Genome-wide Sequencing Ontario

Mature Minor Secondary Findings Choices

TYPE OF RESULT

PRIMARY FINDINGS

Disease-causing variants in genes that explain your medical condition

SECONDARY FINDINGS

Disease-associated variants in genes that are not related to the primary medical conditions for which the test has been offered, but which are associated with a high risk for other medical conditions during childhood and/or later in life.

What does *medically actionable* mean?

81 different genes are currently considered *medically actionable* secondary findings because if a disease-associated variant is found, there are clear medical recommendations that may be made to reduce the risk that the genetic variant will impact a person's health in the future.

Only secondary findings identified in you can be analyzed in other family members participating in genome-wide sequencing. The choice you make regarding secondary findings will not impact the results of your test.

Actionable in Childhood

All patients under 18 will automatically have analysis of 71 genes that are medically actionable in childhood

All Secondary Findings

You may **choose** whether **you** wish to have analysis of all available secondary findings genes.

This includes an additional 10 genes that are not medically actionable until adulthood

For more information on Genome-wide Sequencing Ontario and other patient resources please visit www.gsontario.ca

Genome-wide Sequencing Ontario Secondary Findings List

CONDITION	GENE
Adenomatous polyposis coli	APC
Aortic aneurysm, familial thoracic	ACTA2
	MYH11
Arrhythmogenic right ventricular cardiomyopathy	DSC2
	DSG2
	DSP
	PKP2
	TMEM43
Biotinitidase deficiency	BTD
Catecholaminergic polymorphic ventricular tachycardia	CASQ2
	RYR2
	TRDN
Ehlers-Danlos syndrome, vascular type	COL3A1
Familial hypercholesterolemia	APOB
	LDLR
	PCSK9
Familial hypertrophic cardiomyopathy, dilated cardiomyopathy	ACTC1
	BAG3
	DES
	FLNC
	GLA
	LMNA
	MYBPC3
	MYH7
	MYL2
	MYL3
	PRKAG2
	RBM20
	TNNC1
	TNNI3
	TNNT2
	TPM1
	TTN
Hereditary breast and/or ovarian cancer	BRCA1*
	BRCA2*
	PALB2*
Hereditary hemochromatosis	HFE* (C282Y only)
Hereditary hemorrhagic telangiectasia	ACVRL1
	ENG
Hereditary TTR amyloidosis	TTR*

CONDITION	GENE
Juvenile polyposis syndrome	BMPR1A
	SMAD4
Li-Fraumeni syndrome	TP53
Loeys-Dietz syndrome	SMAD3
	TGFBR1
	TGFBR2
Long QT syndrome, Brugada syndrome	KCMH2
	KCNQ1
	SCN5A
	CALM1
	CALM2
	CALM3
Lynch syndrome	MLH1*
	MSH2*
	MSH6*
	PMS2*
Malignant hyperthermia susceptibility	CACNA1S
	RYR1
Marfan syndrome	FBN1
Maturity-onset diabetes of the young	HNF1A
Multiple endocrine neoplasia type 1	MEN1
Multiple endocrine neoplasia, type 2 and Familial medullary thyroid carcinoma	RET
MUTYH-associated polyposis	MUTYH*
Neurofibromatosis, type 2	NF2
Ornithine carbamoyltransferase deficiency	OTC
Paraganglioma-pheochromocytoma syndrome	MAX
	SDHAF2
	SDHB
	SDHC
	SDHD
	TMEM127
Peutz-Jeghers syndrome	STK11
Pompe disease	GAA
PTEN hamartoma tumor syndrome	PTEN
Retinoblastoma	RB1
RPE65-related retinopathy	RPE65
Tuberous sclerosis	TSC1
	TSC2
Von Hippel-Lindau syndrome	VHL
Wilms tumor	WT1
Wilson disease	ATP7B

* Indicates genes that are not *medically actionable* until adulthood