickening of heart muscle that can lead to an abnormal heart rhythm (arrhythmia), heart failure and other cardiac complications.

Phenotype/Associated Condition	Gene	Description
Long QT Syndromes Types 1, 2, and 3, Brugada Syndrome	KCNQ1, KCNH2, SCN5A	Conditions characterized by abnormal heart rhythm (arrhythmia) that can cause fainting, seizures, and other cardiac complications such as sudden death.
Genes related to inborn errors of metabolism		
Biotinidase deficiency (autosomal recessive)	BTD	A disorder where the body cannot normally process the vitamin biotin. The severe form leads to seizures, developmental delays and multiple health issues and is typically recognized in the first months of life.
Fabry disease (X linked)	GLA	A condition that can cause a variety of health complications including kidney disease/ kidney failure; heart disease and heart rhythm issues (arrhythmia), strokes, digestive issues, neuropathy, and temperature sensitivity. Both men and women can be affected with this condition.
Ornithine transcarbamylase deficiency (X-linked)	OTC	A disorder that causes ammonia to accumulate in the blood. This can cause developmental delay, intellectual impairment, altered mental status, and liver damage. This condition can occur at any age, with the late-onset type occurring in both males and females.
Pompe disease (autosomal recessive)	GAA	A progressive muscle disease that causes significant muscle weakness leading to difficulties with movement and breathing issues. Different forms exist with differing ages of onset of symptoms and rates of progression of disease.
Genes related to miscellaneous genetic conditions		
Hereditary hemochromatosis	HFE	A condition that leads to excess iron in the body's tissues and can cause multiple health complications including liver disease, heart problems and diabetes.

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Hereditary hemorrhagic telangiectasia	ACVRL1, ENG	A disorder where some blood vessels have not developed properly leading to abnormal connections between arteries and veins. This condition leads to abnormal bleeding and often affects the brain, lungs, and liver with some complications being life threatening.
Malignant hyperthermia susceptibility	RYR1, CACNA1S	A rare life-threatening condition that is usually triggered by exposure to certain drugs used for general anesthesia
Maturity-onset diabetes of the young (MODY)	HNF1A	An inherited form of diabetes that presents similarly to adult type diabetes but at younger ages than typically seen with adult type diabetes.
RPE65- related retinopathy (autosomal recessive)	RPE65	Disease of the retina of the eye that starts at birth/ early childhood leading to loss of vision.
Wilson Disease (autosomal recessive)	ATP7B	A disorder that causes too much copper to accumulate in the organs, particularly the liver, brain, and eyes causing neurological issues such as uncontrolled movements and problems with speech, swallowing, and coordination.
Hereditary TTR amyloidosis	TTR	A disorder that is caused by a buildup of the protein 'amyloid' that leads to multiple health issues including heart disease and neuropathy.