

## **Molecular Diagnostic Testing**

Tests available	# of genes	Price (CAD)
Targets of Newborn Screening (Price is based on the number of genes tested)	<5	\$1000
range to or treatment of genes testedy	>=5	\$1,500
Severe Combined/Primary Immune Deficiency Panel	251	\$2,425
CYP21A2 Congenital Adrenal Hyperplasia (Sanger sequencing + MLPA)	1	\$1,500
Cystic Fibrosis Common Mutation Panel	1	\$75
Spinal Muscular Atrophy—ddPCR/MLPA	1	\$250
Mitochondrial Diseases: Full Nuclear Gene Panel		
Full Nuclear Gene Panel	437	\$2,425
Mitochondrial Encephalopathy/Leigh Disease	117	\$2,418
mtDNA Depletion and Deletion (Nuclear Gene Panel)	19	\$2,130
Progressive External Ophthalmoplegia (PEO)/Optic Atrophy	77	\$2 <i>,</i> 425
Pyruvate Dehydrogenase Complex Deficiency	16	\$2,130
Sanger Sequencing for Family Variants/ Cascade Testing	N/A	\$400 (+ \$100/ each additional variant)
Cytomegalovirus qPCR (out-of-province requests)	N/A	\$50
T-cell Receptor Excision Circles (TREC) Analysis (out-of-province requests)	N/A	\$50

Please contact us for availability and pricing for single gene sequencing of any gene on an offered panel, or for a gene not currently offered but relevant to the diseases for which we provide testing.

Last update: December 2025

