

Genome-wide Sequencing Ontario

Adult 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.

All Secondary Findings

The laboratory will search for disease-associated variants in 59 *medically actionable* disease genes

No Secondary Findings

The laboratory will **not** report any variants in disease genes that are not associated with your medical condition

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



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Secondary Findings List

CONDITION	GENE	CONDITION	GENE
Adenomatous polyposis coli	APC	Loeys-Dietz syndrome	SMAD3
Aortic aneurysm, familial thoracic	ACTA2		TGFBR1
	MYH11		TGFBR2
Arrhythmogenic right ventricular cardiomyopathy	DSC2	Long QT syndrome, Brugada syndrome	KCMH2
	DSG2		KCNQ1
	DSP		SCN5A
	PKP2	Lynch syndrome	MLH1
	TMEM43		MSH2
	MSH6		
Catecholaminergic polymorphic ventricular tachycardia	RYR2		PMS2
Ehlers-Danlos syndrome, vascular type	COL3A1	Malignant hyperthermia susceptibility	CACNA1S
Familial hypercholesterolemia	APOB		RYR1
	LDLR	Marfan syndrome	FBN1
	PCSK9		
Familial hypertrophic cardiomyopathy, dilated cardiomyopathy	ACTC1	Multiple endocrine neoplasia type 1	MEN1
	GLA	Multiple endocrine neoplasia, type 2 and Familial medullary thyroid carcinoma	RET
	LMNA		
	MYBPC3	MUTYH-associated polyposis	MUTYH
	MYH7	Neurofibromatosis, type 2	NF2
	MYL2	Ornithine carbamoyltransferase deficiency	OTC
	MYL3	Paraganglioma-pheochromocytoma syndrome	SDHAF2
	PRKAG2		SDHB
	TNNI3		SDHC
	TNNT2		SDHD
	TPM1	Peutz-Jeghers syndrome	STK11
Hereditary breast and ovarian cancer	BRCA1	PTEN hamartoma tumor syndrome	PTEN
	BRCA2	Retinoblastoma	RB1
Juvenile polyposis syndrome	BMPR1A	Tuberous sclerosis	TSC1
	SMAD4		TSC2
Li-Fraumeni syndrome	TP53	Von Hippel-Lindau syndrome	VHL
		Wilms tumor	WT1
		Wilson disease	ATP7B

