Test Profiles & Gene Sequence Analysis

Test	Price	CPT code
Glycogen Storage Disease Profile ● Acid and Neutral Maltase ● Myophosphorylase ● Phosphorylase b kinase ● Phosphofructokinase Tissue: muscle	$626	82657 x 5
Mitochondrial Myopathy Profile ● NADH dehydrogenase ● NADH cytochrome c reductase ● Succinate dehydrogenase ● Succinate cytochrome c reductase ● Cytochrome c oxidase ● Citrate synthase Tissue: muscle, liver	$675	82657 x 6
Modified Mitochondrial Myopathy Profile ● Succinate dehydrogenase ● Cytochrome c oxidase ● Citrate synthase Tissue: muscle, liver	$330	82657 x 3
Myoglobinuria Profile ● Myophosphorylase ● Phosphorylase b kinase ● Phosphofructokinase ● Phosphoglycerate kinase ● Phosphoglycerate mutase ● Lactate dehydrogenase ● Carnitine palmitoyltransferase ● Citrate synthase Tissue: muscle	$1100	82657 x 8, 82658

Exercise Intolerance Mutation Profile (*CPT2*, *PYGM*, *AMPD1* genes) (8 mutations)	$579	81401, 81479 x 2
Exercise Intolerance Mutation Profile + Expanded *CPT2* Gene Analysis (10 mutations)	$660	81401, 81479 x 2
Carnitine Palmitoyltransferase (*CPT2* gene) Mutation Profile (6 mutations)	$479	81479
Myophosphorylase [McArdle Disease; GSD V] (*PYGM* gene) Mutation Profile (2 mutations)	$253	81401
Individual Mutation Analysis*	$230	81401
Gene Sequence Analysis		
Carnitine Palmitoyltransferase II Deficiency (*CPT2* gene)	$715	81404
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (*ACAVDL* gene)	$924	81406
Caveolin-3 Deficiency; Rippling Muscle Disease; Limb Girdle Muscular Dystrophy type 1C; HyperCKemia; Distal Myopathy (*CAV3* gene)	$424	81404
Myophosphorylase Deficiency; McArdle Disease (*PYGM* gene) Level 1: Exons 1 & 5	$374	81479
Myophosphorylase Deficiency; McArdle Disease (*PYGM* gene) Level 2: Exons 2, 3, 4, 6-20	$743	81406
Myophosphorylase Deficiency; McArdle Disease (*PYGM* gene) Level 1 and 2: Exons 1-20	$1073	81406
Malignant Hyperthermia [additional indications: exertional rhabdomyolysis; statin myopathy; hyperCKemia; heat stroke] (*RYR1* gene – 23 exons, *CACNA1S* gene – 3 exons)**	$1150	81408, 81479
RYR1* gene – 23 exons*	$820	81408
CACNA1S* gene – 3 exons*	$330	81479
Familial Studies for Malignant Hyperthermia: *RYR1*		
● one exon	$125	81403
● two exons	$175	81403
● three exons	$225	81403

Familial Studies for Malignant Hyperthermia: *CACNA1S*		
● one exon	$125	81479
● two exons	$175	81479
● three exons	$225	81479

*Any single mutation analysis may be selected from the above profiles.

***RYR1* exons 2, 6-12, 14-17, 39-41, 43-47, 100-102; *CACNA1S* exons 4, 5, 26