NEWBORN SCREENING **ONTARIO**
DÉPISTAGE NÉONATAL **ONTARIO**

MOLECULAR REQUISITION

SHIP SAMPLES TO: **NSO SPECIMEN HUB**
415 Smyth Road Ottawa, ON K1H 8M8

Lab Use Only

PATIENT INFORMATION

Health Card Number

Sex Assigned at Birth
○ Male ○ Female ○ Ambiguous ○ Unknown

Gender Identity (Optional)
○ Male ○ Female ○ Non-binary/X/U

Date of Birth (yyyy/mm/dd)

MRN/Hospital

Patient's Last Name

Patient's First Name

Patient's Address

Patient's Telephone Contact Number

Ancestry: _____ ☐ Fetal Sample

For STAT requests please indicate how a shorter TAT will change patient management:
○ Ongoing pregnancy; result needed for decision making within 6wks (i.e. termination or birth)
Estimated due date:
○ Positive newborn screens where molecular results are essential for treatment decisions
○ Expedited results will directly impact management decisions
For inquiries please contact nsomolecular@cheo.on.ca

ORDERING PROVIDER
Name
Email
Phone Fax
Institution

Copy results to clinician/practitioner:
Name
Phone Fax

Copy results to clinician/practitioner:
Phone Fax

TEST REQUESTED *see FAQ section on NSO website for more information*

- ☐ Targets of Newborn Screening – targeted panel (complete Section 1)
- ☐ Primary Immune Deficiencies – augmented exome slice (complete Section 2; whole blood or DNA)
- ☐ Mitochondrial Diseases – augmented exome slice (complete Section 3; whole blood or DNA)
- ☐ TREC (DBS only; for out-of-province newborn screening samples only)
- ☐ SMA - ddPCR/MLPA (*SMN1* and *SMN2* copy number only)
- ☐ *CFTR* common mutation panel (for carrier testing and CF newborn screen positive only)
- ☐ Familial Variant Testing (complete table below)
- ☐ Maternal Cell Contamination (MCC) studies (for prenatal and umbilical cord blood testing)

SPECIMEN TYPE

- ☐ Whole Blood (room temp, EDTA tubes)
  Adult 1x5mL, Children 1x4mL, Infant ≤2yo 1x3mL
- ☐ Umbilical Cord blood (maternal sample for MCC studies required) ☐ EDTA blood ☐ DBS
- ☐ DNA (>5 µg with at least [50ng/ µL])
  Source: _____
- ☐ DBS (Dried bloodspot - Whatman 903)
- ☐ Other:_____
  Contact NSO prior to sending

☐ Variant reinterpretation (must attach NSO report issued >=1 year ago)

SPECIMEN COLLECTION

Date of collection (YYYY/MM/DD)		Time of Collection (24HR)	
# Tubes (if applicable)		Specimen ID	

Please contact us if this is a precious sample. For more information on precious samples and our sample retention policy, please visit our website.

AUTHORISATION

I certify that the patient and/or legal guardian has been informed of the nature of the genetic test requested, including benefits, risks, possible results, limitations and possible implications for himself/herself and his/her family. I have answered this person's questions and have obtained informed consent for this testing.

Signature of the ordering healthcare provider:

TESTING FOR KNOWN FAMILIAL VARIANT(S)

☐ **Please provide proband's report or NSO report number and family history**

Proband's Name / DOB:		Relationship to Proband:	
Gene and Variant(s): *Transcript (NM number) required if report not attached*			

Personal History: ☐ Asymptomatic ☐ Symptomatic:

Family History:

Name(s) and DOB of other submitted family members:

Complete NSO's billing form if patient is not covered by OHIP; attach subsequent pages/sections as needed

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SECTION 1: MOLECULAR TESTING FOR DISEASES TARGETED BY NEWBORN SCREENING ONTARIO

Disease Targeted:

Gene (or choose from list below); *If a multi-gene panel is being requested, please indicate if you are suspicious of a specific gene(s)*:

Clinical Indication:

Family History (please attach all relevant documents related to previous test results and clinical diagnosis):

