Veritas
The Genome Company

myGenome Standard/Premium Gene-Disease Reference List for Providers

We aim to inform and educate our customers and partners with clear, concise and convenient resources. The Clinical Section of the myGenome test is designed to examine genes associated with a set of actionable genetic disorders as well as genes associated with common carrier conditions.

The list below shows all interpreted genes and their associated conditions/diseases. Both myGenome test options (Standard and Premium) include interpretation of 59 genes deemed medically actionable by the American College of Medical Genetics and Genomics and 24 genes associated with recessive carrier conditions.

If you have any questions, please contact us at 1 (888) 507-6619 or email clinical@veritasgenetics.com.

Clinically Actionable Disease Group*	Gene	ACMG (Y/N)	Included in myGenome Standard	Mode of Inheritance	Clinically Actionable Disease by Gene	Additional Disease Associations (Sources: OMIM, GeneReviews, and/or Genetics Home Reference)
Cancer Susceptibility						
Hereditary Breast Cancer (45 genes) • Hereditary Breast and Ovarian Cancer syndrome • PTEN Hamartoma tumor syndrome • Li-Fraumeni syndrome • Reproductive (gynecological, prostate)	*BRCA1*	Yes	Yes	AD	Hereditary Breast and Ovarian Cancer syndrome (female and male breast cancer, ovarian/fallopian tube, prostate, pancreatic)	N/A
	BRCA2	Yes	Yes	AD, AR	Hereditary Breast and Ovarian Cancer syndrome (female and male breast cancer, ovarian/fallopian tube, prostate, pancreatic, melanoma) (AD)	Fanconi anemia type D1 (AR); Glioblastoma; medulloblastoma; Wilms tumor
	PTEN	Yes	Yes	AD	PTEN Hamartoma tumor syndrome; Cowden syndrome; Banayan-Riley-Ruvalcaba syndrome	Lhermitte-Duclos syndrome; macrocephaly/autism syndrome; VATER association with macrocephaly and ventriculomegaly; Glioma susceptibility; meningioma
	TP53	Yes	Yes	AD	Li-Fraumeni syndrome	Bone marrow failure syndrome; Adrenocortical carcinoma (pediatric); Basal cell carcinoma; Choroid plexus papilloma; Colorectal cancer; glioma susceptibility; osteosarcoma
	ABRAXAS1	No	No	AD	Possible association with breast cancer	Possible association with other cancer types (ovarian, prostate, lymphoma, skin, endometrial, colon, lip, carcinosarcoma)
	AKT1	No	No	AD	Cowden syndrome	Possible association with lung cancer; susceptibility to Schizophrenia
	AR	No	No	XL	Susceptibility to prostate cancer	Androgen insensitivity; XL hypospadias; Spinal and bulbar muscular atrophy of Kennedy; Possibly polycystic ovary syndrome
	ATM	No	No	AD, AR	Susceptibility to breast cancer and other cancer (pancreatic, prostate) types (AD)	Ataxia telangiectasia (AR)
	BARD1	No	No	AD	Susceptibility to breast cancer	N/A
	BRIP1	No	No	AD, AR	Susceptibility to breast cancer and Possibly other cancer types (ovarian, pancreatic, prostate) (AD)	Fanconi anemia type J (AR)
	CD82	No	No	AD	Susceptibility to prostate cancer	N/A
	CHEK2	No	No	AD	Susceptibility to breast, prostate, and colorectal cancers and Possibly other cancer types	N/A
	CYP19A1	No	No	Unknown, AD, AR	Possible association with breast cancer (Unknown)	Aromatase deficiency (AR); Aromatase excess syndrome (AD)
	DICER1	No	No	AD	DICER1-related disorders (includes susceptbility to pleuropulmonary blastoma (PPB); ovarian sex cord-stromal tumors (Sertoli-Leydig cell tumor, juvenile granulosa cell tumor, and gynandroblastoma); cystic nephroma; and thyroid gland neoplasia (multinodular goiter, adenomas, or differentiated thyroid cancer; ciliary body medulloepithelioma; botryoid-type embryonal rhabdomyosarcoma of the cervix or other sites; nasal chondromesenchymal hamartoma; renal sarcoma; pituitary blastoma; pineoblastoma; and Possibly other cancer types) (also known as DICER1-Pleuropulmonary Blastoma Familial Tumor Predisposition Syndrome; DICER1 Syndrome)	N/A
	ESR1	No	No	Unknown, AD, AR	Possible association with breast cancer (Unknown)	Susceptibility to atherosclerosis, migraine, myocardial infarction (all AD); estrogen resistance (AR)
	GEN1	No	No	AR	Susceptibility to breast cancer and Possibly other cancer types (prostate)	N/A
	HIP1	No	No	AD	Associated with prostate cancer progression and metastasis	A chromosomal aberration involving HIP1 is found in a form of chronic myelomonocytic leukemia (CMML). Translocation t(5;7)(q33;q11.2) with PDGFRB (PubMed:9616134). The chimeric HIP1-PDGFRB transcript results from an in-frame fusion of the two genes (PubMed:9616134)
	HMMR	No	No	AD	Susceptibility to breast cancer	N/A
	HOXB13	No	No	AD	Hereditary prostate cancer	N/A

	KLLN	No	No	AD	Cowden syndrome (germline KLLN promoter methylation epimutation is disease causing mechanism for Cowden and Cowden-like syndromes - 30% of cases)	N/A
	LSP1	No	No	Unknown	Possible association with breast cancer	N/A
	MAP3K1	No	No	Unknown, AD	Possible association with breast cancer (Unknown)	46XY sex reversal/Swyer syndrome (AD)
	MRE11	No	No	AD, AR	Susceptibility to breast cancer and Possibly other cancer types (AD); Ataxia-telangiectasia-like disorder (AR)	N/A
	MSR1	No	No	AD	Barrett esophagus/esophageal adenocarcinoma; hereditary prostate cancer (AD)	N/A
	MXI1	No	No	Unknown	Possible association with prostate cancer	N/A
	NBN	No	No	AD, AR	Susceptibility to breast cancer and Possibly other cancer types (prostate, ovarian) (AD)	Nijmegen breakage syndrome (AR)
	NQO2	No	No	AD	Possible association with breast cancer	N/A
	OPCML	No	No	AD	Susceptibility to ovarian cancer	N/A
	PALB2	No	No	AD, AR	Susceptibility to breast and pancreatic cancers and Possibly other cancer types (AD)	Fanconi anemia type N (AR)
	PHB	No	No	AD	Susceptibility to breast cancer	N/A
	PIK3CA	No	No	AD	Cowden syndrome	N/A
	PPM1D	No	No	Unknown, AD	Possible association with breast cancer (Unknown)	Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold (AD)
	RAD50	No	No	AD, AR, Unknown	Susceptibility to breast cancer and Possibly other cancers (ovarian) (AD)	Possibly Nijmegen breakage syndrome-like disorder (AR); Possibly polycystic ovary syndrome (Unknown)
	RAD51	No	No	AD	Susceptibility to breast cancer	Possibly Fanconi anemia type R; Mirror movements 2
	RAD51C	No	No	AD, AR	Susceptibility to ovarian cancer and Possibly breast cancer (AD)	Fanconi anemia type O (AR)
	RAD51D	No	No	AD	Susceptibility to ovarian cancer and Possibly breast cancer (AD)	N/A
	RAD54L	No	No	AD	Susceptibility to breast cancer	N/A
	RBBP8	No	No	Unknown, AR	Possible association with breast cancer (Unknown)	Jawad syndrome; Seckel syndrome 2 (both AR)
	RECQL	No	No	Unknown	Possible association with breast cancer	N/A
	RECQL4	No	No	Unknown, AR	Possible association with breast cancer (Unknown)	Baller-Gerold syndrome; RAPADILINO syndrome; Rothmund-Thomson syndrome (all AR)
	RINT1	No	No	Unknown	Possible association with breast and colorectal cancers	N/A
	SEC23B	No	No	AD, AR	Possibly Cowden syndrome (AD)	Congenital dyserythropoietic anemia, type II (CDAII) (AR)
	SMARCA4	No	No	AD	Susceptibility to small cell carcinoma of the ovary, hypercalcemic type (SCCOHT)	Coffin-Siris syndrome; Rhabdoid tumor predisposition syndrome (RTPS)
	TOX3	No	No	Unknown	Possible association with breast cancer	Possibly polycystic ovary syndrome and restless legs syndrome
	XRCC2	No	No	AD, AR	Possible association with breast cancer and other cancer types (gynecological, gastrointestinal) (AD)	Fanconi anemia type U (AR)
Hereditary Gastrointestinal Polyposis and Cancer (37 genes) • Lynch syndrome • Familial adenomatous polyposis • MUTYH-Associated Polyposis • Juvenile Polyposis • Peutz-Jeghers syndrome • Hereditary Pancreatitis	*APC*	Yes	Yes	AD	Familial Adenomatous Polyposis (FAP); Attenuated FAP; Gardner syndrome; Turcot syndrome	N/A
	BMPR1A	Yes	Yes	AD	Juvenile Polyposis syndrome	Hereditary Mixed Polyposis syndrome
	MLH1	Yes	Yes	AD, AR	Hereditary Non-Polyposis Colorectal syndrome (HNPCC) (AD; also known as Lynch syndrome and/or Muir-Torre syndrome)	Constitutional Mismatch Repair Deficiency (CMMR) syndrome (AR)
	MSH2	Yes	Yes	AD, AR	Hereditary Non-Polyposis Colorectal syndrome (HNPCC) (AD; also known as Lynch syndrome and/or Muir-Torre syndrome)	Constitutional Mismatch Repair Deficiency (CMMR) syndrome (AR)
	MSH6	Yes	Yes	AD, AR	Hereditary Non-Polyposis Colorectal syndrome (HNPCC) (AD; also known as Lynch syndrome and/or Muir-Torre syndrome)	Constitutional Mismatch Repair Deficiency (CMMR) syndrome (AR)
	MUTYH	Yes	Yes	AR, AD	MUTYH-Associated Polyposis (AR; also known as MYH-Associated Polyposis); Adenomas, multiple colorectal, FAP type 2; Autosomal Recessive Colorectal adenomatous polyposis with pilomatricomas	Susceptibility to colorectal cancer (AD)
	PMS2	Yes	Yes	AD, AR	Hereditary Non-Polyposis Colorectal syndrome (HNPCC) (AD; also known as Lynch syndrome and/or Muir-Torre syndrome)	Constitutional Mismatch Repair Deficiency (CMMR) syndrome (AR)
	SMAD4	Yes	Yes	AD	Juvenile Polyposis syndrome	Hereditary Hemorrhagic Telangiectasia; Myrhe syndrome
	STK11	Yes	Yes	AD	Peutz-Jeghers syndrome	N/A
	AURKA	No	No	AD	Susceptibility to colorectal cancer	N/A
	AXIN2	No	No	AD	Hereditary Oligodontia-colorectal cancer syndrome	Oligodontia-colorectal cancer syndrome
	BUB1B	No	No	AD, AR	Susceptibility to colorectal cancer (AD)	Mosaic variegated aneuploidy syndrome 1 (AR); Premature chromatid separation trait (AD)
	CCND1	No	No	AD	Susceptibility to colorectal cancer	Possible modifier of von-Hippel Lindau syndrome
	CDH1	No	No	AD	Hereditary Diffuse Gastric Cancer syndrome	N/A

