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NEUROCHEMISTRY AND METABOLIC TEST REQUEST FORM

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GENETIC TESTING GIMETOT) CSF AMINO ACIDS [METOT) CSF LACTATE [METOT) CSF LACTATE [METOT) CSF PERUVATE [MC01) CSF PERUVATE [MC01) CSF PERUVATE [MC02) CSF PERUVATE [MC02) CSF NEMERIVITERING NOR SIMMUNE SYSTEM STIMULATION] [MC02) CSF NEMERIVITERING NOR SIMMUNE SYSTEM STIMULATION] [MC02) CSF NEMERIVITERING NOR SIMMUNE SYSTEM STIMULATION] [MC02) CSF TERRANDROBIODER TERRONS NOR SIMMUNE SYSTEM STIMULATION] [MC02) CSF TERRANDROBIODER SERIOR SYSTEM STIMULATION] [MC02) CSF TERRANDROBIODER SERIOR SEQUENCY [MC02) CSF TERRANDROBIODER SERIOR SEQUENCY [MC02) CSF TERRANDROBIODER SERIOR SEQUENCY [MC03) CSF PERIVOLAS 5-PROSPHATE [PYRIDOXIA] [ME PROSPHATE [MC07) CSF SAMIC ADD [DISORGES WITH HYPOMENIATE DEFICIENCY] [MC07) CSF SAMIC ADD [DISORGES WITH HYPOMENIATION OF UNKNOWN ETOLOGY (SALUC ADD [SUCCESS WITH HYPOMENIATI	PATIENT NAME:	DOB:		
Common C				
(MET1) (SF AMINO ACIDS (MET1) (SF PYRIUVATE (MET1) (SF PYRIUVA				
(MET10) CSF PARUATE				
(MC11) CSF PYRUVATE				
(NCO1) CSF 5-METHYLTETRAHYDROFOLATE	, ,		, or ji	
(NC02) CSF Neurotransmitter Metabolism; 93 ceies			AES: 6 GENES	
(NC03) CSF TETRALYDROBIOPTERIN AND NEOPTERIN PROFILE (NC04) CSF NEUROTRANSMITTER METABOLITES SHIAA, HVA, 30MD (INCLUDES BIOMARKERS FOR PYRIDDXINE RESPONSIVE SEIZURES) (INC05) CSF PYRIDDXIA.5 "-PHOSPHATE PYRIDOX[AM]INE PHOSPHATE CIVIDADE SHOWARKERS FOR PYRIDDXIA.5 "-PHOSPHATE PROXIDAL SHOP PRIDDXIAD SHOP PRIDX SHOP PRIDX SHOP PRIDX SHOP PR				
(NCO4) CSF NEUROTRANSMITTER METABOLITES 5HIAA, HVA, 30MD (INCUDES BIOMARKERS FOR PYRIDOXINE RESPONSIVE SEIZURES) (INCO5) CSF PYRIDOXAL 5'-PHOSPHATE [PYRIDOX[AM]INE PHOSPHATE OXIDASE DEFICIENCY + CNS PYRIDOXAL 5'-PHOSPHATE DEFICIENCY] (INCO5) CSF PYRIDOXAL 5'-PHOSPHATE DEFICIENCY] (INCO5) CSF SUCILIVIA DEMONSINE [ADENTVO DEMONSICAL 5'-PHOSPHATE DEFICIENCY] (INCO5) CSF SUCILIVIA DEMONSINE [ADENTVO DEMONSICAL 5'-PHOSPHATE DEFICIENCY] (INCO5) CSF SUCILIVIA DEMONSINE [ADENTVO DEMONSICAL 5'-PHOSPHATE DEFICIENCY] (INCO5) CSF SUCINIVIA DEMONSINE [ADENTVO DEMONSICAL 5'-PHOSPHATE DEFICIENCY] (INCO5) CSF SUCINIVIA DEMONSINE [ADENTVO DEMONSINE] (INCO5) CSF SUCINIVIA DEMONSINE [ADENTVO DEMONSINE] (INCO5) CSF ALPHA-AMINOADIPIC SEMIALDEHYDE [PYRIDOXINE-RESPONSIVE SEIZURES] (INCO5) CSF ALPHA-AMINOADIPIC SEMIALDEHYDE DEHYDROCGENASE DEFICIENCY] (INCO1) CSF GLUCOSE (GLUCOSE TRANSPORTER DEFICIENCY) (INCO1) CSF GLUCOSE (GLUCOSE (GLUCOSE TRANSPORTER DEFICIENCY) (INCO1) CALPART (INCO1) CALP				