AMINO ACID DISORDERS *(requesting a panel is equivalent to requesting all related subpanels)*

x	PANEL	x	SUBPANEL	GENES
☐	Homocystinuria	☐	Hypermethioninemia	*ADK, AHCY, CBS, GNMT, MAT1A, SLC25A13*
		☐	Hypomethioninemia	*MTHFR, MTR, MTRR*
☐	Phenylketonuria	☐	PAH Deficiency	*PAH* (sequencing + reflex MLPA as needed)
		☐	Biopterin Deficiencies	*DNAJC12, GCH1, PCBD1, PTS, QDPR, SPR*
☐	Tyrosinemia	☐	Elevated Succinylacetone	*FAH, GSTZ1*
		☐	Elevated Tyrosine	*HPD, TAT*
☐	Urea Cycle Diseases	☐	High citrulline	*ASS1, SLC25A13*
		☐	High ASA	*ASL*
		☐	Low citrulline	*CPS1, NAGS, OTC*
		☐	Other	*ARG1, CA5A, GLUL, GLUD1, OAT, SLC7A7, SLC25A2, SLC25A15*
☐	Maple Syrup Urine Disease			*BCKDHA, BCKDHB, DBT, DLD*

ORGANIC ACID DISORDERS *(requesting a panel is equivalent to requesting all related subpanels)*

x	PANEL	x	SUBPANEL	GENES
☐	Multiple carboxylase Deficiency	☐	Biotinidase Deficiency	*BTD*
		☐	Other	*CA5A, HLCS*
☐	Propionic / Methylmalonic acidemias	☐	PA	*PCCA, PCCB*
		☐	MMA	*ACSF3, ALDH6A1, MCEE, MLYCD, MMAA, MMAB, MMUT, SUCLA2, SUCLG1*
		☐	MMA + Homocysteinemia	*ABCD4, AMN, CBLIF, CD320, CUBN, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2*
☐	Isovaleric acidemia			*ACADSB, FLAD1, IVD*
☐	Glutaric aciduria Type 1			*GCDH*
☐	Isobutyryl-CoA dehydrogenase deficiency			*ACAD8*
☐	Succinic semialdehyde dehydrogenase deficiency			*ALDH5A1*
☐	b-ketothiolase deficiency			*ACAT1*
☐	Guanidinoacetate Methyltransferase Deficiency			*GAMT*

FATTY ACID OXIDATION DISORDERS ☐ Check here to request ALL genes noted below

x	PANEL	GENES
☐	Carnitine Uptake Deficiency	*SLC22A5*
☐	MCAD Deficiency	*ACADM* (sequencing + reflex MLPA as needed)
☐	LCHAD/MTP Deficiency	*HADHA, HADHB*
☐	VLCAD Deficiency	*ACADVL*
☐	MADD/Glutaric Aciduria Type2	*ETFA, ETFB, ETFDH, FLAD1, SLC52A2, SLC52A3, SLC52A1*
☐	CPT2 Deficiency	*CPT2*
☐	CACT Deficiency	*SLC25A20*
☐	CPT1 Deficiency	*CPT1A*
☐	Other FAOD	*ACAA2, ACAD9, ACADL, ACADS, ECHS1, HADH*

CONGENITAL ADRENAL HYPERPLASIA (if both requested, *CYP21A2* will be performed first and reflex to panel)

☐	21-Hydroxylase Deficiency	*CYP21A2* (includes MLPA and long-range PCR analyses for CNVs and common rearrangements)
☐	Other	*ARMC5, CYP11B1, CYP11B2, CYP17A1, HSD3B2, POR, PRKAR1A, STAR*

GALACTOSEMIA

☐	GALT Deficiency	*GALT*
☐	Other	*GALK1, GALE, GALM, GLUT2 (SLC2A2)*

MUCOPOLYSACCHARIDOSIS TYPE I

☐	Mucopolysaccharidosis type I	*IDUA*

PEROXISOMAL STORAGE DISORDERS ☐ Check here to request ALL genes noted below

☐	XL-Adrenoleukodystrophy	*ABCD1*
☐	Zellweger Spectrum	*ACBD5, ACOX1, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7*