	CTNNA1	No	No	AD	Possibly Hereditary Diffuse Gastric Cancer syndrome	Macular dystrophy, patterned
	CTNNB1	No	No	Unknown, AD	Possible association for colorectal cancer (Unknown)	Exudative vitreoretinopathy (AD); Mental retardation (AD)
	CTRC	No	No	AD	Susceptibility to chronic pancreatitis	N/A
	ENG	No	No	AD	Possibly Juvenile Polyposis syndrome	Hereditary Hemorrhagic Telangiectasia (HHT); Hereditary Pulmonary Arterial Hypertension (PAH)
	EPCAM	No	No	AD, AR	Hereditary Non-Polyposis Colorectal syndrome (HNPCC) (AD; also known as Lynch syndrome and/or Muir-Torre syndrome)	Constitutional Mismatch Repair Deficiency (CMMR) syndrome (AR); Diarrhea 5, with tufting enteropathy, congenital (AR)
	FAN1	No	No	AD, AR	Possibly Hereditary Non-Polyposis Colorectal Cancer syndrome/Lynch syndrome (AD)	Interstitial nephritis, karyomegalic (AR)
	GALNT12	No	No	Unknown	Susceptibility for colorectal cancer	N/A
	GREM1	No	No	AD	Hereditary mixed polyposis syndrome	N/A
	KIT	No	No	AD	Gastrointestinal Stromal Tumors (GISTs)	Partial albinism (Piebaldism); Acute Myeloid Leukemia; Cutaneous Mastocytosis
	MLH3	No	No	AD	Hereditary Non-Polyposis Colorectal syndrome (HNPCC) also known as Lynch syndrome	N/A
	MSH3	No	No	AR	MSH3-Associated Polypisis (also known as familial adenomatous polyposis 4)	N/A
	NTHL1	No	No	AR	NTHL1-Associated Polyposis (also known as Familial adenomatous polyposis 3)	N/A
	ODC1	No	No	AD	Possibly reduced risk for colorectal adenoma recurrence	N/A
	PALLD	No	No	Unknown	Susceptibility to pancreatic cancer	N/A
	PDGFRA	No	No	Unknown	Possible association for Gastrointestinal Stromal Tumor (GIST)	N/A
	PLA2G2A	No	No	AD	Susceptibility to colorectal cancer	N/A
	PMS1	No	No	Unknown	Possibly Hereditary Non-Polyposis Colorectal Cancer syndrome (also known as Lynch syndrome)	N/A
	POLD1	No	No	AD	Polymerase proofreading-associated polyposis (PPAP) (AD)	Mandibular hypoplasia, Deafness, Progeroid features, and Lipodystrophy (MDPL) syndrome
	POLE	No	No	AD, AR	Polymerase proofreading-associated polyposis (PPAP) (AD)	Facial dysmorphism, immunodeficiency, livedo, and short stature (FILS) syndrome (AR)
	PRSS1	No	No	AD, AR	Hereditary pancreatitis (AD)	Trypsinogen deficiency (AR)
	RPS20	No	No	AD	Possibly Hereditary Non-Polyposis Colorectal Cancer syndrome (also known as Lynch syndrome)	N/A
	SPINK1	No	No	AD, AR	Hereditary pancreatitis (AD)	Tropical calcific pancreatitis (AD/AR); Susceptibility to Fibrocalculous pancreatic diabetes (AD/AR)
	TLR2	No	No	AD, Unknown	Susceptibility to colorectal cancer (AD)	Susceptibility to Leprosy (AD); Susceptibility to Mycobacterium tuberculosis (AD, Unknown)
Neurocutaneous and Ocular (6 genes) • Neurofibromatosis types 1 & 2 • Tuberous Sclerosis Complex types 1 & 2 • Retinoblastoma	*NF2*	Yes	Yes	AD	Neurofibromatosis type 2	N/A
	RB1	Yes	Yes	AD	Retinoblastoma	N/A
	TSC1	Yes	Yes	AD, Unknown	Tuberous Sclerosis Complex type 1 (AD)	Lymphangioleiomyomatosis (Unknown)
	TSC2	Yes	Yes	AD	Tuberous Sclerosis Complex type 2	N/A
	NF1	No	No	AD	Neurofibromatosis type 1	Leukemia, juvenile myelomonocytic; Watson syndrome
	SMARCB1	No	No	AD	Susceptibility to Schwannomatosis	Rhabdoid tumor predisposition syndrome; Coffin-Siris syndrome
Neuro/endocrine and Renal (31 genes) • Familial Medullary Thyroid Cancer • Hereditary Paraganglioma-Pheochromocytoma syndrome • Multiple Endocrine Neoplasia types 1 & 2 • von Hippel-Lindau syndrome • WT1-related Wilm's tumor	*MEN1*	Yes	Yes	AD, Unknown	Multiple Endocrine Neoplasia type 1 (AD)	Lung carcinoid (Unknown)
	RET	Yes	Yes	AD	Familial Medullary Thyroid Cancer (FMTC)	Multiple Endocrine Neoplasia Type IIA and IIB; Pheochromocytoma; Central hypoventilation syndrome, congenital; Susceptibility to and protection against Hirschsprung disease
	SDHAF2	Yes	Yes	AD	Hereditary Paraganglioma-Pheochromocytoma Syndrome	N/A
	SDHB	Yes	Yes	AD	Hereditary Paraganglioma-Pheochromocytoma Syndrome	Cowden syndrome; Gastrointestinal stromal tumor (GIST)
	SDHC	Yes	Yes	AD	Hereditary Paraganglioma-Pheochromocytoma Syndrome	Cowden syndrome; Gastrointestinal stromal tumor (GIST)
	SDHD	Yes	Yes	AD, AR	Hereditary Paraganglioma-Pheochromocytoma Syndrome (AD)	Gastrointestinal stromal tumor (GIST; AD); Cowden syndrome (AD); mitochondrial complex II deficiency (AR)
	VHL	Yes	Yes	AD, AR	von Hippel-Lindau Syndrome (AD)	Familial erythrocytosis (AR)
	WT1	Yes	Yes	AD, Unknown, AR	WT1-related Wilm's tumor (AD)	WAGR syndrome (Wilms tumor, aniridia, genital anomalies, retardation; AD); Denys-Drash syndrome (AD); Frasier syndrome (AD); Congenital nephrotic syndrome (AD); Meacham syndrome (AR, Unknown)
	AIP	No	No	AD	Familial isolated pituitary adenomas (FIPA; also known as Pituitary Adenoma Predisposition (PAP) syndrome)	N/A
	BAP1	No	No	AD	BAP1 tumor predisposition syndrome	N/A
	CDC73	No	No	AD	Hyperparathyroidism-Jaw Tumor syndrome (HPT-JT)	Familial isolated hyperparathyroidism (FIH); Parathyroid adenoma with cystic changes; Parathyroid carcinoma
	CDKN1B	No	No	AD	Multiple Endocrine Neoplasia type 4	N/A
	CDKN1C	No	No	AD	Beckwith-Wiedeman syndrome	IMAGE syndrome
	DIS3L2	No	No	AR	Perlman syndrome	N/A

