



## Molecular Diagnostic Testing

| Tests available                                                                                                                                                                                                                                                                | # of genes                   | Price (CAD)                                         |
|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------------------------------|-----------------------------------------------------|
| <b>Targets of Newborn Screening</b> ( <i>Price is based on the number of genes tested</i> )                                                                                                                                                                                    | <5<br>≥5                     | \$1000<br>\$1,500                                   |
| <b>CYP21A2 Congenital Adrenal Hyperplasia</b> (Sanger sequencing +MLPA)<br>Reflex to Congenital Adrenal Hyperplasia "Other" panel                                                                                                                                              | 1<br>8                       | \$1,500<br>\$1,000                                  |
| <b>Cystic Fibrosis Common Mutation Panel</b>                                                                                                                                                                                                                                   | 1                            | \$75                                                |
| <b>Spinal Muscular Atrophy – ddPCR/MLPA</b>                                                                                                                                                                                                                                    | 1                            | \$250                                               |
| <b>Mitochondrial Diseases:</b><br>Full Nuclear Gene Panel<br>Mitochondrial Encephalopathy/Leigh Disease<br>mtDNA Depletion and Deletion ( <i>Nuclear Gene Panel</i> )<br>Progressive External Ophthalmoplegia (PEO)/Optic Atrophy<br>Pyruvate Dehydrogenase Complex Deficiency | 437<br>117<br>19<br>77<br>16 | \$2,425<br>\$2,418<br>\$2,130<br>\$2,425<br>\$2,130 |
| <b>Sanger Sequencing for Familial Variants/Cascade Testing</b>                                                                                                                                                                                                                 | N/A                          | \$400 (+ \$100/each additional variant)             |
| <b>Cytomegalovirus qPCR</b> (out-of-province requests)                                                                                                                                                                                                                         | N/A                          | \$50                                                |
| <b>T-cell Receptor Excision Circles (TREC) Analysis</b> (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: October 2026