

# 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 81 *medically actionable* disease genes

#### 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 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](http://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
Biotinidase 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
Lynch syndrome	CALM3
	MLH1*
	MSH2*
	MSH6*
Malignant hyperthermia susceptibility	PMS2*
	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
Neurofibromatosis, type 2	NF2
Ornithine carbamoyltransferase deficiency	OTC
Paranglioma-pheochromocytoma syndrome	MAX
	SDHAF2
	SDHB
	SDHC
	SDHD
Peutz-Jeghers syndrome	TMEM127
	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