	EGLN1	No	No	AD	Possible association with paraganglioma and/or pheochromocytoma	Familial erythrocytosis, type 3; Hemoglobin, high altitude adaptation
	EGLN2	No	No	AD	Possible association with paraganglioma-pheochromocytoma and polycythemia	N/A
	EPAS1	No	No	AD	Possible association with paraganglioma-pheochromocytoma and polycythemia/somatostatinoma	Familial erythrocytosis, type 4
	FH	No	No	AD, AR	Hereditary Leiomyomatosis and Renal Cell Cancer (HLRCC; AD)	Fumarase deficiency (AR)
	FLCN	No	No	AD	Birtt-Hogg-Dube syndrome	Pneumothorax, primary spontaneous
	H19	No	No	AD	Suscpetibiity to Wilm's tumor	Beckwith-Wiedeman syndrome; Russell Silver syndrome
	HABP2	No	No	AD	Possible association with non-medullary thyroid cancer	Susceptibility to venous thromboembolism
	KIF1B	No	No	AD	Susceptibility to pheochromocytoma	Susceptibility to neuroblastoma; Possible association with Charcot-Marie-Tooth disease, type 2A1
	MAX	No	No	AD	Hereditary Paraganglioma-Pheochromocytoma Syndrome	N/A
	MET	No	No	AD, AR	Familial papillary renal cell carcinoma (AD)	Autosomal recessive (AR) deafness; Susceptibility to osteofibrous dysplasia (AD)
	MITF	No	No	AD, AR	Susceptibility to cutaneous malignant melanoma and Possibly renal cell carcinoma (AD)	COMMAD syndrome (AR); Tietz albinism-deafness syndrome (AD); Waardenburg syndrome, type 2A (AD); Waardenburg syndrome/ocular albinism, digenic (AD)
	PRKAR1A	No	No	AD	Carney complex, type 1	Acrodysostosis 1, with or without hormone resistance; Myxoma, intracardiac; Pigmented nodular adrenocortical disease, primary, 1
	SDHA	No	No	AD, AR, Mito, Unknown	Hereditary Paraganglioma-Pheochromocytoma Syndrome (AD); Dilated cardiomyopathy (Unknown)	Gastrointestinal stromal tumor (GIST); Mitochondrial respiratory chain complex II deficiency (AR); Leigh syndrome (AR, mitochondrial)
	SRGAP1	No	No	AD	Susceptibility to non-medullary thyroid cancer	N/A
	TMEM127	No	No	AD	Hereditary Paraganglioma-Pheochromocytoma Syndrome	N/A
	TRIM37	No	No	AR	Mulibrey nanism (susceptibility to Wilm's tumor)	N/A
	TSHR	No	No	Unknown, AD, AR	Susceptibility to thyroid carcinoma (Unknown)	Hyperthyroidism (includes familial gestational; nonautoimmune (AD); congenital, nongoitrous (AR))
Other cancers (115 genes) • Skin • Hemotological • Solid tumors	*ACD*	No	No	AD, AR	Dyskeratosis congenita (AD, AR)	N/A
	ALK	No	No	AD	Susceptibility to neuroblastoma	N/A
	ARID5B	No	No	Unknown	Possible association with acute lymphoblastic leukemia (ALL)	N/A
	ATR	No	No	AD, AR	Familial cutaneous telangiectasia and cancer syndrome (FCTCS) (AD)	Seckel syndrome (AR)
	CASP8	No	No	AD, AR	Protection against breast cancer (AD) and lung cancer (AR)	Autoimmune lymphoproliferative syndrome, type IIB (ALPS; AR)
	CBX8	No	No	Unknown	Possible association with cancer susceptibility	N/A
	CDK4	No	No	AD	Susceptibility to cutaneous malignant melanoma	N/A
	CDKN2A	No	No	AD	Familial Atypical Mole-Malignant Melanoma syndrome (FAMMM; also known as autosomal dominant melanoma-pancreatic cancer syndrome)	Melanoma and neural system tumor syndrome (melanoma -NST)
	CEBPA	No	No	AD	Susceptiblity to Familial acute myeloid leukemia (AML)	N/A
	CEBPE	No	No	AR	Susceptibility to neutrophil-specific granule deficiency	N/A
	CEP57	No	No	AR	Mosaic Variegated Aneuploidy (MVA) syndrome 2	N/A
	COL7A1	No	No	AD, AR	Epidermolysis bullosa (includes dystrophica (AR, AD); inversa (AR); Bart type (AD); pruriginosa (AD, AR); pretibial (AR, AD)) and susceptibility to squamous cell carcinoma	Isolated toenail dystrophy (AD); Transient bullous of the newborn (AR, AD)
	CREBBP	No	No	AD	Rubenstein-Taybi syndrome	this gene is commonly deleted in the recurrent 16p13.3 microdeletion syndrome
	CTC1	No	No	AR	Dyskeratosis congenita	Cerebroretinal microangiopathy with calcifications and cysts
	CTLA4	No	No	AD, Unknown	Autoimmune lymphoproliferative syndrome, type V (ALPS; AD)	Susceptibility to Celiac disease (Unknown); Susceptibility to Diabetes mellitus, insulin-dependent (Unknown); Susceptibility to Hashimoto thyroiditis (AD); Susceptibility to systemic lupus erythematosus (AD)
	CYLD	No	No	AD	Brooke-Spiegler syndrome (BSS); Familial Cylindromatosis; Multiple familial Trichoepithelioma	N/A
	DDB2	No	No	AR	Xeroderma Pigmentosum, group E, DDB-negative subtype	N/A
	DDR2	No	No	Unknown, AD, AR	Possible susceptbility to lung cancer (Unknown)	Spondylometaepiphyseal dysplasia, short limb-hand type (AR); Warburg-Cinotti syndrome (AD)
	DDX41	No	No	AD	Susceptibility to Familial Myeloproliferative/ lymphoproliferative neoplasms (multiple types)	N/A
	DKC1	No	No	XLR	Dyskeratosis congenita	N/A
	DOCK8	No	No	AR	Hyper-IgE recurrent infection syndrome	N/A
	EGFR	No	No	AD, AR	Susceptibility to non-small cell lung cancer (NSCLC; AD)	Inflammatory skin and bowel disease, neonatal, 2 (AR); Response to tyrosine kinase inhibitor in NSCLC (AD)

ELANE	No	No	AD	Severe congenital neutropenia, type 1	Cyclic neutropenia, cyclic
EP300	No	No	AD	Rubinstein-Taybi syndrome	Menke-Hennekam syndrome 2; Possibly Cornelia de Lange syndrome
ERBB2	No	No	AD	Possible suceptibility to various cancers	N/A
ERCC1	No	No	AR	Cerebrooculofacioskeletal syndrome 4	N/A
ERCC2	No	No	AR	Xeroderma pigmentosum, group D	Possibly Cerebrooculofacioskeletal syndrome 2; Trichothiodystrophy 1, photosensitive
ERCC3	No	No	AR	Xeroderma pigmentosum, group B	Trichothiodystrophy 2, photosensitive
ERCC4	No	No	AR	Xeroderma pigmentosum, type F +/- Cockayne syndrome	Possibly XFE progeroid syndrome; Fanconi anemia, complementation group Q
ERCC5	No	No	AR	Xeroderma pigmentosum, group G +/- Cockayne syndrome	Cerebrooculofacioskeletal syndrome 3
ETV6	No	No	AD	Thrombocytopenia type 5 (includes susceptibility to hematological cancers and Possibly solid tumors)	N/A
EXO1	No	No	Unknown	Possible susceptbility to colorectal cancer	N/A
EXT1	No	No	AD, AR	Hereditary Multiple Osteochondromas type 1 (AD)	Chondrosarcoma (AR); multiple exostoses type 1 (AD)
EXT2	No	No	AD, AR	Hereditary Multiple Osteochondromas type 2 (AD)	Possibly seizures, scoliosis, and macrocephaly syndrome (AR); multiple exostoses type 2 (AD)
EZH2	No	No	AD	Weaver syndrome	N/A
FANCA	No	Yes	AR	Fanconi anemia, complementation group A	N/A
FANCB	No	No	XLR	Fanconi anemia, complementation group B	N/A
FANCC	No	Yes	AR	Fanconi anemia, complementation group C	N/A
FANCD2	No	No	AR	Fanconi anemia, complementation group D2	N/A
FANCE	No	No	AR	Fanconi anemia, complementation group E	N/A
FANCF	No	No	AR	Fanconi anemia, complementation group F	N/A
FANCG	No	Yes	AR	Fanconi anemia, complementation group G	N/A
FANCI	No	No	AR	Fanconi anemia, complementation group I	N/A
FANCL	No	No	AR	Fanconi anemia, complementation group L	N/A
FANCM	No	No	AR	Fanconi anemia, complementation group M	Possibly premature ovarian failure; Spermatogenic failure
FAS	No	No	AD	Autoimmune lymphoproliferative syndrome, type IA	N/A
FAT1	No	No	Unknown, AD, AR	Possible association with head and neck squamous cell carcinoma (Unknown)	Possibly facioscapulohumeral muscular dystrophy like phenotype (AD, Unknown); Possibly steroid-resistant nephrotic syndrome (AD, AR, Unknown)
FGFR2	No	No	AD, Unknown	Possible association for various cancer types (Unknown)	Antley-Bixler syndrome (AD); Apert syndrome (AD); Beare-Stevenson cutis gyrata syndrome (AD); bent bone dysplasia syndome (AD); craniosynostosis (nonspecific; Unknown); Crouzon syndrome (AD); Jackson-Weiss syndrome (AD); LADD syndrome (AD); Pfeiffer syndrome (AD);; Saethre-Chotzen syndrome (AD);; Scaphocephaly and Axenfeld-Rieger anomaly (Unknown); Scaphocephaly, maxillary retrusion, and mental retardation (Unknown)
G6PC3	No	No	AR	Severe congenital neutropenia, type 1	Dursun syndrome
GATA2	No	No	Unknown, AD	Susceptibility to Acute Myeloid Leukemia (AML; AD) and Myelodysplastic syndrome (Unknown)	Emberger syndrome (AD); Immunodeficiency 21 (AD)
GATA1	No	No	XLR	GATA1-related X-linked Thrombocytopenia (includes Anemia with/without neutropenia and/or platelet abnormalities; Thrombocytopenia with beta-thalassemia; Thrombocytopenia with or without dyserythropoietic anemia)	Diamond-Blackfan Anemia
GPC3	No	No	XLR	Simpson-Golabi-Behmel syndrome, type 1	N/A
HAX1	No	No	AR	Severe congenital neutropenia, type 1	N/A
HMBS	No	No	AD	Acute intermittent porphyria	N/A
HNF1A	No	No	Unknown, AD, AR	Susceptibility to renal cell carcinoma (Unknown)	diabetes mellitus (both insulin (AR) and non-insulin dependent (AD)); MODY type III (AD)
IKZF1	No	No	AD	Common Variable Immunodeficiency (CVID)	N/A
ITK	No	No	AR	Lymphoproliferative syndrome 1	N/A
JAK2	No	No	AD	Thrombocythemia 3	N/A
MC1R	No	No	Unknown, AR	Susceptibility to cutaneous malignant melanoma (Unknown)	Analgesia from kappa-opioid receptor agonist, female-specific; Skin/hair/eye pigmentation 2, blond hair/fair skin (AR); Skin/hair/eye pigmentation 2, red hair/fair skin (AR); Albinism, oculocutaneous, type II, modifier of (AR); UV-induced skin damage} (AR)
MGMT	No	No	Unknown	Possible association with colorectal cancer and Possibly other cancer types (promoter methylation reported in several cancer types, including individuals with Lynch syndrome)	N/A
MPL	No	No	AD, AR	Thrombocythemia 2 (AD)	Thrombocytopenia, congenital amegakaryocytic (AR)
MTAP	No	No	AD	Diaphyseal medullary stenosis with malignant fibrous histiocytoma	N/A
NHP2	No	No	AR	Dyskeratosis congenita	N/A
NOP10	No	No	AR	Dyskeratosis congenita	N/A
NSD1	No	No	AD	Susceptibility to acute myeloid leukemia (AML)	Sotos syndrome
PARN	No	No	AR, AD	Dyskeratosis congenita (AR)	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4 (AD)

