London Health Sciences Centre—Molecular Diagnostics

Requisition for DNA Testing: Biochemical Genetics

Family Information

Have samples from this family been sent to a DNA lab before?

☐ Yes ☐ No

If Yes, specify:_________________________________

☐ This individual is the index case

☐ Name of index case in the family:___________________
Date of birth:_____________________________________
Relationship t this patient: __________________________

Patient information:

Name: ____

Birthdate:(YYMMDD)___________________________

Address: _____________________________________

Health Card Number: ___________________________

Pedigree:______________________________________

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Test Request:

- ☐ Adenosine deaminase deficiency (ADA)
- ☐ Arginase deficiency (ARG1)
- ☐ Biotinidase deficiency (BTD)
- ☐ Chondrodysplasia punctata 1 (ARSE)
- ☐ Cystinosis (CTNS)
- ☐ Glutaric acidemia, type I (GCDH)
- ☐ Glycogen storage disease, type IV (GBE1)
- ☐ GTP cyclohydrolase I deficiency (GCH1)
- ☐ Lesch-Nyhan syndrome (HPRT1)
- ☐ Metachromatic leukodystrophy (ARSA)
- ☐ Mevalonic aciduria (MVK)
- ☐ Microcephaly, Amish type (SLC25A19)
- ☐ Niemann Pick disease, type C1/C2 (NPC1, NPC2)
- ☐ Ornithine transcarbamylase deficiency (OTC)
- ☐ Transcobalamin II deficiency (TCN2)
- ☐ Other rare or familial mutation (specify):

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Mitochondrial disorders:

- ☐ Whole mtDNA genome NGS including deletion detection and heteroplasmy analysis

Point mutaitons

- ☐ 3243A>G (MELAS)
- ☐ 3260A>G (myopathy)
- ☐ 3303C>T (cardiomyopathy)
- ☐ 8344A>G (MERRF)
- ☐ 8993T>G/C (NARP/Leigh disease)
- ☐ Other rare or familial mutation (specify):__________
- ☐ Hepatocerebral mtDNA depletion syndrome (DGUOK)
- ☐ Myopathic mtDNA depletion syndrome (TK2)

Reason for Referral

- ☐ Documented family history of indicated disease
- ☐ Possible family history of indicated disease
- ☐ Symptoms of indicated disease in this individual
- ☐ Other________________________________

Information Requested

- ☐ Bank until further notice
- ☐ Confirm clinical diagnosis
- ☐ Carrier status
- ☐ To be referred out (specify):_________________
- ☐ Other

Sample Collection

- ☐ EDTA blood(lavender top)_______cc room temp
- ☐ DNA ___________ng/ul
- ☐ Fibroblast culture
- ☐ Tissue (specify):____________________________
- ☐ Prenatal (specify):___________________________
- ☐ Other

AUTHORIZED SIGNATURE IS REQUIRED

Referring Physician:_____________.

Signature: ___________________________________

Address: ____________________________________

Telephone: ______________Fax:_________________

Billing address:_______________________________

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Pathology and Laboratory Medicine