INHERITED METABOLIC DISORDERS INVESTIGATION
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IWK LAB USE ONLY	

Biochemical Genetics Testing Requisition

PATIENT DEMOGRAPHICS

_____ _____
Last Name, First Gender

_____ _____
Birth Name/Mom's Last Name (patient < 18yr) K# if Born after2015

_____ _____
Date of Birth (dd/mm/yy) Health Card # (province)

_____
Mailing Address Postal Code

PERTINENT CLINICAL HISTORY

☐ Dev. Delay ☐ Epilepsy ☐ Hypoglycemia ☐ Acidosis

☐ Hypotonia ☐ Encephalopathy ☐ Family Hx of IEM

☐ NBS positive for _____

☐ Renal stones ☐ Other (specify):

_____

_____

MEDICATIONS (list all):

_____

_____

NUTRITION/DIET:

☐Normal Diet ☐ TPN ☐ MCT oil ☐ Formula

☐ Other _____

ORDERING PHYSICIAN

☐Medical Geneticist ☐Neurologist ☐ Other _____

_____
Last Name, First Phone/Pager/FAX#

_____ _____
SIGNATURE Date (dd/mm/yy)

***fill below if you are Not a Medical Geneticist and order MMGS-only tests. Indicate if the Geneticist would like a copy of result.**

☐Discussed with Geneticist ☐Copy report to

_____
Last Name, First Phone/Pager/FAX#

SPECIMEN COLLECTION

☐ Diagnostic ☐ Monitoring

☐ STAT ☐ URGENT ☐ ROUTINE

_____ _____
Time/Date of collection (dd/mm/yy) Collector's Initials

☐ Blotter ☐ Plasma ☐ CSF ☐ Serum ☐ Other _____

Urine: □ random □ 24hr

Start: Stop: TV (ml): pH:

TEST REQUESTED (IN HOUSE)

☐ Blotter Carnitine/Acylcarnitine - **CARNBS**

☐ Plasma Amino Acid – **AAAP**

☐ Urine Organic Acid – **ORGA**

☐ CSF Amino Acid – **AAACSF (simultaneous CSF & plasma)**

☐ Urine Amino Acid – **AAU (please check indications)**

☐ Blotter GALT Deficiency Screen – **GALTSCR***

☐ Blotter Biotinidase Deficiency Screen – **BIOTSCR***

REFERRED OUT TESTS (MMGS ONLY)*

☐ Serum Carnitine/Acylcarnitine - **CARNFRSER**

☐ Urine Mucopolysaccharide Screen - **MPS**

☐ Urine Oligosaccharide Screen - **OLIGSU**

☐ CUD (NBS positive follow/up) - **FCUD (plasma & urine)**

☐ Galactosemia confirmation (NBS+ F/U) - **GALT & GALTGA**

☐ Serum Biotinidase (NBS+ F/U) - **BIOTS**

☐ Plasma Homocysteine (>5y) - **HOMOCYS**

☐ Plasma Homocysteine (=/<5y) - **HOMOCYST**

☐ Other: _____

_____

MMGS Only*: These Tests must be ordered by Maritime Medical Genetics Service (MMGS) or discussed with a Medical Geneticist.

Please see the EXPLANATORY NOTES in next page.

https://www.iwk.nshealth.ca/newbornscreening

EXPLANATORY NOTES

1. *COMPLETION OF THE FORM*

- Young patient's birth name and K# (if applicable) are very important to link the patient's previous newborn screening (NBS) result to facilitate result interpretation and avoid duplicate K# in the IWK LIS system.
- It is important to provide relevant clinical history, abnormalities found on examination, all medications, nutritional intervention. Current biochemical data (e.g. glucose, ketones, ammonia, etc.) may aid interpretation greatly.

Most of the Referred-out tests require Pathologist/Biochemist approval before sending. If there is insufficient information (including physician's signature and contact), these tests may be put on-hold. Ordering physician is required to fill and submit the referring lab's requisition along with IWK requisition.