PAX5	No	No	Unknown	Susceptibility to acute lymphoblastic leukemia (ALL)	N/A
PHOX2B	No	No	Unknown, AD	Susceptibility to Neuroblastoma +/- Hirschsprung disease (Unknown)	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease (AD)
PICALM	No	No	Unknown	Possible association with acute myeloid leukemia (AML)	N/A
POLH	No	No	AR	Xeroderma pigmentosum, variant type (without neurologic abnormalities)	N/A
POT1	No	No	AD	Susceptibility to cutaneous malignant melanoma	Susceptibility to glioma
PRF1	No	No	AR, Unknown	Familial Hemophagocytic Lymphohistiocytosis-2 (FHL2)	Aplastic anemia; non-Hodgkin Lymphoma (both Unknown)
PRKDC	No	No	AR	Immunodeficiency 26, with or without neurologic abnormalities	N/A
PTCH1	No	No	AD	Nevoid Basal Cell Carcinoma syndrome (NBCSS; also known as Gorlin syndrome and Gorlin-Goltz syndrome)	Holoprosencephaly 7
PTCH2	No	No	AD	Nevoid Basal Cell Carcinoma syndrome (NBCSS; also known as Gorlin syndrome and Gorlin-Goltz syndrome)	N/A
RAD51B	No	No	AD	Possible association with various cancer types (RAD51B involved in recurring chromosomal translocation, t(6;14)(p21;q23-24))	N/A
RBM15	No	No	Unknown	Susceptbility to Acute megakaryoblastic leukemia	N/A
RHBDF2	No	No	AD	Susceptbility to Tylosis with esophageal cancer	N/A
ROBO2	No	No	Unknown, AD	Possible association with cholangiocarcinoma (Unknown)	Vesicoureteral reflux 2 (AD)
ROS1	No	No	Unknown	Possible association with lung cancer and other cancer types (chromosomal aberration involving ROS1 is found in a glioblastoma multiforme sample)	N/A
RPL11	No	No	AD	Diamond-Blackfan anemia 7	N/A
RPL15	No	No	AD	Diamond-Blackfan anemia 12	N/A
RPL26	No	No	AD	Diamond-Blackfan anemia 11	N/A
RPL27	No	No	AD	Diamond-Blackfan anemia 16	N/A
RPL31	No	No	Unknown	Diamond-Blackfan anemia	N/A
RPL35A	No	No	AD	Diamond-Blackfan anemia 5	N/A
RPL5	No	No	AD	Diamond-Blackfan anemia 6	N/A
RPS10	No	No	AD	Diamond-Blackfan anemia 9	N/A
RPS19	No	No	AD	Diamond-Blackfan anemia 1	N/A
RPS24	No	No	AD	Diamond-blackfan anemia 3	N/A
RPS26	No	No	AD	Diamond-blackfan anemia 10	N/A
RPS27	No	No	AD	Diamond-blackfan anemia 17	N/A
RPS28	No	No	AD	Diamond Blackfan anemia 15 with mandibulofacial dysostosis	N/A
RPS29	No	No	AD	Diamond-Blackfan anemia 13	N/A
RPS7	No	No	AD	Diamond-Blackfan anemia 8	N/A
RTEL1	No	No	AD, AR	Dyskeratosis congenita (AD, AR)	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3 (AD)
RUNX1	No	No	AD	Familial platelet disorder with associated myeloid malignancy	Susceptibility to acute myeloid leukemia
SBDS	No	No	AR, Unknown	Shwachman-Diamond syndrome (AR)	Susceptibility to Aplastic anemia (Unknown)
SH2D1A	No	No	XLR	X-linked lymphoproliferative syndrome-1	N/A
SLX4	No	No	AR	Fanconi anemia, complementation group P	N/A
SMARCE1	No	No	AD	Susceptibility to familial meningioma	Coffin-Siris syndrome 5
STAT3	No	No	Unknown, AD	Possible association with prostate cancer and other cancer types (Unknown)	Autoimmune disease, multisystem, infantile-onset, 1 (AD); Hyper-IgE recurrent infection syndrome (AD); Possible association with Autoimmune diseases, Crohn disease, and Shingles (Unknown)
SUFU	No	No	AD, AR	Nevoid Basal Cell Carcinoma syndrome (NBCSS; also known as Gorlin syndrome and Gorlin-Goltz syndrome) (AD)	Susceptibility to Familial meningioma (AD); Joubert syndrome (AR); Susceptibility to demoplastic medullablastoma (AR, AD)
TERC	No	No	AD	Dyskeratosis congenita	Susceptibility to Idiopathic Pulmonary Fibrosis; Aplastic anemia
TERT	No	No	AD, AR, Unknown	Dyskeratosis congenita (AD, AR)	Acute myeloid leukemia (AD); Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1 (AD); Susceptibility to cutaneous malignant melanoma (Unknown)
TINF2	No	No	AD	Dyskeratosis congenita	Revesz syndrome
TYR	No	No	Uknown, AR, AD	Susceptibility to cutaneous malignant melanoma (Unknown)	Oculocutaneous albinism types IA (AR) and IB (AR); Waardenburg syndrome (albinism, digenic; AD); Skin/hair/eye pigmentation 3, blue/green eyes (Unknown); Skin/hair/eye pigmentation 3, light/dark/freckling skin (Unknown)
UROD	No	No	AD, AR	Porphyria cutanea tarda (AD, AR); Porphyria, hepatoerythropoietic (AD, AR)	N/A
USB1	No	No	AR	Poikiloderma with neutropenia (AR)	N/A
WAS	No	No	XLR	Wiskott-Aldrich syndrome	Severe congenital Neutropenia; Thrombocytopenia
WRAP53	No	No	AR	Dyskeratosis congenita	N/A
WRN	No	No	AR	Werner syndrome	N/A
XPA	No	No	AR	Xeroderma pigmentosum, group A	N/A

