

Mitochondrial Diagnostics Test List

Test Code	Test Name	Shipping Temp	CPT Code
L7087	Blue Native Page Electrophoresis with in-gel mitochondrial enzyme analysis – Muscle, Liver or Heart ¹	Frozen on Dry Ice	82664 82657x4
L7078	Blue Native Page Electrophoresis with in-gel mitochondrial enzyme analysis – Skin Fibroblasts ²	Ambient	82664 82657x4
	Glycine Cleavage Enzyme Assay – Liver Tissue ONLY ¹	Frozen on Dry Ice	82657 82658
L7086	Mitochondrial Respiratory Chain Enzyme – Muscle, Liver or Heart ¹	Frozen on Dry Ice	82657x6
L7079	Mitochondrial Respiratory Chain Enzyme – Skin Fibroblasts ²	Ambient	82657x6
L7085	Pyruvate Dehydrogenase Enzyme Assay – Skin Fibroblasts ²	Ambient	82657x1 82658x1

¹ Tissue Specimens must be immediately frozen on dry ice, stored at -70°C, and shipped frozen on dry ice. Minimum 60 mg muscle tissue required, 120 mg recommended.

² An additional fee of \$460 will be charged for skin specimens that have not been cultured into confluent fibroblasts before shipment.

Turnaround Times (TAT)

Turnaround times are based on a Monday – Friday schedule.

If test results are delayed for longer than specified turnaround time, the requesting physician will be notified of the delay.

Test Name	TAT
Glycine Cleavage Assay	4 weeks
Pyruvate Dehydrogenase Enzyme Assay	90 days*
Respiratory Chain Enzyme Assay – Muscle, Liver or Heart	4-6 weeks
Respiratory Chain Enzyme Assay – Skin Fibroblasts	90 days*
Quantitative Stable Isotope Dilution	1-5 days
Acylcarnitine Profile	1-3 days
Urine/CSF succinylpurines	1-10 days
Urine Trimethylamine (TMA)	3 weeks
Enzyme assays for GAI and GAI1	90 days
Enzyme assay for VLCADD	7 days
HVA and VMA	1-3 days

* Fibroblast turnaround time depends on the growth of the fibroblasts