

Genome-wide Sequencing Ontario

Pediatric 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?

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 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.

Actionable in Childhood

All patients under 18 will automatically have analysis of 74 genes that are medically actionable in childhood

All Secondary Findings

You may **choose** whether **you** wish for your child 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
Genes related to cardiovascular disease predisposition	
Aortic aneurysm, familial thoracic	ACTA2 MYH11
Marfan syndrome	FBN1
Loeys-Dietz syndrome	SMAD3 TGFB1 TGFB2
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	BMPT1A SMAD4 MLH1* MSH2* MSH6* PMS2*
Lynch syndrome	MEN1
Multiple endocrine neoplasia type 1	RET
Multiple endocrine neoplasia, type 2 and Familial medullary thyroid carcinoma	MUTYH*
MUTYH-associated polyposis	TP53
Li-Fraumeni syndrome	NF2
Neurofibromatosis, type 2	MAX SDHAF2 SDHB SDHC SDHD TMEM127
Paraganglioma-pheochromocytoma syndrome	STK11
Peutz-Jeghers syndrome	PTEN
PTEN hamartoma tumor syndrome	TSC1 TSC2
Tuberous sclerosis	RB1
Retinoblastoma	VHL
Von Hippel-Lindau syndrome	WT1
Wilms tumor	
Genes related to inborn errors of metabolism	
Adrenal Leukodystrophy	ABCD1
Biotinidase deficiency	BTB
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