	XPC	No	No	AR	Xeroderma pigmentosum, group C	N/A
	XRCC3	No	No	AD, Unknown	Possible association with breast cancer (AD), cutaneous malignant melanoma, and other cancer types (Unknown)	N/A
Cardiac Disease						
Cardiomyopathy (78 genes) • Hypertrophic cardiomyopathy • Dilated cardiomyopathy • Arrhythmogenic right ventricular cardiomyopathy • Left ventricular non-compaction	*ACTC1*	Yes	Yes	AD	Dilated cardiomyopathy; Hypertrophic cardiomyopathy; Left ventricular noncompaction	Atrial septal defect; restrictive cardiomyopathy
	DSC2	Yes	Yes	AD, AR	Arrythmogenic right ventricular cardiomyopathy (AD, AR)	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair (AR, AD)
	DSG2	Yes	Yes	AD, AR, Unknown	Dilated cardiomyopathy (Unknown)	Arrhythmogenic right ventricular dysplasia 10 (AD) Arrthythmogenic right ventriuclar cardiomyopathy (AR)
	DSP	Yes	Yes	AD, AR, Unknown	Arrythmogenic right ventricular cardiomyopathy (AD); Dilated cardiomyopathy (Unknown)	Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis (AD); Dilated cardiomyopathy with woolly hair and keratoderma (AR); Lethal acantholytic epidermolysis bullosa (AR); Skin fragility-woolly hair syndrome (AR); Keratosis palmoplantaris striata II (Unknown)
	GLA	Yes	Yes	XL	Hypertrophic cardiomyopathy (XL)	Fabry disease (XL)
	LMNA	Yes	Yes	AD, AR	Dilated cardiomyopathy (AD)	Congenital muscular dystrophy (AD); Limb-girdle muscular dystrophy, type 1B (AD); Heart-hand syndrome, Slovenian type (AD); Lipodystrophy, familial partial, type 2 (AD); Malouf syndrome (AD); Emery-Dreifuss muscular dystrophy (AD, AR); Hutchinson-Gilford progeria (AD, AR); Charcot-Marie-Tooth disease, type 2B1 (AR); Mandibuloacral dysplasia (AR); Restrictive dermopathy, lethal (AR)
	MYBPC3	Yes	Yes	AD	Dilated cardiomyopathy; Hypertrophic cardiomyopathy; Left ventricular noncompaction	N/A
	MYH7	Yes	Yes	AD, AR	Dilated cardiomyopathy (AD); Hypertrophic cardiomyopathy (AD); Left ventricular noncompaction (AD)	Laing distal myopathy (AD); Scapuloperoneal syndrome, myopathic type (AD); Myosin storage myopathy (AD,AR)
	MYL2	Yes	Yes	AD	Hypertrophic cardiomyopathy	N/A
	MYL3	Yes	Yes	AD	Hypertrophic cardiomyopathy	N/A
	PKP2	Yes	Yes	AD	Arrythmogenic right ventricular cardiomyopathy	N/A
	PRKAG2	Yes	Yes	AD	Hypertrophic cardiomyopathy	Glycogen storage disease of heart, lethal congenital; Wolff-Parkinson-White syndrome
	TMEM43	Yes	Yes	AD	Arrhythmogenic right ventricular cardiomyopathy	Emery-Dreifuss muscular dystrophy
	TNNI3	Yes	Yes	AD, AR, Unknown	Dilated cardiomyopathy (AR, Unknown); Hypertrophic cardiomyopathy (AD)	familial restrictive cardiomyopathy (AD)
	TNNT2	Yes	Yes	AD	Dilated cardiomyopathy; Hypertrophic cardiomyopathy; Left ventricular noncompaction	familial restrictive cardiomyopathy
	TPM1	Yes	Yes	AD	Dilated cardiomyopathy; Hypertrophic cardiomyopathy; Left ventricular noncompaction	N/A
	ACTA1	No	No	AD, AR	Dilated cardiomyopathy (AD); Hypertrophic cardiomyopathy (AD)	Scapulohumeroperoneal myopathy (AD); Congenital myopathy with fiber-type disproportion (AD,AR); Nemaline myopathy (Cap myopathy, Actin-accumulation myopathy, intranuclear rod myopathy) (AD, AR)
	ACTN2	No	No	AD	Dilated cardiomyopathy; Hypertrophic cardiomyopathy	N/A
	ALMS1	No	No	AR	Dilated cardiomyopathy (Alstrom syndrome)	Alstrom syndrome
	ANKRD1	No	No	AD	Dilated cardiomyopathy; Hypertrophic cardiomyopathy	N/A
	BAG3	No	No	AD	Dilated cardiomyopathy	Myofibrillar myopathy
	CALR3	No	No	AD	Hypertrophic cardiomyopathy	N/A
	CAV3	No	No	AD, AR	Hypertrophic cardiomyopathy (AD) Long QT (AD)	Elevated serum creatine phosphokinase (AD); Myopathy, distal, Tateyama type (AD); Rippling muscle disease (AD); Limb-girdle muscular dystrophy, type IC (AD, AR)
	CAVIN4	No	No	Unknown	Possible association with cardiomyopathy	N/A
	CHRM2	No	No	Unknown	Possible association with cardiomyopathy	Alcohol use disorder; Major depressive disorder
	COX15	No	No	AR, Mito	Hypertrophic cardiomyopathy (Leigh syndrome - AR, Mito)	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2 (AR); Leigh syndrome (AR, Mito)
	CRYAB	No	No	AD, AR	Dilated cardiomyopathy (AD)	Myofibrillar myopathy (AD); Cataract, multiple types (AD/AR); Myofibrillar myopathy, fatal infantile hypertonic, alpha-B crystallin-related (AR)
	CSRP3	No	No	AD	Dilated cardiomyopathy; Hypertrophic cardiomyopathy	N/A
	CTF1	No	No	Uknown	Dilated cardiomyopathy (preliminary evidence)	N/A
	DES	No	No	AD, AR	Dilated cardiomyopathy (AD)	Scapuloperoneal syndrome, neurogenic, Kaeser type (AD); Myobrillar myopathy (AD, AR); Limb-girdle muscular dystrophy, type 2R (AR)

DMD	No	No	XL	Dilated cardiomyopathy	Duchenne muscular dystrophy; Becker muscular dystrophy
DOLK	No		Unknown, AR	Dilated cardiomyopathy (Unknown)	Congenital disorder of glycosylation, type Im (AR)
DPP6	No	No	AD	Arrythmogenic right ventricular cardiomyopathy	Mental retardation
DTNA	No	No	Unknown, AD	Dilated cardiomyopathy (Unknown); Left ventricular noncompaction (with or without congenital heart defects) (AD)	N/A
EMD	No	No	XL	Dilated cardiomyopathy; Hypertrophic cardiomyopathy	Emery-Dreifuss muscular dystrophy
EYA4	No	No	AD	Dilated cardiomyopathy	Deafness
FHL1	No	No	XL	Dilated cardiomyopathy; Hypertrophic cardiomyopathy	Uruguay faciocardiomusculoskeletal syndrome; Emery-Dreifuss muscular dystrophy; Myopathy with postural muscle atrophy; Reducing body myopathy (severe, infantile or early childhood onset); Reducing body myopathy (late childhood or adult onset); Scapuloperoneal myopathy
FHL2	No	No	Unknown	Possible association with hypertrophic cardiomyopathy	N/A
GATAD1	No	No	AR	Dilated cardiomyopathy	N/A
HADHA	No	No	AR, Unknown	Possible association with cardiomyopathy (AR, Unknown)	LCHAD deficiency (AR); trifunctional protein deficiency (AR); Acute fatty liver of pregnancy and maternal HELLP syndrome of pregnancy (when fetus carries two variants)
ILK	No	No	Unknown	Possible association with dilated cardiomyopathy	N/A
JPH2	No	No	AD	Hypertrophic cardiomyopathy	N/A
JUP	No	No	AD/AR	Arrythmogenic right ventricular cardiomyopathy (AD)	Naxos disease (AR)
KLF10	No	No	Unknown	Possible association with hypertrophic cardiomyopathy	N/A
LAMA4	No	No	AD	Dilated cardiomyopathy	N/A
LAMP2	No	No	XL	Dilated cardiomyopathy; Hypertrophic cardiomyopathy	Danon disease
LDB3	No	No	AD	Dilated cardiomyopathy; Hypertrophic cardiomyopathy; Left ventricular noncompaction	Myofibrillar myopathy
MIB1	No	No	Unknown	Possible association with Left ventricular noncompaction	N/A
MYH6	No	No	AD, Unknown	Dilated cardiomyopathy (Unknown); Hypertrophic cardiomyopathy (AD)	Sick sinus syndrome (Unknown); Atrial septal defect (Unknown)
MYLK2	No	No	Digenic	Hypertrophic cardiomyopathy	N/A
MYO6	No	No	AD, AR	Hypertrophic cardiomyopathy (with deafness) (AD)	Deafness (AD, AR)
MYOM1	No	No	Unknown	Possible association with dilated cardiomyopathy; Possible association with hypertrophic cardiomyopathy	N/A
MYOZ2	No	No	AD	Hypertrophic cardiomyopathy	N/A
MYPN	No	No	AD, AR	Dilated cardiomyopathy (AD); Hypertrophic cardiomyopathy (AD)	Familial restrictive cardiomyopathy (AD); Nemaline myopathy (AR)
NEBL	No	No	Unknown	Possible association with dilated cardiomyopathy	N/A
NEXN	No	No	AD	Dilated cardiomyopathy; Hypertrophic cardiomyopathy	N/A
PDLIM3	No	No	Unknown	Possible association with hypertrophic cardiomyopathy	N/A
PLN	No	No	Unknown, AD	Dilated cardiomyopathy (Unknown); Hypertrophic cardiomyopathy (AD)	N/A
PRDM16	No	No	AD	Dilated cardiomyopathy; Left ventricular noncompaction	N/A
PSEN1	No	No	AD	Dilated cardiomyopathy	Alzheimer disease; Alzheimer disease with spastic paraparesis and apraxia; Alzheimer disease with spastic paraparesis and unusual plaques; Frontotemporal dementia; Pick disease; Familial acne inversa
PSEN2	No	No	AD	Dilated cardiomyopathy	Alzheimer disease
RBM20	No	No	AD	Dilated cardiomyopathy	N/A
SCO2	No	No	AD, AR	Hypertrophic cardiomyopathy (AR)	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency (AR); Myopia (AD)
SELENON	No	No	AD, AR	Dilated cardiomyopathy (AD, AR)	Congenital myopathy with fiber-type disproportion (AD, AR); Rigid spine muscular dystrophy (AR); Multiminicore disease (AR)
SGCB	No	No	AR	Dilated cardiomyopathy	Limb-girdle muscular dystrophy, type 2E
SGCD	No	No	Unknown, AR	Dilated cardiomyopathy (Unknown)	Limb-girdle muscular dystrophy, type 2F (AR)
SLC25A13	No	No	AR	Possible association with hypertrophic cardiomyopathy	Citrullinemia, type II (adult-onset and neonatal-onset)

