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 59 *medically actionable* disease genes

What does *medically actionable* mean?

59 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 your child 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 child'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
Adenomatous polyposis coli	APC
Aortic aneurysm, familial thoracic	ACTA2
	MYH11
Arrhythmogenic right ventricular cardiomyopathy	DSC2
	DSG2
	DSP
	PKP2
	TMEM43
Catecholaminergic polymorphic ventricular tachycardia	RYR2
Ehlers-Danlos syndrome, vascular type	COL3A1
Familial hypercholesterolemia	APOB
	LDLR
	PCSK9
Familial hypertrophic cardiomyopathy, dilated cardiomyopathy	ACTC1
	GLA
	LMNA
	MYBPC3
	MYH7
	MYL2
	MYL3
	PRKAG2
	TNNI3
	TNNT2
	TPM1
Hereditary breast and ovarian cancer	BRCA1
	BRCA2
Juvenile polyposis syndrome	BMPR1A
	SMAD4
Li-Fraumeni syndrome	TP53

CONDITION	GENE
Loeys-Dietz syndrome	SMAD3
	TGFBR1
	TGFBR2
Long QT syndrome, Brugada syndrome	KCMH2
	KCNQ1
	SCN5A
Lynch syndrome	MLH1
	MSH2
	MSH6
	PMS2
Malignant hyperthermia susceptibility	CACNA1S
	RYR1
Marfan syndrome	FBN1
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	SDHAF2
	SDHB
	SDHC
	SDHD
Peutz-Jeghers syndrome	STK11
PTEN hamartoma tumor syndrome	PTEN
Retinoblastoma	RB1
Tuberous sclerosis	TSC1
	TSC2
Von Hippel-Lindau syndrome	VHL
Wilms tumor	WT1
Wilson disease	ATP7B