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SECTION 2: MOLECULAR TESTING FOR INBORN ERRORS OF IMMUNITY

PANEL SELECTION

☐ Severe Combined/Primary Immune Deficiency (251 gene augmented exome slice), please visit our website for full list of genes

SUBPANELS

☐ ADA Deficiency (*ADA*)

☐ Chronic Granulomatous Disease (*CYBA, CYBB, CYBC1, G6PD, NCF1*, NCF2, NCF4*) [*limited coverage due to high homology with duplicated regions in genome; please note that this gene is not included in the full severe combined/primary immune deficiency panel]

**Additional testing to ensure full coverage of NCF1 can only be requested if patient has had an abnormal neutrophil oxidative burst index*

☐ Aicardi-Goutières syndrome (*ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1*)

CLINICAL DETAILS

Please provide detailed information regarding patient's phenotype, age of onset of symptoms, previous tests completed, and family history:

- Age of onset:
- Family history:
- Other:

CLASSIC PRESENTATIONS

☐ ADA deficiency
☐ Aicardi-Goutières syndrome
☐ Autoimmune lymphoproliferative syndrome
☐ Chronic granulomatous disease
☐ Common variable immunodeficiency
☐ Familial cold autoinflammatory syndrome
☐ G6PD deficiency
☐ Hyper IgE syndrome – Autosomal Dominant
☐ Hyper IgE syndrome – Autosomal Recessive
☐ Mendelian susceptibility to mycobacterial disease
☐ Severe combined immunodeficiency
☐ Wiskott-Aldrich syndrome
☐ **Other (indicate if you are suspicious of a specific gene):**

LABORATORY FEATURES

☐ Elevated inflammatory markers
☐ Anemia
☐ Neutropenia
☐ Lymphopenia
☐ Thrombocytopenia
☐ Eosinophilia
☐ Abnormal Neutrophil Oxidative Burst Index
☐ Abnormal TREC assay
☐ Low or absent CD4+ T cell number
☐ Low or absent CD8+ T cell number
☐ Abnormal T cell function
☐ Low or absent NK function
☐ Low or absent B cell number
☐ Agammaglobulinemia
☐ Increased immunoglobulins: ☐ IgG ☐ IgA ☐ IgM ☐ IgE
☐ Decreased immunoglobulins: ☐ IgG ☐ IgA ☐ IgM ☐ IgE
☐ Poor specific antibody response to vaccine

CLINICAL FEATURES

RHEUMATOLOGICAL/IMMUNE DYSREGULATION

☐ Arthritis
☐ Granulomas
☐ Hepato/splenomegaly
☐ Lymphadenopathy
☐ Recurrent fevers
☐ Systemic lupus erythematosus
☐ Vasculitis

HEMATOLOGICAL

☐ Autoimmune cytopenia
☐ Bone marrow failure
☐ Evan's syndrome
☐ Hemophagocytic lymphohistiocytosis
☐ Lymphoma

GASTROINTESTINAL

☐ Chronic diarrhea
☐ Celiac disease
☐ Enteropathy
☐ Inflammatory bowel disease
☐ Perianal abscess/fistula
☐ Liver/biliary disease

INFECTIONS

☐ Abscesses
☐ Candidiasis
☐ Epstein-Barr virus
☐ Mycobacterium tuberculosis
☐ Non-tuberculous mycobacteria
☐ Recurrent infections: ☐ bacterial ☐ fungal ☐ viral
☐ Recurrent pneumonia
☐ Skin and/or connective tissue infections

DERMATOLOGICAL

☐ Alopecia
☐ Bullous pemphigoid
☐ Dermatitis/eczema
☐ Psoriasis
☐ Urticaria
☐ Vitiligo
☐ Warts

PULMONARY

☐ Asthma
☐ Bronchiectasis
☐ Chronic obstructive pulmonary disease
☐ Interstitial lung disease

OTHER

☐ Developmental delay
☐ Endocrinopathy
☐ Facial dysmorphisms
☐ Failure to thrive
☐ Hearing loss
☐ Microcephaly
☐ Short stature
☐ Unexplained weight loss