	SLC25A4	No	No	AD, AR	Hypertrophic cardiomyopathy (AD, AR)	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) (AD) and 12B (AR); Progressive external ophthalmoplegia with mitochondrial DNA deletions (AD)
	TAZ	No	No	XL	Dilated cardiomyopathy; Left ventricular noncompaction	Barth syndrome
	TCAP	No	No	AD, AR	Hypertrophic cardiomyopathy (AD)	Limb-girdle muscular dystrophy, type 2G (AR)
	TMPO	No	No	Unknown	Possible association with dilated cardiomyopathy	N/A
	TNNC1	No	No	Unknown, AD	Dilated cardiomyopathy (Unknown); Hypertrophic cardiomyopathy (AD)	N/A
	TRIM63	No	No	Unknown	Possible association with hypertrophic cardiomyopathy	N/A
	TTN	No	No	Unknown, AD, AR	Dilated cardiomyopathy (Unknown); Hypertrophic cardiomyopathy (AD)	Tibial muscular dystrophy, tardive (AD); Myopathy, proximal, with early respiratory muscle involvement (AD); Limb-girdle muscular dystrophy, type 2J (AR); Salih myopathy (AR)
	TTR	No	No	AD	Possible association with cardiomyopathy	Dystransthyretinemic hyperthyroxinemia; Carpal tunnel syndrome, familial
	TXNRD2	No	No	Unknown, AR	Possible association with dilated cardiomyopathy (Unknown)	Glucocorticoid deficiency (AR)
	VCL	No	No	Unknown, AD	Dilated cardiomyopathy (Unknown); Hypertrophic cardiomyopathy (AD)	N/A
	ZBTB17	No	No	AD	Possible association with dilated cardiomyopathy	N/A
RASopathy (13 genes) • Noonan syndrome • Costello syndrome • Cardiofaciocutaneous syndrome	*BRAF*	No	No	AD	Hypertrophic cardiomyopathy	Noonan syndrome; Noonan syndrome with multiple lentigenes; Cardiofaciocutaneous syndrome
	CBL	No	No	AD	Hypertrophic cardiomyopathy; Susceptibiltity to cancer (juvenile myelomonocytic leukemia)	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia
	HRAS	No	No	AD	Hypertrophic cardiomyopathy	Costello syndrome
	KRAS	No	No	AD, Unknown	Hypertrophic cardiomyopathy (AD); Susceptibiltity to cancer (Possibly AD/Unknown, usually somatic)	Noonan syndrome (AD); Cardiofaciocutaneous syndrome (AD); RAS-associated autoimmune leukoproliferative disorder (AD); Leukemia, acute myeloid (AD)
	LZTR1	No	No	AD	Hypertrophic cardiomyopathy	Noonan syndrome; Schwannomatosis
	MAP2K1	No	No	AD	Hypertrophic cardiomyopathy	Cardiofaciocutaneous syndrome
	MAP2K2	No	No	AD	Hypertrophic cardiomyopathy	Cardiofaciocutaneous syndrome
	NRAS	No	No	AD	Hypertrophic cardiomyopathy	Noonan sydrome
	PTPN11	No	No	AD	Hypertrophic cardiomyopathy	Noonan syndrome; Noonan syndrome with multiple lentigenes; Metachondromatosis
	RAF1	No	No	AD, Unknown	Dilated cardiomyopathy (AD)	Noonan syndrome (AD); Noonan syndrome with multiple lentigenes (Unknown)
	RIT1	No	No	AD	Hypertrophic cardiomyopathy	Noonan syndrome
	SHOC2	No	No	AD	Possible association with cardiomyopathy	Noonan-like syndrome with loose anagen hair
	SOS1	No	No	AD	Hypertrophic cardiomyopathy	Noonan sydrome; Fibromatosis, gingival
Channelopathy and Arrhythmia (34 genes) • Romano-Ward Long QT syndromes types 1, 2, and 3 • Brugada syndrome • Catecholaminergic polymorphic ventricular tachycardia	*KCNQ1*	Yes	Yes	AD, AR	Long QT syndrome (AD); Atrial fibrillation, familial (AD); Short QT syndrome (AD); Jervell and Lange-Nielsen syndrome (AR)	N/A
	SCN5A	Yes	Yes	AD, AR, Unknown	Long QT syndrome (AD); Brugada syndrome (AD); Atrial fibrillation, familial (AD); Dilated cardiomyopathy (AD)	Heart block (progressive and nonprogressive), type IA (AD); Sick sinus syndrome (AR); Susceptibility to Sudden Infant Death Syndrome (AR): Ventricular fibrillation, familial (Unknown)
	KCNH2	Yes	Yes	AD, Unknown	Long QT syndrome (AD); Short QT syndrome (Unknown)	N/A
	RYR2	Yes	Yes	AD	Catecholaminergic polymorphic ventricular tachycardia; Arrythmogenic right ventricular cardiomyopathy	N/A
	ABCC9	No	No	AD	Atrial fibrillation, familial; Dilated cardiompyopathy	Hypertrichotic osteochondrodysplasia (Cantu syndrome)
	AKAP9	No	No	AD	Possible association with Long QT syndrome; Modifier of Romano-Ward Syndrome	N/A
	ANK2	No	No	AD	Cardiac arrhythmia, ankyrin-B-related	N/A
	CACNA1C	No	No	Unknown, AD	Brugada syndrome (Unknown)	Timothy syndrome (AD)
	CACNB2	No	No	AD	Brugada syndrome	N/A
	CALM1	No	No	AD	Long QT syndrome; Catecholaminergic polymorphic ventricular tachycardia	N/A
	CALM2	No	No	AD	Long QT syndrome	N/A
	CASQ2	No	No	AR	Catecholaminergic polymorphic ventricular tachycardia	N/A
	GJA5	No	No	AD, Digenic	Atrial fibrillation, familial (AD); Atrial standstill (digenic - GJA5/SCN5A)	N/A
	GPD1L	No	No	AD	Brugada syndrome	N/A
	HCN4	No	No	AD	Brugada syndrome	Sick sinus syndrome
	KCNA5	No	No	AD	Atrial fibrillation, familial	N/A
	KCND3	No	No	AD	Brugada syndrome	Spinocerebellar ataxia
	KCNE1	No	No	AD, AR	Long QT syndrome (AD); Jervell and Lange-Nielsen syndrome (AR)	N/A
	KCNE2	No	No	AD	Long QT syndrome; Atrial fibrillation, familial	N/A

	KCNE3	No	No	AD	Brugada syndrome	N/A
	KCNE5	No	No	AD	Brugada syndrome	N/A
	KCNJ2	No	No	AD, Unknown	Atrial fibrillation, familial (AD); Short QT syndrome (Unknown)	Andersen syndrome (AD)
	KCNJ5	No	No	AD	Long QT syndrome	Hyperaldosteronism, familial, type III
	KCNJ8	No	No	AD	Brugada syndrome	N/A
	NPPA	No	No	AD, AR	Atrial fibrillation, familial (AD); Atrial standstill (AR)	N/A
	RANGRF	No	No	AD	Brugada syndrome	N/A
	SCN1B	No	No	AD, AR, Unknown	Atrial fibrillation, familial (AD); Brugada syndrome (Unknown)	Epilepsy, generalized, with febrile seizures plus, type 1 (AD); Epileptic encephalopathy, early infantile (AR); Cardiac conduction defect, nonspecific (Unknown)
	SCN2B	No	No	AD	Atrial fibrillation, familial	N/A
	SCN3B	No	No	AD	Atrial fibrillation, familial; Brugada syndrome	N/A
	SCN4B	No	No	AD	Long QT syndrome; Atrial fibrillation, familial	N/A
	SLMAP	No	No	AD	Brugada syndrome	N/A
	SNTA1	No	No	AD	Long QT syndrome	N/A
	TRPM4	No	No	AD	Progressive familial heart block, type IB	N/A
	TRDN	No	No	AR	Catecholaminergic polymorphicv entricular tachycardia, with or without muscle weakness	N/A
Familial Hypercholesterolemia (14 genes)	*APOB*	Yes	Yes	AD, AR	Familial hypercholesterolemia (AD)	Hypobetalipoproteinemia (AR)
	LDLR	Yes	Yes	AD	Familial hypercholesterolemia	N/A
	PCSK9	Yes	Yes	AD	Familial hypercholesterolemia	N/A
	ABCG5	No	No	AR	Sitosterolemia	N/A
	ABCG8	No	No	AR	Sitosterolemia	N/A
	APOA5	No	No	AD	Familial hypercholesterolemia; Susceptibility to familial hypertriglyceridemia	N/A
	APOC2	No		AR	Hyperlipoproteinemia, type Ib	N/A
	APOE	No	No	AD, AR	Hyperlipoproteinemia, type III (AD, AR)	Susceptibility to Alzheimer disease (AD); lipoprotein glomerulopathy (AD); age-related macular degeneration (AD); coronary artery disease (AD/AR); sea-blue histiocyte disease (AR)
	CETP	No	No	AD	Hyperalphalipoproteinemia	N/A
	CREB3L3	No	No	Unknown	Hypertriglyceridemia	N/A
	GPIHBP1	No	No	AD, AR	Hyperlipoproteinemia, type 1D (AD)	Hyperlipoproteinemia, type 1D (AR)
	LDLRAP1	No	No	AR	Familial hypercholesterolemia	N/A
	LMF1	No	No	AR	Combined lipase deficiency	N/A
	SREBF2	No	No	Unknown	Possible association with familial hypercholesterolemia	N/A
Connective Tissue						
Marfan syndrome, Loeys-Dietz syndromes, and Familial Thoracic Aortic Aneurysms and Dissections (21 genes) (includes other conditions with vascular involvement)	*ACTA2*	Yes	Yes	AD	Familial thoracic aortic aneurysm	Moyamoya disease; Multisystemic smooth muscle dysfunction syndrome
	FBN1	Yes	Yes	AD, Unknown	Marfan syndrome (AD)	Acromicric dysplasia (AD); familial ectopia lentis (AD); geleophysic dysplasia (AD); Marfan lipodystrophy syndrome (AD); stiff skin syndrome (AD); Weill-Marchesani syndrome (AD); MASS syndrome (Unknown)
	MYH11	Yes	Yes	AD	Familial thoracic aortic aneurysm	N/A
	SMAD3	Yes	Yes	AD	Loeys-Dietz syndrome	N/A
	TGFBR1	Yes	Yes	AD	Loeys-Dietz syndrome	Susceptibility to multiple self-healing squamous epithelioma
	TGFBR2	Yes	Yes	AD	Loeys-Dietz syndrome	Colorectal cancer, hereditary nonpolyposis
	CCM2	No	No	AD	Cerebral cavernous malformations	N/A
	ELN	No	No	AD	Cutis laxa	Supravalvar aortic stenosis
	FBN2	No	No	AD	Congenital contractural arachnodactyly	Macular degeneration, early-onset
	LTBP2	No	No	AR	Weill-Marchesani syndrome	Primary congenital glaucoma; icrospherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma
	MYLK	No	No	AD	Familial thoracic aortic aneurysm	N/A
	NKX2-5	No	No	AD, Unknown	Possible association with cardiomyopathy (AD)	Atrial septal defect, with or without AV conduction defects (AD); Hypoplastic left heart syndrome (AD); Hypothyroidism, congenital nongoitrous (AD); Tetralogy of Fallot (AD); ventricular septal defect (AD); conotruncal heart malformations (variable) (Unknown)
	NODAL	No	No	AD	Visceral heterotaxy	N/A
	NOTCH1	No	No	AD	Aortic valve disease	Adams-Oliver syndrome
	NOTCH3	No	No	AD	CADASIL	Myofibromatosis, infantile; lateral meningocele syndrome
	PDCD10	No	No	AD	Cerebral cavernous malformations	N/A
	PRKG1	No	No	AD	Familial thoracic aortic aneurysm	N/A
	PRNP	No	No	AD, Unknown	Cerebral amyloid angiopathy, PRNP-related (AD)	Creutzfeldt-Jakob disease (AD); Prion disease with protracted course (AD); Gerstmann-Straussler disease (AD); Huntington disease-like (AD); Insomnia, fatal familial (AD); susceptibility to Kuru (Unknown)
	SLC2A10	No	No	AR	Arterial tortuosity syndrome	N/A
	TGFB2	No		AD	Loeys-Dietz syndrome	N/A