(NC05) CSF PYRIDOXAL 5'-PHOSPHATE [PYRIDOX[AM]NINE PHOSPHATE OXIDASE DEFICIENCY + CNS PYRIDOXAL 5'-PHOSPHATE DEFICIENCY] (NC05) CSF PYRIDOXAL 5'-PHOSPHATE DEFICIENCY] (NC06) CSF SUCCINYLADENOSINE [ADENYLOSUCCINATE LYASE DEFICIENCY] (NC07) CSF SIALIC ACID [DISORDERS WITH HYPOMYELIANTION OF UNKNOWN ETIOLOGY SIALIC ACID STORAGE DISORDERS] (NC08) CSF ALPHA-AMINOADIPIC SEMIALDEHYDE [PYRIDOXINE-RESPONSIVE SEIZURES] (MC0.38) ADAR (MC0.306) ARRIGEF9 (MC0.38) CSF ALPHA-AMINOADIPIC SEMIALDEHYDE [PYRIDOXINE-RESPONSIVE SEIZURES] (MC0.237) DHFR (MC0.236) DDC (
□ (NC05) CSF PYRIDOXAL 5'-PHOSPHATE [PYRIDOX,M]IMP PHOSPHATE ○ (NC05) CSF PYRIDOXAL 5'-PHOSPHATE DEFICIENCY] ○ (NC05) CSF SUCCINYLADENOSINE [ADENYLOSUCCINATE LYASE DEFICIENCY] ○ (NC05) CSF SUCCINYLADENOSINE [ADENYLOSUCCINATE LYASE DEFICIENCY] ○ (NC05) CSF SIALLO ACID [DISORDERS WITH HYPOMYELINATION OF UNKNOWN ETIOLOGY (SIALLO ACID [DISORDERS WITH HYPOMYELINATION OF UNKNOWN ETIOLOGY (SIALLO ACID STORAGE DISORDERS) ○ (NC08) CSF ALPHA-AMINOADIPIC SEMIALDEHYDE [PYRIDOXINE-RESPONSIVE SEIZURES] ○ (MC003) ALDHAA1 ○ (MOL125) ALDHAA1 ○ (NC08) CSF ALPHA-AMINOADIPIC SEMIALDEHYDE [PYRIDOXINE-RESPONSIVE SEIZURES] ○ (MOL141) DBH ○ (MOL025) DDC ○ (NC09) CSF 4-HYDROXYBUTYRIC ACID [SUCCINIC SEMIALDEHYDE DEHYDROGENASE DEFICIENCY] ○ (MOL141) DBH ○ (MOL166) FOLR1 ○ (NET00) CSF GLUCOSE (GLUCOSE TRANSPORTER DEFICIENCY) ○ (MOL237) DHFR ○ (MOL237) DHFR ○ (MOL166) FOLR1 ○ (MET010) CSF GLUCOSE (GLUCOSE TRANSPORTER DEFICIENCY) ○ (MOL234) GCH1+MLPA [DEL/DUP] ○ (MOL234) GCH1+MLPA [DEL/DUP] ○ (MOL234) GCH1+MLPA [DEL/DUP] ○ (MOL234) GCH1+MLPA [DEL/DUP] ○ (MOL236) GCH1 ○ (MOL236) GCH1 ○ (MOL236) GCH1 ○ (MOL236) GCH1 ○ (MOL236) GCH1+MLPA [DEL/DUP] ○ (MOL236) GCH1-MLPA [DEL/DUP] ○ (MOL236) GCH1-MLPA [DEL/DUP] ○ (MOL236) GCH1-MLPA [DEL/DUP] ○ (MOL237) DCH2-M		• •		
OXIDASE DEFICIENCY + CNS PYRIDOXAL 5'-PHOSPHATE DEFICIENCY] (NCO1) CSF SUCCINYLADENOSINE [ADENYLOSUCCINATE LYASE DEFICIENCY] (NCO1) CSF SIALIC ACID [DISORDERS WITH HYPOMYELINATION OF UNKNOWN ETIOLOGY /SIALIC ACID [SIORDERS] (NCO8) CSF ALPHA-AMINOADIPIC SEMIALDEHYDE [PYRIDOXINE-RESPONSIVE SIZURES] (NCO9) CSF ALPHA-AMINOADIPIC SEMIALDEHYDE [PYRIDOXINE-RESPONSIVE SIZURES] (NCO9) CSF ALPHA-AMINOADIPIC SEMIALDEHYDE [PYRIDOXINE-RESPONSIVE SIZURES] (NCO1) CSF GLUCOSE (GLUCOSE TRANSPORTER DEFICIENCY) (NC10) CSF GLUCOSE (GLUCOSE TRANSPORTER DEFICIENCY) (NC10) CSF GLUCOSE (GLUCOSE TRANSPORTER DEFICIENCY) (METO2) ANIMO ACIDS [PLASMA] (MOL234) GCH1+MLPA [DEL/Dup] (MOL213) GLDC MILPA [DEL/Dup] (MOL223) GLDC MILP				
NC06 CSF Succinvladenosine [Adenylosuccinate Lyase deficiency] (NC07) CSF Sulcic Acid [Disorders with Hyponytelination of Unknown Etiology / Sialic Acid [Disorders with Hyponytelination of Unknown Etiology / Sialic Acid [Disorders with Hyponytelination of Unknown Etiology / Sialic Acid [Disorders with Hyponytelination of Unknown Etiology / Sialic Acid [Disorders with Hyponytelination of Unknown Etiology / Sialic Acid [Disorders with Hyponytelination of Unknown Etiology / Sialic Acid [Disorders with Hyponytelination of Unknown Etiology / Sialic Acid [Disorders with Hyponytelination of Unknown Etiology / Sialic Acid [Disorders with Hyponytelination of Unknown Etiology / Sialic Acid [Disorders with Hyponytelination of Unknown Etiology (MOL39) ADAR (MOL141) DBH (MOL123) ACID (MOL237) DHFR (MOL237) DHFR (MOL237) DHFR (MOL233) CEH1+MLPA [Det/Dup] (MOL234) CEH1+MLPA [Det/Dup] (MOL234) CEH1+MLPA [Det/Dup] (MOL236) GLD (HMLPA [Det/Dup] (MOL346) GLD (HMLPA				
