

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?

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.

Actionable in Childhood

All patients under 18 will automatically have analysis of 52 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 7 genes that are not medically actionable until adulthood



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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	FBN1	
	PCSK9	Marfan syndrome	FBN1
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	MUTYH-associated polyposis	MUTYH*
	MYBPC3	Neurofibromatosis, type 2	NF2
	MYH7	Ornithine carbamoyltransferase deficiency	OTC
	MYL2	Paraganglioma-pheochromocytoma syndrome	SDHAF2
	MYL3		SDHB
	PRKAG2		SDHC
	TNNI3		SDHD
	TNNT2	Peutz-Jeghers syndrome	STK11
TPM1	PTEN hamartoma tumor syndrome	PTEN	
Hereditary breast and ovarian cancer	BRCA1*	Retinoblastoma	RB1
	BRCA2*	Tuberous sclerosis	TSC1
Juvenile polyposis syndrome	BMPR1A		TSC2
	SMAD4	Von Hippel-Lindau syndrome	VHL
Li-Fraumeni syndrome	TP53	Wilms tumor	WT1
		Wilson disease	ATP7B

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

