# Genome-wide Sequencing Ontario

# Mature Minor 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 you 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 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 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 |
| | TGFBR1 |
| | TGFBR2 |
| 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 | BMPR1A |
| | SMAD4 |
| Lynch syndrome | MLH1* |
| | MSH2* |
| | MSH6* |
| | PMS2* |
| Multiple endocrine neoplasia type 1 | MEN1 |
| Multiple endocrine neoplasia, type 2 and Familial medullary thyroid carcinoma | RET |
| MUTYH-associated polyposis | MUTYH* |
| Li-Fraumeni syndrome | TP53 |
| Neurofibromatosis, type 2 | NF2 |
| Paraganglioma-pheochromocytoma syndrome | MAX |
| | SDHAF2 |
| | SDHB |
| | SDHC |
| | SDHD |
| | TMEM127 |
| Peutz-Jeghers syndrome | STK11 |
| PTEN hamartoma tumor syndrome | PTEN |
| Tuberous sclerosis | TSC1 |
| | TSC2 |
| Retinoblastoma | RB1 |
| Von Hippel-Lindau syndrome | VHL |
| Wilms tumor | WT1 |
| **Genes related to inborn errors of metabolism** | |
| Adrenal Leukodystrophy | ABCD1 |
| Biotinitidase deficiency | BTD |
| 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