(MC07) CSF SALIC ACID DISGORDERS WITH HYPOMYELINATION OF UNKNOWN ETIOLOSY /SALIC ACID STORAGE DISGORDERS] (NC08) CSF ALPHA-AMINOADIPIC SEMIALDEHYDE [PYRIDOXINE-RESPONSIVE SEZURES] (MC01) CSF ALPHA-AMINOADIPIC SEMIALDEHYDE				
ETIOLOGY /SIALIC ACID STORAGE DISORDERS] (NC08) CSF ALPHA-AMINOADIPIC SEMIALDEHYDE [PYRIDOXINE-RESPONSIVE SEIZURES] (NC09) CSF 4-HYDROXYBUTYRIC ACID [SUCCINIC SEMIALDEHYDE DEHYDROGENASE DEFICIENCY] (NC109) CSF 6 CLUCOSE (GLUCOSE (GLUCOSE TRANSPORTER DEFICIENCY) (NC10) CSF 6 CLUCOSE (GLUCOSE TRANSPORTER DEFICIENCY) (MET02) AMINO ACIDS [PLASMA] (MET02) AMINO ACIDS [PLASMA] (MET03) CORNYME Q10 LEVEL [LEUKOCYTES] (MET08) LACTATE [PLASMA] (MET09) PHENYLALANINE LOADING ASSAY [PLASMA] (MET109) PYRUVATE' [BLOOD] (MET09) PHENYLALANINE LOADING ASSAY [PLASMA] (MET12) THYMIDINE AND DEOXYURIDINE [PLASMA] (MET12) THYMIDINE AND DEOXYURIDINE [PLASMA] (MET12) CREATINE & GUANIDINOACETATE [PLASMA] (MET12) CREATINE & GUANIDINOACETATE [PLASMA] (MET13) AMINO ACIDS [URINE] (MET03) AMINO ACIDS [URINE] (MET03) ALPHA-AMINOADIPIC SEMIALDEHYDE [URINE; FOR PYRIDOXINE-RESPONSIVE SEIZURES] (MET120) ALPHA-AMINOADIPIC SEMIALDEHYDE [URINE; FOR PYRIDOXINE-RESPONSIVE SEIZURES] (MC120) AROMATIC L-AMINO ACID DECARBOXYLASE [AADC] ENZYMOLOGY [PLASMA] (MET20) AROMATIC L-AMINO ACID DECARBOXYLASE [AADC] ENZYMOLOGY [PLASMA] (MC120) AROMATIC L-AMINO ACID DECARBOXYLASE [AADC] ENZYMOLOGY [PLASMA] (MC120) AROMATIC L-AMINO ACID DECARBOXYLASE [ADC] ENZYMOLOGY [PLASMA] (MC127) AROMATIC L-AMINO ACID DECARBOXYLASE [ADC] ENZYMOLOGY [PLASMA] (MC127) AROMATIC L-AMINO ACID DECARBOXYLASE [ADC] ENZYMOLOGY [PLASMA] (MC127) AROMATIC R-AMINO ACID DECARBO			☐ (MOL 125) AL DH5A1	
(NC08) CSF ALPHA-AMINOADIPIC SEMIALDEHYDE [PYRIDOXINE-RESPONSIVE SEIZURES] (MOL1341) DBH				
SETURES (NC09) CSF 4-HYDROXYBUTYRIC ACID [SUCCINIC SEMIALDEHYDE DEHYDROGENASE DEFICIENCY] (NC10) CSF GLUCOSE (GLUCOSE TRANSPORTER DEFICIENCY) (NC10) CSF GLUCOSE (GLUCOSE TRANSPORTER DEFICIENCY) (MET02) AMINO ACIDS [PLASMA] (MET02) AMINO ACIDS [PLASMA] (MET05) COENZYME Q10 LEVEL [LEUCKOCYTES] (MET05) COENZYME Q10 LEVEL [MUSCLE] (MET06) LACTATE [PLASMA] (MET10) PYRIVALANINIE LOADING ASSAY [PLASMA] (MET10) PYRIVATE* [BLOOD] (MET10) PYRIVATE* [BLOOD] (MET11) PYRIVATE* [BLOOD] (MET12) HYMIDINE AND DEOXYURIDINE [PLASMA] (MET12) HYMIDINE AND DEOXYURIDINE [PLASMA] (MET24) GLUCOSE [PLASMA] (MET24) GLUCOSE [PLASMA] (MET24) GLUCOSE [PLASMA] (MET03) AMINO ACIDS [URINE] (MET19) CREATINE & GUANIDINOACETATE [URINE] (MET19) ALPHA-AMINOADIPIC SEMIALDEHYDE [URINE; FOR PYRIDOXINE- RESPONSIVE SEIZURES] (MET24) AROMATIC L-AMINO ACID DECARBOXYLASE [AADC] ENZYMOLOGY (PLASMA] (MC117) SLC6A3 (MOL127) SLC6A3 (MOL128) SLC18A2 (MOL197) SLC6A3 (MOL125) SLC18A2 (MOL197) TREXT (MOL197) TREXT (MOL197) TREXT (MOL197) TREXT (MOL197) TREXT (MOL197) TREXT (MOL177) AROMATIC L-AMINO ACID DECARBOXYLASE [AADC] ENZYMOLOGY (WHOLE BLOOD ACD] (ENZOS) THYMIDINE PHOSPHORYLASE ENZYMOLOGY (WHOLE BLOOD ACD] (MOL177) AROMATIC L-AMINO ACID DECARBOXYLASE ENZYMOLOGY (WHOLE BLOOD ACD] (MOL177) AROMATIC L-AMINO ACID DECARBOXYLASE ENZYMOLOGY (WHOLE BLOOD ACD] (MOL177) AROMATIC L-AMINO ACID DECARBOXYLASE ENZYMOLOGY (WHOLE BLOOD ACD] (MOL177) AROMATIC L-AMINO ACID DECARBOXYLASE ENZYMOLOGY (WHOLE BLOOD ACD] (MOL177) AROMATIC L-AMINO ACID DECARBOXYLASE ENZYMOLOGY (WHOLE BLOOD ACD] (MOL177) AROMATIC L-AMINO ACID DECARBOXYLASE (GENES - ADAR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREXT) (MOL177) AROMATIC L-AMINO ACID DECARBOXYLASE (GENES - ADAR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREXT)	<u> </u>			
(NC09) ČSF 4-HYDROXYBUTYRIC ACID [SUCCINIC SEMIALDEHYDE DEHYDROGENASE DEFICIENCY]				
DEHYDROGENASE DEFICIENCY] (NC10) CSF GLUCOSE (GLUCOSE TRANSPORTER DEFICIENCY) (MC10) CSF GLUCOSE (GLUCOSE TRANSPORTER DEFICIENCY) (MET03) AMINO ACIDS [PLASMA] (MET04) COENZYME Q10 LEVEL [LEUOKOCYTES] (MET05) COENZYME Q10 LEVEL [MUSCLE] (MET08) LACTATE [PLASMA] (MET08) PLENYLALANINE LOADING ASSAY [PLASMA] (MET09) PRENYLALANINE LOADING ASSAY [PLASMA] (MET10) PYRUVATE* [BLOOD] (MET10) PYRUVATE* [BLOOD] (MET12) THYMIDINE AND DEOXYURIDINE [PLASMA] (MET12) THYMIDINE AND DEOXYURIDINE [PLASMA] (MET24) GLUCOSE [PLASMA] (MET24) GLUCOSE [PLASMA] (MET24) GLUCOSE [PLASMA] (MET03) AMINO ACIDS [URINE] (MET03) AMINO ACIDS [URINE] (MET109) CREATINE & GUANIDINOACETATE [URINE] (MET109) CREATINE & GUANIDINOACETATE [URINE] (MET109) ALPHA-AMINOADIPIC SEMIALDEHYDE [URINE; FOR PYRIDOXINE-RESPONSIVE SEIZURES] (MOL127) SLC6A3 (MOL127) SLC6A5 (MOL127) SLC6A5 (MOL127) SLC6A5 (MOL128) SLC1A3 (MOL127) SLC6A5 (MOL127) SLC6A5		□ (MOLZ31) DIII K	LI (MOL 100) I OLIVI	
(NC10) CSF GLUCOSE (GLUCOSE TRANSPORTER DEFICIENCY) (MOL234) GCH1+MLPA [DEL/DuP]		+□ (MOL060) GCH1		
CMCT215 GCH1 MLPA [DEL/DUP ONLY]				
(MET02) AMINO ACIDS [PLASMA]			ur v1	
☐ (MET04) COENZYME Q10 LEVEL [LEUOKOCYTES] ☐ (MOL236) GLDC+MLPA [DEL/DUP] ☐ (MET05) COENZYME Q10 LEVEL [MUSCLE] ☐ (MOL219) GLDC MLPA [DEL/DUP ONLY] ☐ (MET08) PHENYLALANINE LOADING ASSAY [PLASMA] ☐ (MOL116) GLRA1 ☐ (MOL120) GLRB ☐ (MET10) PYRUVATE* [BLOOD] ☐ (MOL251) GPHN ☐ (MOL171) MTHFR ☐ (MET23) CREATINE & GUANIDINOACETATE [PLASMA] ☐ (MOL026) PC ☐ (MOL349) PAH ☐ (MET24) GLUCOSE [PLASMA] ☐ (MOL074) PNPO ☐ (MOL0247) PDXK ☐ (MET24) GLUCOSE [PLASMA] ☐ (MOL171) QDPR ☐ (MOL124) RNASEH2A ☐ (MET103) AMINO ACIDS [URINE] ☐ (MOL172) SAMHD1 ☐ (MOL124) RNASEH2A ☐ (MET14) ORGANIC ACIDS [URINE] ☐ (MOL172) SAMHD1 ☐ (MOL124) RNASEH2C ☐ (MET19) CREATINE & GUANIDINOACETATE [URINE] ☐ (MOL095) SLC18A2 ☐ (MOL095) SLC18A2 ☐ (MET20) ALPHA-AMINOADIPIC SEMIALDEHYDE [URINE; FOR PYRIDOXINE-RESPONSIVE SEIZURES] ☐ (MOL127) SLC6A5 ☐ (MOL127) SLC6A4 ☐ (MOL127) SLC6A5 ☐ (MOL126) SPR ☐ (MOL199) THEX ☐ (MOL199) TREX1 GENEZYMOLOGY BLOOD ☐ (MOL181) AICARDI-GOUTIERES (6 GENES - ADAR, RNASEH2A, RNASEH2A, RNASEH2C, SAMHD1, TREX1) ☐ (MOL279) AICARDI-GOUTIERES (6 GENES) + MLPA [DEL/DUP]			421]	
(MET05) COENZYME Q10 LEVEL [MUSCLE] (MET08) LACTATE [PLASMA] (MET09) PHENYLALANINE LOADING ASSAY [PLASMA] (MOL116) GLRA1 (MOL120) GLRB (MET10) PYRUVATE* [BLOOD] (MOL251) GPHN (MOL251) GPHN (MOL171) MTHFR (MET12) THYMIDINE AND DEOXYURIDINE [PLASMA] (MOL266) PC (MOL247) PDXK (MET23) CREATINE & GUANIDINOACETATE [PLASMA] (MOL074) PNPO (MOL247) PDXK (MET24) GLUCOSE [PLASMA] (MOL074) PNPO (MOL096) PTS (MOL117) QDPR (MOL096) PTS (MOL117) QDPR (MOL124) RNASEH2A (MOL172) SAMHD1 (MOL168) RNASEH2C (MOL172) SAMHD1 (MOL168) RNASEH2C (MOL172) SAMHD1 (MOL095) SLC18A2 (MOL095) SLC18A2 (MOL097) SLC6A3 (MOL095) SLC18A2 (MOL097) SLC6A3 (MOL0252) SLC18A2 (MOL097) SLC6A3 (MOL0252) SLC1A3 (MOL097) THE (MOL097) THE (MOL097) THE (MOL097) THE (MOL097) THE (MOL099) TPH2 (MOL09				
(MET08) Lactate [PLasma]			ui v1	
(MET09) Phenylalanine Loading Assay [Plasma] (MOL116) GLRA1 (MOL120) GLRB (MET10) Pyruvate* [Blood] (MOL171) MTHFR (MOL251) GPHN (MOL171) MTHFR (MOL349) PAH (MCT23) CREATINE & GUANIDINOACETATE [PLASMA] (MOL226) PC (MOL349) PAH (MOL274) GLUCOSE [PLASMA] (MOL096) PTS (MOL096) PTS (MOL171) QDPR (MOL096) PTS (MOL171) QDPR (MOL096) PTS (MOL172) SAMHO ACIDS [URINE] (MOL172) SAMHOD1 (MOL188) RNASEH2A (MOL172) SAMHOD1 (MOL168) RNASEH2C (MOL172) SAMHOD1 (MOL095) SLC18A2 (MOL095) SLC18A2 (MOL095) SLC18A2 (MOL097) SLC6A3 (MOL228) SLC1A3 (MOL228) SLC1A3 (MOL127) SLC6A5 (MOL091) TH (MOL092) TPH2 (MOL091) TH (MOL092) TPH2 (MOL092) TPH2 (MOL092) TPH2 (MOL091) TREX1 (ENZ06) Thymidine Phosphorylase Enzymology [Whole Blood ACD] (MOL279) Alcardi-Goutieres (6 Genes) + MLPA [Det/Dup]		(MOLZ 19) OLDO MEI A [DEL/DOF OF	vL1]	
(MET10) PYRUVATE* [BLOOD]		☐ (MOI 116) GI RA1	☐ (MOL 120) GLRB	
(MET12) THYMIDINE AND DECXYURIDINE [PLASMA] (MOL226) PC (MOL349) PAH (MET23) CREATINE & GUANIDINOACETATE [PLASMA] (MOL074) PNPO (MOL247) PDXK (MOL117) QDPR (MOL096) PTS (MOL117) QDPR (MOL118) RNASEH2B (MOL124) RNASEH2A (MOL172) SAMHD1 (MOL124) RNASEH2A (MOL172) SAMHD1 (MOL168) RNASEH2C (MOL172) SAMHD1 (MOL095) SLC18A2 (MOL095) SLC18A2 (MOL095) SLC18A2 (MOL095) SLC18A2 (MOL095) SLC18A2 (MOL097) SLC6A3 (MOL228) SLC1A3 (MOL228) SLC1A3 (MOL127) SLC6A5 (MOL252) SLC6A4 (MOL091) TH (MOL091) TH (MOL091) TH (MOL092) TPH2 (MOL091) TPH2 (MOL191) TREX1 (MOL191) TREX1 (MOL191) TREX1 (MOL191) TREX1 (MOL191) TREX1 (MOL192) ACARDI-GOUTIERES (6 GENES - ADAR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1) (MOL279) ACARDI-GOUTIERES (6 GENES) + MLPA [DEL/Dup]			•	