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SECTION 3: MOLECULAR TESTING FOR MITOCHONDRIAL DISEASES

Criteria for testing requires selections from at least one classic presentation OR at least one pathologic/lab feature OR at least one biochemical feature OR (at least one sign in CNS/heart/eyes/muscles AND one "other")

PANEL SELECTION

☐ Full Mitochondrial Nuclear Gene Panel (437 gene augmented exome slice), please visit our website for full list of genes

SUBPANELS

- ☐ Mitochondrial Encephalopathy / Leigh Disease (117 genes)
- ☐ mtDNA Depletion and Deletion (19 genes)
- ☐ Progressive External Ophthalmoplegia (PEO) / Optic Atrophy (77 genes)
- ☐ Pyruvate Dehydrogenase Complex Deficiency (16 genes)
- ☐ Hydroxyglutaric Aciduria (4 genes)

Please contact laboratory to request another subset of the full nuclear gene panel

CLINICAL DETAILS

Please provide detailed information regarding patient's phenotype, age of onset of symptoms, previous tests completed, and family history:

- Age of onset:
- Family history:
- Other:

CLASSIC PRESENTATIONS

- ☐ Alpers disease
- ☐ Chronic progressive external ophthalmoplegia (CPEO)
- ☐ Gentamicin-related sensorineural hearing loss
- ☐ Kearns-Sayre syndrome
- ☐ Leber's hereditary optic neuropathy (LHON)
- ☐ Leigh disease
- ☐ Mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS)
- ☐ Mitochondrial neuro-gastro-intestinal encephalopmyopathy (MNGIE)
- ☐ Multiple symmetric lipomatosis
- ☐ Myoclonic epilepsy with ragged-red fibers (MERRF)
- ☐ Neuropathy, ataxia, and retinitis pigmentosa (NARP)
- ☐ Pearson syndrome
- ☐ Primary lactic acidosis
- ☐ Sensory-ataxia, neuropathy, dysarthria and ophthalmoparesis (SANDO)
- ☐ **Other (indicate if you are suspicious of a specific gene):**

PATHOLOGIC/LABORATORY FEATURES

- ☐ Ragged red fibers: _____ %
- ☐ COX-negative fibers: _____%
- ☐ Ultrastructurally abnormal mitochondria by electron microscopy
- ☐ Muscle biopsy consistent with mitochondriopathy (affix report)

BIOCHEMICAL FEATURES

- ☐ Persistent hyperalaninemia
- ☐ Persistent abnormal excretion of lactate, pyruvate of TCA intermediates in urine
- ☐ Evidence of mtDNA depletion or multiple mtDNA deletions (affix results)
- ☐ <30% activity of any RC complex in tissue or cell line
- ☐ Increased lactate pyruvate ratio (>25) in skin fibroblasts

CLINICAL FEATURES

CENTRAL NERVOUS SYSTEM (CNS)

- ☐ Developmental delay
- ☐ Regression
- ☐ Movement disorder
- ☐ Seizures
- ☐ Hemiplegic or complicated migraine
- ☐ Peripheral neuropathy
- ☐ Sensorineural hearing loss

HEART

- ☐ Arrhythmias
- ☐ Cardiomyopathy
- ☐ Conduction block

EYES

- ☐ Optic atrophy
- ☐ Pigmentary retinopathy

MUSCLES

- ☐ Hypotonia
- ☐ Rhabdomyolysis
- ☐ Fixed weakness of skeletal muscle
- ☐ Ataxia

OTHER

- ☐ Clinical progression with stepwise exacerbation of symptoms
- ☐ Elevated alanine (PAA)
- ☐ Elevated 3-methylglutaconic acid (UOA)
- ☐ GI tract: pseudoobstruction
- ☐ GI tract: hepatopathy
- ☐ Lactic acidosis (in non-acute illness setting)
- ☐ Proximal renal tubulopathy (Fanconi syndrome)
- ☐ Sideroblastic anemia
- ☐ Short stature (<2 SD below normal)
- ☐ Type 2 diabetes mellitus
- ☐ Unexplained failure to thrive

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