NEWBORN SCREENING ONTARIO
DÉPISTAGE NÉONATAL ONTARIO
MOLECULAR DIAGNOSTICS

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
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* Mitochondrial Encephalopathy/Leigh Disease* mtDNA Depletion and Deletion (*Nuclear Gene Panel*)* Progressive External Ophthalmoplegia (PEO)/Optic Atrophy* Pyruvate Dehydrogenase Complex Deficiency	 437 117 19 77 16	 $2,425 $2,418 $2,130 $2,425 $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

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.

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415 Smyth Road, Ottawa, ON K1H 8M8 · Phone 613-738-4209 · 1-877-NBS-8330 · Fax: 613-738-4214
www.newbornscreening.on.ca /en/diagnostic-testing · NSOmolecular@cheo.on.ca