NEW Solutions for your Complex Mitochondrial and Metabolic Testing Needs

The Medical Genetics Laboratories at Baylor College of Medicine have been providing the medical genetics community with high quality comprehensive diagnostic services for over 40 years. By building on our institution's strengths in research and discovery, we are committed to providing quality genetic testing services relevant to twenty-first century medicine.

Three reasons to use BCM's Medical Genetics Laboratories:

1. ***On time delivery*** *of test results including panel testing by Next Generation Sequencing. The last six months,* ***99%*** *of the Next Generation Sequencing tests were reported out on time.*
2. ***Lower cost*** *and time to diagnosis by using pathway driven gene panels.*
3. ***Industry leading expertise*** *in clinical and diagnostic interpretation.*

New panels and single gene analysis tests are now available:

Next Generation Sequencing Panels	Test Code
Cholestasis Panel (4 genes)	2105
Cobalamin Metabolism Panel (8 genes)	2120
Coenzyme Q10 Deficiency Panel (5 genes)	2100
Glycogen Metabolism Disorders (16 genes)	2125
Glycogen Metabolism Disorders Muscle form (9 genes)	2126
Glycogen Metabolism Disorders Liver form (10 genes)	2127
Mitochondrial Depletion and Integrity Panel (14 genes)	2130
Mitochondrial or Metabolic Myopathy/Rhabdomyosis (25 genes)	2300
Mitochondrial Respiratory Chain Complex I Panel (25 genes)	2155
Mitochondrial Respiratory Chain Complex II Panel (6 genes)	2160
Mitochondrial Respiratory Chain Complex III Panel (4 genes)	2165
Mitochondrial Respiratory Chain Complex IV Panel (12 genes)	2170
Mitochondrial Respiratory Chain Complex V Panel (3 genes)	2175
Mitochondrial Respiratory Chain Complex I – V Panel (50 genes)	2180
PDH & Mitochondrial Respiratory Chain Complex V Panel (9 genes)	2185
Mitochondrial Whole Genome Sequencing Analysis	2055
Progressive External Ophthalmoplegia Panel (6 genes)	2140

Sanger Sequencing Panels	Test Code
Methylmalonic Acidemia Sequencing Panel (*MMAA, MMAB,* and *MUT* genes)	3579
Single Gene Analysis	**Test Code**
3-Methylglutaconic Aciduria Type I - *AUH* Sequence Analysis	3910
Alagille Syndrome - *JAG1* Sequence Analysis	3755
Glycogen Storage Disease Type II - *GAA* Sequence Analysis	3400
Glycogen Storage Disease Type XI - *LDHA* Sequencing Analysis	3785
Glycogen Storage Disease Type XII - *ALDOA* Sequence Analysis	2515
Glycogen Storage Disease Type XIII - *ENO3* Sequence Analysis	2525
Glycogen Storage Disease Type XIV - *PGM1* Sequence Analysis	2520
Leukoencephalopathy (LBSL) - *DARS2* Sequence Analysis	3715
LMBR1 Domain-Containing Protein 1 - *LMBRD1* Sequence Analysis	2560
Menkes Disease - *ATP7A* Sequence Analysis	2545
Methionine Synthase Reductase - *MTRR* Sequence Analysis	2565
Nuclear Encoded complex I deficiency - *C20orf7* Sequence Analysis	2655
Nuclear Encoded complex I deficiency - *FOXRED1* Sequence Analysis	2660
Nuclear Encoded complex I deficiency - *NDUFA2* Sequence Analysis	2665
Nuclear Encoded complex I deficiency - *NDUFA8* Sequence Analysis	2670
Nuclear Encoded complex I deficiency - *NDUFA10* Sequence Analysis	2675
Nuclear Encoded complex I deficiency - *NDUFA11* Sequence Analysis	2680
Nuclear Encoded complex I deficiency - *NDUFA13* Sequence Analysis	2685
Nuclear Encoded complex I deficiency - *NDUFAF3* Sequence Analysis	2690
Nuclear Encoded complex I deficiency - *NDUFB6* Sequence Analysis	2500
Nuclear Encoded complex I deficiency - *NDUFS1* Sequence Analysis	2700
Nuclear Encoded complex I deficiency - *NDUFV3* Sequence Analysis	2705
Nuclear Encoded complex I deficiency - *NUBPL* Sequence Analysis	2710
Nuclear Encoded complex III Deficiency - *TTC19* Sequence Analysis	2715
Nuclear Encoded complex III Deficiency - *UQCRB* Sequence Analysis	2720
Nuclear Encoded complex III Deficiency - *UQCRQ* Sequence Analysis	2725
Nuclear Encoded complex IV deficiency - *COX4I1* Sequence Analysis	2730
Nuclear Encoded complex IV deficiency - *COX4I2* Sequence Analysis	2735
Nuclear Encoded complex IV deficiency - *COX7A1* Sequence Analysis	2740
Nuclear Encoded complex IV deficiency - *TACO1* Sequence Analysis	2745
Nuclear Encoded complex IV deficiency - *ATP5E* Sequence Analysis	3290
Nuclear Encoded complex V deficiency - *ATPAF2* Sequence Analysis	3270
Mitochondrial Myopathy and Sideroblastic Anemia Type 2 - *YARS2* Sequence Analysis	3755
Molybdenum Cofactor Deficiency - *MOCS2* Sequence Analysis	3615
Pontocerebellar Hypoplasia - *LARS2* Sequence Analysis	2535
Pontocerebellar Hypoplasia Type 6 - *RARS2* Sequence Analysis	3725
Pyruvate Dehydrogenase E1-Beta Deficiency - *PDHB* Sequence Analysis	3895
Pyruvate Dehydrogenase E2 Deficiency - *DLAT* Sequence Analysis	3915
Pyruvate Dehydrogenase E3-Binding Protein Deficiency - *PDHX* Sequence Analysis	3920
Pyruvate Dehydrogenase Phosphatase Deficiency - *PDP1* Sequence Analysis	3890
Wilson Disease - *ATP7B* Sequence Analysis	2550

Now offering Comprehensive testing (Sequencing & Deletion/Duplication analysis) and Individual Sequencing or Targeted Deletion/Duplication analysis for the following genes:

Disorders	Comprehensive Analysis (Seq & Del/Dup)	Seq Analysis	Del/Dup Analysis
ABCB11 Related Disorders	3314	3310	3313
ABCB4 Related Disorders	3319	3315	3318
ALDOB Related Disorders	3129	3125	3128
ATP8B1 Related Disorders	3309	3305	3308
C10orf2 Related Disorders	3179	3175	3178
CPT2 Related Disorders	3164	3160	3163
DGUOK Related Disorders	3079	3075	3078
MPV17 Related Disorders	3324	3320	3323
OTC Related Disorders	3144	3140	3143
PDHA1 Related Disorders	3169	3165	3168
POLG Related Disorders	3069	3065	3068
SLC22A5 Related Disorders	3364	3360	3363
SUCLA2 Related Disorders	3379	3375	3378
TK2 Related Disorders	2180	3070	3073

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