	TGFB3	No	No	AD	Loeys-Dietz syndrome; Arrthmogenic right ventricular cardiomyopathy	N/A
Ehlers-Danlos syndrome (17 genes) (vascular and other types)	*COL3A1*	Yes	Yes	AD	Ehlers-Danlos syndrome, vascular type	N/A
	ADAMTS2	No	No	AR	Ehlers-Danlos syndrome, dermatosparaxix type	N/A
	B3GALT6	No	No	AR	Ehlers-Danlos syndrome, spondylodysplastic type	Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures
	B4GALT7	No	No	AR	Ehlers-Danlos syndrome, spondylodysplastic type	N/A
	CHST14	No	No	AR	Ehlers-Danlos syndrome, musculocontractural type	N/A
	COL1A1	No	No	AD	Ehlers-Danlos syndrome, arthrochalasia type; Ehlers-Danlos, classic type	Caffey disease; Osteogenesis imperfecta, types I, II, III, IV
	COL1A2	No	No	AD, AR	Ehlers-Danlos syndrome, arthrochalasia type (AD); Ehlers-Danlos syndrome, cardiac valvular type (AR)	Oseogenesis imperfecta, types II, III, and IV (AD)
	COL5A1	No	No	AD	Ehlers-Danlos syndrome, classic type	N/A
	COL5A2	No	No	AD	Ehlers-Danlos syndrome, classic type	N/A
	DSE	No	No	AR	Ehlers-Danlos syndrome, musculocontractural type	N/A
	EFEMP2	No	No	AR	Cutis laxa, autosomal recessive, type IB	N/A
	FKBP14	No	No	AR	Ehlers-Danlos syndrome, kyphoscoliotic type	N/A
	PLOD1	No	No	AR	Ehlers-Danlos syndrome, kyphoscoliotic type	N/A
	PRDM5	No	No	AR	Ehlers-Danlos syndrome	Brittle cornea syndrome
	SLC39A13	No	No	AR	Ehlers-Danlos syndrome	Spondylocheirodysplasia (Ehlers-Danlos syndrome-like)
	TNXB	No	No	AD, AR	Ehlers-Danlos syndrome, classic-like and hypermobile (AR)	Vesicoureteral reflux (AD)
	ZNF469	No	No	AR	Ehlers-Danlos syndrome	Brittle cornea syndrome
Other Actionable Diseases						
Malignant Hyperthermia Susceptibility (3 genes)	*CACNA1S*	Yes	Yes	AD	Malignant hyperthermia susceptibility	Hypokalemic periodic paralysis
	RYR1	Yes	Yes	AD, AR	Malignant hyperthermia susceptibility (AD)	King-Denborough syndrome (AD); Central core disease (AD, AR); Neuromuscular disease, congenital, with uniform type 1 fiber (AD, AR); Minicore myopathy with external ophthalmoplegia (AR)
	CACNA2D1	No	No	AD	Malignant hyperthermia; Possible association with Brugada syndrome	N/A
Ornithine Transcarbamylase Deficiency	*OTC*	Yes	Yes	XL	Ornithine transcarbamylase deficiency	N/A
Wilson's Disease	*ATP7B*	Yes	Yes	AR	Wilson Disease	N/A
Other (29 genes)	*ABCD1*	No	No	XL	Adrenoleukodystrophy; Adrenomyeloneuropathy	N/A
	ACVRL1	No	No	AD	Hereditary hemorrhagic telangiectasia	N/A
	APOA4	No	No	Unknown	Possible association with lipid metabolism disease	N/A
	CASR	No	No	AD, AR	Hypocalcemia (AD); Hyperparathyroidism, neonatal (AD, AR); Hypocalciuric hypercalcemia, type I (AD)	N/A
	CRELD1	No	No	AD	Atrioventricular septal defect with heterotaxy syndrome	N/A
	EGR2	No	No	AD, AR	Charcot-Marie-Tooth disease, type 1D (AD); Dejerine-Sottas disease (AD, AR); Neuropathy, congenital hypomyelinating (AD, AR)	N/A
	F5	No	No	AD, AR	Factor V deficiency (AR); Susceptibility to thrombophilia due to factor V Leiden (AD)	N/A
	F8	No	No	XL	Hemophilia A	N/A
	F9	No	No	XL	Hemophilia B	N/A
	FKRP	No	No	AR	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies; congenital with or without mental retardation; limb-girdle)	N/A
	FXN	No	No	AR	Freidreich ataxia	N/A
	GCKR	No	No	Unknown	Possible effect on fasting plasma glucose levels; non-alcoholic fatty liver disease	N/A
	GDF2	No	No	AD	Hereditary hemorrhagic telangiectasia	N/A
	HSPB8	No	No	AD	Charcot-Marie-Tooth disease, axonal, type 2L; Neuropathy, distal hereditary motor, type IIA	N/A
	JAG1	No	No	AD	Alagille syndrome; Tetralogy of Fallot	N/A
	LITAF	No	No	AD	Charcot-Marie-Tooth disease, type 1C	N/A
	MPZ	No	No	AD, AR	Charcot-Marie-Tooth disease, types dominant intermediate D, type 1B, type 2I, type 2J (all AD); Roussy-Levy syndrome (AD); Dejerine-Sottas disease (AD, AR); Neuropathy, congenital hypomyelinating (AD, AR)	N/A
	MYCN	No	No	AD	Feingold syndrome	N/A
	NEFL	No	No	AD, AR	Charcot-Marie-Tooth disease, dominant intermediate G and type 2E (AD); type 1F (AR)	N/A
	PKD2	No	No	AD	Polycystic kidney disease	N/A
	PMP22	No	No	AD, AR	Charcot-Marie-Tooth disease, types 1A & 1E (AD); Neuropathy, inflammatory demyelinating (AD); Neuropathy, recurrent, with pressure palsies (AD); Roussy-Levy syndrome (AD); Dejerine-Sottas disease (AD, AR)	N/A

	PRKN	No	No	AR	Parkinson disease, juvenile (AR)	N/A
	SALL4	No	No	AD	Duane-radial ray syndrome; IVIC syndrome	N/A
	SERPINA1	No	No	AR	Alpha-1 antitrypsin deficiency	N/A
	TBX20	No	No	AD	Atrial septal defect	N/A
	TBX3	No	No	AD	Ulnar mammary syndrome	N/A
	TBX5	No	No	AD	Holt Oram syndrome	N/A
	ZHX3	No	No	Unknown	Possible association with congenital heart defects	N/A
	ZIC3	No	No	XL	Congenital heart defects (nonsyndromic); Visceral heterotaxy; VACTERL association	N/A