(MET23) CREATINE & GUANIDINOACETATE [PLASMA] (MOL074) PNPO (MOL247) PDXK (MET24) GLUCOSE [PLASMA] (MOL117) QDPR (MOL096) PTS (MET03) AMINO ACIDS [URINE] (MOL118) RNASEH2B (MOL124) RNASEH2A (MET14) ORGANIC ACIDS [URINE] (MOL095) SLC18A2 (MOL095) SLC18A2 (MET19) CREATINE & GUANIDINOACETATE [URINE] (MOL095) SLC18A2 (MOL095) SLC18A2 (MET20) ALPHA-AMINOADIPIC SEMIALDEHYDE [URINE; FOR PYRIDOXINE-RESPONSIVE SEIZURES] (MOL097) SLC6A3 (MOL228) SLC1A3 (MOL127) SLC6A5 (MOL252) SLC6A4 (MOL091) TH (MOL126) SPR (MOL092) TPH2 (MOL119) TREX1 (MOL119) TREX1 (ENZ01) AROMATIC L-AMINO ACID DECARBOXYLASE [AADC] ENZYMOLOGY (MOL181) AICARDI-GOUTIERES (6 GENES-ADAR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1) (MOL181) AICARDI-GOUTIERES (6 GENES) + MLPA [DEL/Dup]		•		
(MET24) GLUCOSE [PLASMA] (MOL196) PTS URINE (MET03) AMINO ACIDS [URINE] (MOL172) SAMHD1 (MOL168) RNASEH2A (MET14) ORGANIC ACIDS [URINE] (MOL172) SAMHD1 (MOL095) SLC18A2 (MET19) CREATINE & GUANIDINOACETATE [URINE] (MOL095) SLC18A2 (MOL095) SLC18A2 (MET20) ALPHA-AMINOADIPIC SEMIALDEHYDE [URINE; FOR PYRIDOXINE-RESPONSIVE SEIZURES] (MOL097) SLC6A3 (MOL228) SLC1A3 (MOL097) SLC6A5 (MOL127) SLC6A5 (MOL126) SPR (MOL091) TH (MOL126) SPR (MOL092) TPH2 (MOL119) TREX1 (MOL191) TREX1 (MOL191) TREX1 (MOL119) TREX1 (MOL181) AICARDI-GOUTIERES (6 GENES- ADAR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1) (MOL172) SCAMBOL (MOL172) AICARDI-GOUTIERES (6 GENES) + MLPA [DEL/Dup]				
URINE		•		
(MET03) AMINO ACIDS [URINE] (MOL168) RNASEH2C (MOL095) SLC18A2 (MOL095) SLC18A3 (MOL095) SLC18A3 (MOL095) SLC18A3 (MOL095) SLC18A3 (MOL097) SLC6A5 (MOL095) SLC18A3 (MOL097) SLC6A5 (MOL095) SLC1A3 (MOL177) SLC6A5 (MOL177) SLC6A5 (MOL177) SLC6A5 (MOL197) SLC6A5		•	• •	
(MET14) Organic acids [URINE] (MOL095) SLC18A2 (MOL095) SLC18A2 (MOL095) SLC18A2 (MOL228) SLC1A3 (MET19) Creatine & Guanidinoacetate [Urine] (MOL097) SLC6A3 (MOL228) SLC1A3 (MOL228) SLC1A3 (MOL228) SLC1A3 (MOL127) SLC6A5 (MOL252) SLC6A4 (MOL091) TH (MOL126) SPR (MOL091) TH (MOL126) SPR (MOL092) TPH2 (MOL119) TREX1 (MOL119) TREX1 (MOL119) TREX1 (MOL181) Aicardi-Goutieres (6 Genes- ADAR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1) (MOL279) Aicardi-Goutieres (6 Genes) + MLPA [DeL/Dup]				
(MET19) CREATINE & GUANIDINOACETATE [URINE] (MOL228) SLC1A3 (MOL228) SLC1A3 (MET20) ALPHA-AMINOADIPIC SEMIALDEHYDE [URINE; FOR PYRIDOXINE-RESPONSIVE SEIZURES] (MOL127) SLC6A5 (MOL252) SLC6A4 (MOL091) TH (MOL126) SPR (MOL092) TPH2 (MOL119) TREX1 (MOL181) AICARDI-GOUTIERES (6 GENES-ADAR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1) (MOL279) AICARDI-GOUTIERES (6 GENES) + MLPA [DEL/Dup]				
