Genome-wide Sequencing Ontario

Adult with Power of Attorney or Substitute Decision Maker 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.

Secondary Findings Analysis

The laboratory will search for disease-associated variants in 84 *medically actionable* disease genes

What does *medically actionable* mean?

84 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 the patient can be analyzed in other family members participating in genome-wide sequencing. The choice that is made regarding secondary findings will not impact the results of the patient's test.

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
Genes related to cardiovascular disease predisposition	
Aortic aneurysm, familial thoracic	ACTA2
	MYH11
Marfan syndrome	FBN1
Loeys-Dietz syndrome	SMAD3
	TGFBR1
	TGFBR2
Arrhythmogenic right ventricular cardiomyopathy	DSC2
	DSG2
	DSP
	PKP2
	TMEM43
Catecholaminergic polymorphic ventricular tachycardia	CASQ2
	RYR2
	TRDN
Familial hypertrophic cardiomyopathy, dilated cardiomyopathy	ACTC1
	BAG3
	DES
	FLNC
	GLA
	LMNA
	MYBPC3
	MYH7
	MYL2
	MYL3
	PLN
	PRKAG2
	RBM20
	TNNC1
	TNNI3
	TNNT2
	TPM1
	TTN
Long QT syndrome, Brugada syndrome	KCNH2
	KCNQ1
	SCN5A
	CALM1
	CALM2
	CALM3
Ehlers-Danlos syndrome, vascular type	COL3A1
Familial hypercholesterolemia	APOB
	LDLR
	PCSK9

CONDITION	GENE
Genes related to cancer predisposition	
Adenomatous polyposis coli	APC
Hereditary breast and/or ovarian cancer	BRCA1*
	BRCA2*
	PALB2*
Juvenile polyposis syndrome	BMPR1A
	SMAD4
Lynch syndrome	MLH1*
	MSH2*
	MSH6*
	PMS2*
Multiple endocrine neoplasia type 1	MEN1
Multiple endocrine neoplasia, type 2 and Familial medullary thyroid carcinoma	RET
MUTYH-associated polyposis	MUTYH*
Li-Fraumeni syndrome	TP53
Neurofibromatosis, type 2	NF2
Paraganglioma-pheochromocytoma syndrome	MAX
	SDHAF2
	SDHB
	SDHC
	SDHD
	TMEM127
Peutz-Jeghers syndrome	STK11
PTEN hamartoma tumor syndrome	PTEN
Tuberous sclerosis	TSC1
	TSC2
Retinoblastoma	RB1
Von Hippel-Lindau syndrome	VHL
Wilms tumor	WT1
Genes related to inborn errors of metabolism	
Adrenal Leukodystrophy	ABCD1
Biotinitidase deficiency	BTD
Cerebrotendinous xanthomatosis	CYP27A1
Hereditary hemochromatosis	HFE* (C282Y only)
Ornithine carbamoyltransferase deficiency	OTC
Pompe disease	GAA
Genes related to other disease phenotypes	
Hereditary hemorrhagic telangiectasia	ACVRL1
	ENG
Hereditary TTR amyloidosis	TTR*
Malignant hyperthermia susceptibility	CACNA1S
	RYR1
Maturity-onset diabetes of the young	HNF1A
RPE65-related retinopathy	RPE65
Wilson disease	ATP7B

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