

Molecular Diagnostic Testing

Tests available	# of genes	Price (CAD)
Targets of Newborn Screening (Price is based on the number of genes tested)	<5 >=5	\$1000 \$1,500
	/-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
Mitochondrial Diseases: Full Nuclear Gene Panel		
Full Nuclear Gene Panel*	425	\$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	\$2,425
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

^{*}No additional cost for a custom subpanel or for release of exome data with research consent.

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: October 2022

