

lab use only

Ship to: Neurochemistry/AminoAcid Lab
Massachusetts General Hospital
55 Fruit St GRJ-239, Boston MA 02114 USA

CLIA ID #: 22D0650226

Tel: (617) 726-3884

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Patient Information

Name: _____

Sex: Male Female

I.D #: _____

Birthdate: _____

Working diagnosis: _____

Medications/Comments: _____

Specimen: _____

Date/Time of collection (required) _____

R = 5mL red L = 3mL Lavender P = 10mL Purple u = 5-10mL urine CSF = 1 mL plain s = Special tube

Amino Acid Analysis

- AA, quant, complete panel, plasma PQAA
- Special panels, plasma
- MSUD: ala, alloile, gln, ile, leu, val MSUDQ
- ORNITHINE: arg, cit, gln, lys, orn ORNQ
- PHE/TYR: phe, tyr, met PKU carrier test PKUQ
- SULFUR: cysta, cys, h-c disulf, met, ser, tau SAAQ
- UREA CYCLE: arg, asa, cit, gln UCDQ
- AA, quant, complete panel, urine VQAA
- Special panels, urine:
- CYS/LPI: arg, cys, lys, orn CYSQ
- HCS: cys, homocys-cys disulf, hcs, met VHCSQ
- UREA CYCLE: arg, asa, cit, hcit, lys, orn VUCDQ
- AA screen, urine VAA
- AA, quant, complete panel, CSF CQAA
- Alanine, plasma PALA
- CSF Alanine, CSF CSFALA
- Arginine, plasma PARG
- s Argininosuccinate/AA, quant, amn fluid AFQAA
- Aspartic acid, plasma PASPA
- Citrulline, plasma PCIT
- Cystine, urine V1CYS
- Glutamine, plasma PGLN
- CSF Glutamine, CSF CSFGLN
- Glycine, plasma PGLY
- CSF Glycine, CSF CSFGLY
- CSF Glycine plasma/CSF ratio (NKH Test) NKHQ
- Homocitrulline, urine HCIT
- Kynurenine, plasma PKYN
- Ornithine, plasma PORN
- Tryptophan, plasma PTRYP

* call for special handling instructions

Organic Acids / Urine testing / Misc tests

- α -ketoacids/ketones (MSUD) ACDNPH
- Canavan Disease / N-acetylaspartate OA
- Cystine/Homocystine/Nitroprusside, urine NPRUSS
- GAMT def / Guanidinoacetate screen MISCT
- Ketones/ α -ketoacids, urine ACDNPH
- Organic acids, semi-quant, GC-MS OA
- Reducing Substances CLINI
- Succinylpurines/sugars by TLC TLC
- Sulfite (fresh or fresh frozen urine required) MISCT
- Thiosulfate THIOQ

Enzyme Assays — prearrangement only

- s Arginase, RBCs * BUCE
- s Fumarase activity, fibroblasts * ENZY
- s Ornithine aminotransferase, fibroblasts * MISCT
- s Ornithine incorporation assay, fibroblasts * MISCT

DNA Tests — prearrangement only

- Ps DNA isolation/extraction MISCT
- P Homocystinemia, C β S G307S, I278T DNACBS
- P MCAD, 985A>G MCAD
- P MTHFR, 677C>T, 1298A>C, 1317T>C MTHFR
- P Sulfite oxidase, SUOX mutation analysis MISCT

Tissue Culture — prearrangement only

- s Skin biopsy culture SKFB
- s Cryopreservation ea cell line MISCT
- s Cell culture expansion, fibroblasts MISCT

Ordering physician: _____

Address: _____

Telephone/FAX: _____

Requesting location / reporting address:

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