(MET20) ALPHA-AMINOADIPIC SEMIALDEHYDE [URINE; FOR PYRIDOXINE-RESPONSIVE SEIZURES] (MOL252) SLC6A4 (MOL091) TH (MOL126) SPR (MOL092) TPH2 (MOL119) TREX1 (MOL127) AICARDI-GOUTIERES (6 GENES-ADAR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1) (MOL279) AICARDI-GOUTIERES (6 GENES) + MLPA [DEL/Dup]				
(MOL091) TH (MOL126) SPR				
ENZYMOLOGY BLOOD (ENZO1) AROMATIC L-AMINO ACID DECARBOXYLASE [AADC] ENZYMOLOGY [PLASMA] (ENZO6) THYMIDINE PHOSPHORYLASE ENZYMOLOGY [WHOLE BLOOD ACD] IMMUNOASSAYS (MOL119) TREX1 GENE SEQUENCING PANELS [SANGER] + (MOL181) AICARDI-GOUTIERES (6 GENES- ADAR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1) [MOL279) AICARDI-GOUTIERES (6 GENES) + MLPA [DEL/Dup]	· · ·	☐ (MOL091) TH		
BLOOD (ENZ01) AROMATIC L-AMINO ACID DECARBOXYLASE [AADC] ENZYMOLOGY [PLASMA] (ENZ06) THYMIDINE PHOSPHORYLASE ENZYMOLOGY [WHOLE BLOOD ACD] IMMUNOASSAYS (MOL173) FOLIAR OF SERVIN CEREBON (MOL279) AICARDI-GOUTIERES (6 GENES) + MLPA [DEL/Dup]	-		,	
□ (ENZ01) AROMATIC L-AMINO ACID DECARBOXYLASE [AADC] ENZYMOLOGY [PLASMA] □ (ENZ06) THYMIDINE PHOSPHORYLASE ENZYMOLOGY [WHOLE BLOOD ACD] MMUNOASSAYS □ (MOL279) AICARDI-GOUTIERES (6 GENES) + MLPA [DEL/Dup]		☐ (MOL119) TREX1		
[PLASMA] □ (ENZ06) THYMIDINE PHOSPHORYLASE ENZYMOLOGY [WHOLE BLOOD ACD] IMMUNOASSAYS □ (MOL181) AICARDI-GOUTIERES (6 GENES- ADAR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1) □ (MOL279) AICARDI-GOUTIERES (6 GENES) + MLPA [DEL/Dup]				
□ (ENZ06) THYMIDINE PHOSPHORYLASE ENZYMOLOGY [WHOLE BLOOD ACD] IMMUNOASSAYS □ (MOL279) AICARDI-GOUTIERES (6 GENES) + MLPA [DEL/Dup]				
IMMUNOASSAYS ☐ (MOL279) AICARDI-GOUTIERES (6 GENES) + MLPA [DEL/DUP]				
[MCL279] ALARDI-GOOTIERES (O GENES) - MILE A [DELIDOF]				
	☐ (MET22) FOLATE RECEPTOR ANTIBODY ASSAY [PLASMA, SERUM; CEREBRAL			
FOLATE DEFICIENCY]		☐ (MOL284) AICARDI-GOUTIERES PANEL MLPA [DEL/DUP ONLY]		
[(METOS) For the property of	• • • • • • • • • • • • • • • • • • •	☐ (MOL178) DOPA-RESPONSIVE DYSTONIA (3 GENES- GCH1, SPR, TH) +☐ (MOL180) HYPEREKPLEXIA (5 GENES- ARHGEF9, GLRA1, GLRB, GPHN, SLC6A5))		
DEFICIENCY] (MOL178) DOPA-RESPONSIVE DYSTONIA (3 GENES- GCH1, SPR, TH)	• •			
·	DEI IOIENO 1			
, "				
☐ (MOL297) HYPEREKPLEXIA (5 GENES) +MLPA [DEL/DUP INCLUDED] ☐ (MOL285) HYPEREKPLEXIA PANEL MLPA: [DEL/DUP ONLY FOR GLRA1,				
		GLRB, SLC6A5]	FA. [DELIDUP UNLT FUR GLKAT,	

PAGE 1 OF 2 201604NEUROMETA



5424 Glenridge Drive Nf Atlanta, GA 30342 USA toll-free: 844.TESTMNG fax: 678.225.0212

NEUROCHEMISTRY AND METABOLIC TEST REQUEST FORM

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VISIT WWW.MNGLABS.COM/SUPPORT TO SUBMIT QUESTIONS BY SECURE HIPAA-COMPLIANT EMAIL FOR RAPID RESPONSE TO QUESTIONS.
VISIT WWW.MNGLABS.COM/TESTS FOR DESCRIPTIONS OF EACH GENE AND EACH TEST, AS WELL MOLECULAR DIFFERENTIAL DIAGNOSIS SEARCH.
ORDER TEST KITS ONLINE AT WWW.MNGLABS.COM/KITS FOR SIMPLE SAMPLE PROCESSING, AND FREE INBOUND AND RETURN OVERNIGHT SHIPPING.