**** Informed by the American College of Medical Genetics and Genomics (PMID: 23788249)***

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Veritas
The Genome Company

myGenome Standard/Premium Carrier Gene-Disease Reference List for Providers

Gene*	Included in myGenome Standard	Disease**	Other Associated Diseases**
ABCC8	Yes	Familial hyperinsulinism; Neonatal diabetes mellitus	Familial hyperinsulinism (AD); Hypoglycemia of infancy, leucine-sensitive (AD); Neonatal diabetes mellitus (AD)
ACADM	Yes	Medium-chain acyl-CoA dehydrogenase deficiency	
ASPA	Yes	Canavan disease	
BCKDHA	Yes	Maple syrup urine disease, type Ia	
BCKDHB	Yes	Maple syrup urine disease, type Ib	
BLM	Yes	Bloom syndrome	
CFTR	Yes	Congenital bilateral absence of vas deferens; Cystic fibrosis	Hereditary Pancreatitis (AD); Bronchiectasis with or without elevated sweat chloride (modifier of) (AD)
CLRN1	Yes	Retinitis pigmentosa; Usher syndrome, type 3A	
DHCR7	Yes	Smith-Lemli-Opitz syndrome	
ELP1	Yes	Familial dysautonomia	
G6PC	Yes	Glycogen storage disease Ia	
GALK1	Yes	Galactokinase deficiency with cataracts	
GALT	Yes	Galactosemia	
GLB1	Yes	GM1-gangliosidosis, type I, type II, type III; Mucopolysaccharidosis type IVB (Morquio)	
HBB	Yes	Sickle cell anemia; Beta-thalassemia	Delta-beta thalassemia (AD); Heinz body anemia (AD); Hereditary persistence of fetal hemoglobin (AD)
HEXA	Yes	Tay-Sachs disease	
MCOLN1	Yes	Mucolipidosis IV	
MYO7A	Yes	Deafness; Usher syndrome, type 1B	Deafness (AD)
PAH	Yes	Phenylketonuria; Hyperphenylalaninemia (non-PKU mild)	
SMPD1	Yes	Niemann-Pick disease, types A & B	
USH2A	Yes	Retinitis pigmentosa; Usher syndrome, type 2A	
ACADSB	No	Short/branched chain acyl-CoA dehydrogenase deficiency	
ACADVL	No	Very long-chain acyl-CoA dehydrogenase deficiency	
ACSF3	No	Combined Malonic and Methylmalonic Aciduria	
AGA	No	Aspartylglucosaminuria	
AGL	No	Glycogen storage disease type III	
AIRE	No	Autoimmune polyendocrinopathy syndrome, type I	
ALDOB	No	Hereditary fructose intolerance	
ALPL	No	Hypophosphatasia	
ARSA	No	Metachromatic Leukodystrophy	
ASNS	No	Asparagine synthetase deficiency	
BBS1	No	Bardet-Biedl syndrome 1	
BBS2	No	Bardet-Biedl syndrome 2; Retinitis pigmentosa	
BCHE	No	Pseudocholinesterase deficiency	
BTD	No	Biotinidase deficiency	
CBS	No	Homocystinuria; Thrombosis, hyperhomocysteinemic	
CERKL	No	Retinitis pigmentosa; Cone-rod dystrophy	
CHRNE	No	Congenital myasthenic syndrome	Congenital myasthenic syndrome (AD form)
CLN5	No	Ceroid lipofuscinosis, neuronal, 5	
CNGB3	No	Achromatopsia; Juvenile macular degeneration	
CPT1A	No	Carnitine palmitoyl transferase I deficiency	

CPT2	No	Carnitine palmitoyl transferase II deficiency	
CTNS	No	Cystinosis	
CYBA	No	Chronic granulomatous disease	
CYP11B1	No	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	Aldosteronism, glucocorticoid-remediable (AD)
CYP11B2	No	Hypoaldosteronism, congenital, due to CMO I or CMO II deficiency	
CYP1B1	No	Anterior segment dysgenesis; Glaucoma	
CYP21A2	No	Hyperandrogenism and congenital adrenal hyperplasia, both due to 21-hydroxylase deficiency	
CYP27A1	No	Cerebrotendinous xanthomatosis	
DCLRE1C	No	Omenn syndrome; Severe combined immunodeficiency	
DNAJC19	No	3-methylglutaconic aciduria, type V	
DPYD	No	Dihydropyrimidine dehydrogenase deficiency	
DYSF	No	Miyoshi muscular dystrophy; Limb-girdle muscular dystrophy, type 2B; Distal myopathy with anterior tibial onset	
ERCC6	No	Cockayne syndrome; UV-sensitive syndrome	Premature ovarian failure 11 (AD); Susceptibility to lung cancer (AD); Age-related macular degeneration (unknown); Cerebrooculofacioskeletal syndrome 1; De Sanctis-Cacchione syndrome
ETFDH	No	Glutaric acidemia IIC	
EVC	No	Ellis-van Creveld syndrome	Weyers acrofacial dysostosis (AD)
EYS	No	Retinitis pigmentosa	
F11	No	Factor XI deficiency	
FAH	No	Tyrosinemia, type I	
FAM161A	No	Retinitis pigmentosa	
FKTN	No	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies); Limb-girdle muscular dystrophy	Dilated cardiomyopathy
G6PD	No	Glucose-6-phosphate dehydrogenase deficiency (XL)	
GAA	No	Pompe disease (glycogen storage disease II)	
GALC	No	Krabbe disease	
GBA	No	Gaucher disease	Susceptibility to Lewy body dementia (AD) and late-onset Parkinson disease (AD, Multifactorial)
GBE1	No	Glycogen storage disease IV; Polyglucosan body disease, adult form	
GCDH	No	Glutaricaciduria, type I	
GJB2	No	Nonsyndromic hearing loss	Deafness (AD); Bart-Pumphrey syndrome (AD); Hystrix-like ichthyosis with deafness (AD); Keratitis-ichthyosis-deafness syndrome (AD); Keratoderma, palmoplantar, with deafness (AD); Vohwinkel syndrome (AD)
GJB6	No	Deafness	Deafness (AD, digenic GJB2/GJB6); Ectodermal dysplasia 2, Clouston type (AD)
GLE1	No	Lethal congenital contracture syndrome 1	Arthrogryposis, lethal, with anterior horn cell disease (Unknown)
GNE	No	Nonaka myopathy	Sialuria (AD)
HEXB	No	Sandhoff disease, infantile, juvenile, and adult forms	
HFE	No	Hemochromatosis	Susceptibility to porphyria variegata (AD); Susceptibility to porphyria cutanea tarda (AD, AR)
HLCS	No	Holocarboxylase synthetase deficiency	
HPS1	No	Hermansky-Pudlak syndrome 1	
HYLS1	No	Hydrolethalus syndrome	
IVD	No	Isovaleric acidemia	
LAMA2	No	Muscular dystrophy, congenital merosin-deficient	
LIPA	No	Cholesteryl ester storage disease; Wolman disease	
LPL	No	Lipoprotein lipase deficiency	Combined hyperlipidemia, familial (AD)
LRPPRC	No	Leigh syndrome, French-Canadian type	
MED17	No	Microcephaly, postnatal progressive, with seizures and brain atrophy	
MEFV	No	Familial Mediterranean fever	Familial Mediterranean fever (AD form)
MESP2	No	Spondylocostal dysostosis	
MKS1	No	Joubert syndrome; Meckel syndrome; Bardet-Biedl syndrome	
MLC1	No	Megalencephalic leukoencephalopathy with subcortical cysts	
MPV17	No	Mitochondrial DNA depletion syndrome (hepatocerebral type)	
MTHFR	No	Homocystinuria due to MTHFR deficiency	
MUT	No	Methylmalonic aciduria, mut(0) type	
NDRG1	No	Charcot-Marie-Tooth disease, type 4D	
NDUFS6	No	Mitochondrial complex I deficiency	

NPHS1	No	Nephrotic syndrome, type 1	
OPA3	No	3-methylglutaconic aciduria, type III	Optic atrophy 3 with cataract (AD)
PC	No	Pyruvate carboxylase deficiency	
PCDH15	No	Deafness; Usher syndrome, type 1F	Usher syndrome, type 1D/F (digenic)
PEX12	No	Zellweger spectrum disorder	
PEX6	No	Zellweger spectrum disorder; Heimler syndrome	
PKHD1	No	Polycystic kidney disease	
PMM2	No	Congenital disorder of glycosylation, type Ia	
POLG	No	Mitochondrial DNA depletion syndrome, Alpers type and MNGIE type; Mitochondrial recessive ataxia syndrome; Progressive external ophthalmoplegia	Progressive external ophthalmoplegia (AD form)
PPT1	No	CLN1 disease (Ceroid lipofuscinosis, neuronal, 1)	
PYGM	No	McArdle disease	
RMRP	No	Anauxetic dysplasia; Cartilage-hair hypoplasia; Metaphyseal dysplasia without hypotrichosis	
RPE65	No	Leber congenital amaurosis; Retinitis pigmentosa	
SACS	No	Spastic ataxia, Charlevoix-Saguenay type	
SEPSECS	No	Pontocerebellar hypoplasia type 2D	
SGCG	No	Limb-girdle muscular dystrophy	
SLC12A3	No	Gitelman syndrome	
SLC12A6	No	Andermann syndrome (Agenesis of the corpus callosum with peripheral neuropathy)	
SLC17A5	No	Sialic acid storage disorder (infantile, Salla disease, intermediate severe Salla disease)	
SLC22A5	No	Primary carnitine deficiency	
SLC25A15	No	Ornithine translocase deficiency (Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome)	
SLC26A2	No	Achondrogenesis Ib	
SLC26A4	No	Deafness with enlarged vestibular aqueduct; Pendred syndrome	
SPG11	No	Amyotrophic lateral sclerosis, juvenile; Charcot-Marie-Tooth disease, axonal, type 2X; Spastic paraplegia	
TCIRG1	No	Osteopetrosis	
TECPR2	No	Spastic paraplegia	
TOR1A	No	Dystonia-1, torsion (AR, AD)	
TPP1	No	CLN2 disease (Ceroid lipofuscinosis, neuronal, 2); Spinocerebellar ataxia	
TRMU	No	Transient infantile liver failure	
TSFM	No	Combined oxidative phosphorylation deficiency	

**** Informed by American College of Obstetrics and Gynecology (ACOG) Policies for expanded carrier screening recommendations (genes associated with a carrier frequency of 1 in 100 or greater were selected)***

***** All diseases are inherited in an autosomal recessive manner unless otherwise noted.***

AD = autosomal dominant

XL = X-linked

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