2. *SPECIMEN COLLECTION, STORAGE AND SHIPPING REQUIREMENTS (see more detailed User Notes in MEDITECH)*

Analytes	Mnemonics (IWK LIS)	Specimen (minimum amount)	Testing Centre	Storage & shipping	User Notes (simplified)
Carnitine & Acylcarnitine	**CARNBS**	Blotter (1 DBS)	IWK	Ambient	Capillary whole blood preferred, Venous non-EDTA acceptable, 2 filled circles. Dry flat 3 hrs at room temp.
	CARNFRSER	Serum (0.1ml)	CHUS	Frozen	SST tube preferred
	FCUD	Hep. Plasma (0.5ml) & Urine (0.6 ml)**	Duke	Frozen	For NBS positive Carnitine Uptake Defect (CUD) only. Rarely ordered for adults who are suspicious for CUD.
Amino acids	**AAACSF**	CSF (0.5 ml)***	IWK	Frozen	Should have a matching plasma and AAAP ordered
	AAAP	Hep. Plasma (0.5ml)	IWK	Frozen	Fasting preferred. Green-top (no gel) tube. Spin and send aliquot.
	AAU	Urine (1 ml)**	IWK	Frozen	
GALT Activity	**GALTSCR**	Blotter (1 dot)	IWK	Ambient	For non-neonate, screening. Collect as CARNBS above.
	GALT & GALTGA	Whole blood (2 ml)	Mayo	Refrigerate /Ambient	For NBS positive GALT, diagnostic test. Lavender-top EDTA Vacutainer preferred. Refrigerate is preferred
Biotinidase Activity	**BIOTSCR**	Blotter (1 dot)	IWK	Ambient	For non-neonate, screening. Collect as CARNBS above.
	BIOTS	Serum (0.5ml)	Mayo	Frozen	For NBS positive BIOT, diagnostic test
Organic acids	**ORGAT**	Urine (1.5 ml)**	IWK	Frozen	Random urine, freeze
Mucopolysaccharides	**MPS**	Urine (1 ml)**	ICL	Frozen	10ml needed for further analysis
Oligosaccharides	**OLIGSU**	Urine (1 ml)**	ICL	Frozen	Random urine, freeze
	HOMOCYS	EDTA Plasma (2 ml)	ICL	Frozen	Fasting preferred. Spin within 1 hr or keep on ice until spun (max. 4 hrs). Patient > 5 years-old
	HOMOCYST	Serum/hep. Plasma (0.1 ml)	CHUS	Frozen	Fasting preferred. Spin within 1 hr or keep on ice until spun (max. 4 hrs). Patient =/< 5 years-old

** **Urine:** Use clean catch techniques; results may be uninterpretable in the presence of significant proteinuria or urinary tract infection. If a 24-hour collection is needed, precise timing is important. Please make note of the total volume and pH. Please follow testing centre's requirement for specimen type and amount required. Store and send frozen.

*** **CSF (CerebroSpinal Fluid):** Analysis of CSF amino acid is only rarely indicated (EXCEPT non-ketotic hyperglycinemia) and should be accompanied by **matching plasma** for proper interpretation. Store and send frozen – for amino acid profiling.

3. *INDICATIONS FOR SPECIFIC REQUESTS*

Blotter Carnitine/Acylcarnitine profile: Quantitative screening: uses the same platform for NBS. Often used for NBS positives follow up, monitoring treatment, ketogenic diet monitoring and screening test for non-neonate who is clinically suspicious for a Fatty acid oxidation disorder (FAOD) or certain organic acidopathy.

Plasma Carnitine/Acylcarnitine profile: Quantitative: Often used for NBS positive confirmation or non-neonate who is clinically suspicious for an FAOD or certain organoacidopathy.

Plasma Amino Acid Analysis, Quantitative: This test often used for confirmation diagnosis and treatment monitoring of some NBS positives including PKU, MSUD, ASA, Citrullinemia etc. This test is also recommended for a patient who is clinically suspicious for aminoacidopathies including urea cycle defects.

Urine Amino Acid Analysis, Quantitative: Don't routinely order UAA as part of a screen for IEM or in a work-up for critical hypoglycemia. UAA may be indicated to facilitate the diagnosis of Homocystinuria, Cystinuria (renal stones/hematuria) etc. Rarely, this test is used to provide information for renal Fanconi syndrome.

Organic Acid Analysis, Semi- Quantitative: Often used for diagnosis confirmation and treatment monitoring of some NBS positives including PA/MMA, IVA, GA1, MSUD, MCAD etc. Also a recommended screening test for patients with unexplained lactic acidosis, metabolic acidosis, hypo/hyperglycemia, ketonuria, hyperammonenia, anemia/neutropenia, unusual odor, neurological deficit, poor feeding or unusual food aversion, etc.

Oligosaccharide/Mucopolysaccharide Screens (Urine Only – 3 sequential early morning samples are recommended): False negatives are not uncommon and special testing may be indicated if clinical features are compelling. Testing is indicated for coarse facies, short stature, dysostosis (multiplex), neurodegenerative disease, evidence of lysosomal storage disease.

Other: This includes rarer referred out screening tests for defects in purine or pyrimidine metabolism, specific enzyme assays or other specific pre-arranged tests. Please contact lab for specimen collection procedure.