PATIENT NAME:				DOB:			
		PATIEN	NT AND SPECIMEN INF	FORMATIO	N		
PATIENT LAST N	AME		PATIENT FIRST NAME				
PATIENT ID#			DATE OF BIRTH [MM/DD/YY	YY]			
DIAGNOSIS/ICD	-10		COLLECTION DATE [MM/DD	M/DD/YYYY]			
GENDER	☐ MALE ☐ FEMALE		SPECIMEN TYPE	□ WHOLE BLOOD □ FIBROBLA □ SKIN [FOR CULTURE] □ PLASMA □ BUCCAL SWAB □ MUSCLE □ DNA [DNA ISOLATION TISSUE]		☐ MUSCLE	☐ URINE☐ CSF
		REFE	RRING PHYSICIAN INF	ORMATION			
REFERRING PHY	SICIAN NAME				SIGNATURE		
REFERRING PH	REFERRING PHYSICIAN NPI # (REQUIRED)						
FACILITY/ORGANIZATION			PHONE				
SELECT AND PROVIDE EMAIL OR FAX FOR REPORT DELIVERY		PR EMAIL	☐ EMAIL ☐ FAX				
			BILLING INFORMATION				
FACILITY RESPONSIBLE FOR PAYMENT			PHONE				
FACILITY CONTACT PERSON			EMAIL				
FACILITY BILLING ADDRESS 1			FAX				
FACILITY BILLING ADDRESS 2							
CITY, STATE, ZIP CODE							
		RESULTS	S (SENT BY SECURE HIPAA-COMP	PLIANT EMAIL OR	FAX)		
	AUT	HORIZED RESULTS RECIP	PIENT 1	AUTHORIZED RESULTS RECIPIENT 2			
NAME							
FACILITY							
PHONE		MARK BOX AND EILL IN IN	JEORMATION FOR PREFERRED	RESHITS TRANS	SMISSION METHOD		
FAX		MARK BOX AND FILL IN INFORMATION FOR PREFERRED RESULTS TRANSMISSION METHOD					
EMAIL							
		CLINICAL INFORMATIO	ON: PLEASE INCLUDE/ ATT	ACH CLINICA	L INFORMATION		
			ATION FORM AVAILABLE A				



Atlanta, GA 30342 USA toll-free: 844.TESTMNG

CLINICAL INFORMATION FORM

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Patient Name (Last, First):		Date of Birth (MM/D	D/YYYY):
		Gender:	ale 🗆 Female
CLINICAL (CIRCLE	ALL THAT APPLY)	NERVE/ANTERIOR HORN CELL	
NEUROLOGY	COMMENTS	CHARCOT-MARIE-TOOTH	
EYE		NERVE CONDUCTION	
OPTIC ATROPHY		SENSORY	
RETINITIS PIGMENTOSA		Motor	
OTHER		AUTONOMIC	
HEARING		Pain	
SENSORINEURAL		NEUROFIBROMAS	
STICKLER		OTHER	
USHER		CARDIOLOGY	COMMENTS
COGNITIVE/NEUROBEHAVIORAL		CARDIOMYOPATHY	COMMENTO
INTELLECTUAL DISABILITY (ID)		DILATED	
SYNDROMIC ID		HYPERTROPHIC	
Nonsyndromic ID		Noncompaction	
		ARRHYTHMIAS	
AUTISM			
DEMENTIA		VENTRICULAR TACHYCARDIA	
EPILEPSY		Long or Short QT	
TONIC CLONIC		CONDUCTION DEFECT	
ABSENCE		BRUGADA	
MYOCLONIC		CONGENITAL HEART DEFECTS	
EPILEPTIC ENCEPHALOPATHY		DESCRIBE	
OTHER		HETEROTAXY	
NEURONAL MIGRATION		ENDOCRINE	COMMENTS
Joubert		DIABETES MELLITUS	
MECKEL		HYPOTHYROIDISM	
OTHER		OTHER	
STROKE		CONNECTIVE TISSUE/BONE	COMMENTS
MOVEMENT DISORDER		EHLERS DANLOS	
A TAXIA		MARFAN	
EPISODIC ATAXIA		ANEURYSMS	
Dystonia		OSTEOGENESIS IMPERFECTA	
CHOREA/ATHETOSIS			
PARKINSON DISEASE		OTHER	Company
L-Dopa Response		ADDITIONAL	COMMENTS
SPASTICITY			
SPASTIC QUADRIPLEGIA			
SPASTIC PARAPLEGIA			
OTHER			
NEUROMUSCULAR			
PROXIMAL OR DISTAL			
MUSCLE ATROPHY	1		
RHABDOMYOLYSIS	1		
	1		
STATIN USE	1		
MALIGNANT HYPERTHERMIA	1		
CONTRACTURES	i		
ARTHROGRYPOSIS			
Myasthenia	1		
PERIODIC PARALYSIS			



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Patient Name (Last, First):	Date of Birth (MM/DD/YYYY):		
	Gender:	☐ Male ☐ Female	
	3040	Li Male Li Telliale	
IMAGING (CIRCLE ALL THAT APPLY)		AMILY HISTORY	
Brain MRI		D MATERNAL LINEAGE	
LEIGH DISEASE	RELATIONSHIP TO PROBAN	D SYMPTOMS	
BASAL GANGLIA CALCIFICATION			
STROKE CEREBELLAR ATROPHY			
ABNORMAL MYELIN			
(DESCRIBE)			
EEG (DESCRIBE FINDINGS)			
, ,			
	A		
	RELATIONSHIP TO PROBAN	D PATERNAL LINEAGE D SYMPTOMS	
	RELATIONSHIP TO PROBAN	D STMPTOMS	
EMG/NCV (DESCRIBE FINDINGS)			
Zime/Net (Szechisz i insured)			
LABORATORY			
METABOLIC (DESCRIBE FINDINGS)			
		SIBLINGS	
	NUMBER (SPECIFY GENDER	R) HEALTHY/AFFECTED	
СРК			
MAXIMUM			
Мінімим			
MINIMOW			
GENETIC (DESCRIBE FINDINGS)			
CHROMOSOME MICROARRAY	ETHNIC	ITY (PLEASE CIRCLE)	
	AFRICAN		
	EAST ASIAN		
DELETION/INSERTION TESTING	South Asian		
	EUROPEAN (NON- F	INNISH)	
	EUROPEAN (FINNIS		
OTHER	LATINO	•	
	• HEN		