Requisition for DNA Testing

Reason for Referral:

- ☐ Diagnostic Testing:
  - ☐ Affected
  - ☐ Unaffected
- ☐ Carrier testing/Known Family Mutation

Name of index case in the family *(include copy of report):*

_____

Date of Birth: _____

Relationship to this patient: _____

Gene: _____ Mutation: _____

RefSeq:NM: _____

- ☐ Prenatal Diagnosis
- ☐ DNA Banking
- ☐ RNA Banking
- ☐ Referral to an outside laboratory *(must specify lab):*

_____

Sample Collection:

Date drawn: *(YYYY/MM/DD)* _____

- ☐ EDTA blood (lavender top)(min. 2ml at room temp)
- ☐ EDTA bone marrow (lavender top)(min. 2ml at room temp)
- ☐ DNA (100ng to 1ug):_____ ug
- ☐ Fresh/Frozen Tissue *(provide tissue source):* _____
- ☐ Formalin fixed paraffin embedded tissue (FFPE) (slides preferred)
- ☐ Other: _____

Clinical Diagnostics and Family History:

_____

_____

_____

_____

Lab Use Only:

Received date:

_____

Notes:

_____

_____

_____

_____

Patient Information:

INCOMPLETE REQUESTS WILL BE BANKED

Name: _____

Birthdate: _____

Address: _____

Sex: ☐ Male ☐ Female

Health Card Number: _____

Test Requests:

Use attached menu to select panels or individual genes. Panels, sub-panels or individual genes may be selected using the checkbox adjacent to the item of interest.

_____

_____

_____

Request for Expedited Result:

☐ Pregnancy *(L.M.P., YY/MM/DD):* _____

Medical intervention *(specify with date):*

_____

_____

_____

AUTHORIZED SIGNATURE IS REQUIRED

Referring Physician:

Physician name (print): _____

Signature: _____

Address: _____

Telephone: _____ Fax: _____

E-mail address: _____

CC report to:

Name: _____

Address: _____

Telephone: _____ Fax: _____

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Requisition for DNA Testing

FOR SAMPLE REQUIREMENTS SEE: http://www.lhsc.on.ca/lab/molegen/index.htm

Patient Information:

Name: _____ Birthdate: _____

NGS Panels (includes deletion/duplication analysis)

Hereditary Cancer *(if patient meets OBSP criteria, please use provincial cancer requisition)*

☐ **Hereditary Cancer – Comprehensive (38)**
APC (incl. 5'UTR), ATM, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, CTNNA1, EPCAM, FANCC, FANCM, FLCN, GREM1, HOXB13, MEN1, MLH1 (incl. 5'UTR), MSH2, MSH3, MSH6, MUTYH, NBN, NTHL3, PALB2, PMS2, POLE, POLD1, PTEN (incl. 5'UTR), RAD51C, RAD51D, SDHB, SMAD4, STK11, TP53

☐ **Hereditary Cancer – High Penetrance (16)**
APC (incl. 5'UTR), ATM, BRCA1, BRCA2, CDH1, CHEK2, EPCAM, MLH1 (incl. 5'UTR), MSH2, MSH6, MUTYH, PALB2, PMS2, PTEN (incl. 5'UTR), STK11, TP53

☐ **Hereditary Cancer – Breast/Ovarian (19)**
ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, FANCC, FANCM, FLCN, MLH1 (incl. 5'UTR), MSH2, MSH6, NBN, PALB2, PMS2, PTEN (incl. 5'UTR), RAD51D, RAD51C, STK11 TP53

☐ **Hereditary Cancer – Colorectal/Gastric Cancer (20)**
APC (incl. 5'UTR), BMPR1A, CDH1, CDK4, CHEK2, CTNNA1, EPCAM, GREM1, MLH1 (incl. 5'UTR), MSH2, MSH6, MUTYH, PMS2, POLD1, POLE, PTEN (incl. 5'UTR), SDHB, SMAD4, STK11, TP53

Hereditary Cancer – Individual selections

☐ APC (incl. 5'UTR)	☐ ATM	☐ BARD1	☐ BMPR1A	☐ BRCA1	☐ BRCA2	☐ BRIP1	☐ CDH1
☐ CDK4	☐ CDKN2A	☐ CHEK2	☐ CTNNA1	☐ EPCAM	☐ FANCC	☐ FANCM	☐ FLCN
☐ GREM1	☐ HOXB13	☐ MEN1	☐ MLH1 (incl. 5'UTR)	☐ MSH2	☐ MSH3	☐ MSH6	☐ MUTYH
☐ NBN	☐ NTHL1	☐ PALB2	☐ PMS2	☐ POLD1	☐ POLE	☐ PTEN (incl. 5'UTR)	☐ RAD51C
☐ RAD51D	☐ SDHB	☐ SMAD4	☐ STK11	☐ TP53			

Charcot Marie Tooth

☐ **Charcot Marie Tooth, HNPP – Comprehensive (34)**
AARS, AIFM1, DNAJB2, DYNC1H1, EGR2, FGD4, FIG4, GARS, GDAP1, GJB1, HSPB1, HSPB8, IGHMBP2, KIF1B, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MPZ, MTMR2, NDRG1, NEFL, PDK3, PMP22, PRPS1, PRX, RAB7A, SBF2, SH3TC2, SPTLC1, TRPV4, TTR

☐ **Charcot Marie Tooth – Type1 (10)**
EGR2, FIG4, GDAP1, GJB1, LITAF, MPZ, NEFL, PMP22, PRX, SH3TC2

☐ **Charcot Marie Tooth – Type2 (28)**
AARS, AIFM1, DNAJB2, DYNC1H1, FGD4, GARS, GDAP1, GJB1, HSPB1, HSPB8, IGHMBP2, KIF1B, LMNA, LRSAM1, MARS, MED25, MFN2, MPZ, MTMR2, NDRG1, NEFL, PDK3, PRPS1, RAB7A, SBF2, SPTLC1, TRPV4, TTR

Charcot Marie Tooth – Individual selections

☐ AARS	☐ AIFM1	☐ DNAJB2	☐ DYNC1H1	☐ EGR2	☐ FGD4	☐ FIG4	☐ GARS
☐ GDAP1	☐ GJB1	☐ HSPB1	☐ HSPB8	☐ IGHMBP2	☐ KIF1B	☐ LITAF	☐ LMNA
☐ LRSAM1	☐ MARS	☐ MED25	☐ MFN2	☐ MPZ	☐ MTMR2	☐ NDRG1	☐ NEFL
☐ PDK3	☐ PMP22	☐ PRPS1	☐ PRX	☐ RAB7A	☐ SBF2	☐ SH3TC2	☐ SPTLC1
☐ TRPV4	☐ TTR						

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Requisition for DNA Testing

Lysosomal Storage Disorders

☐ **Lysosomal Storage Disorders (50)**
AGA, ARSA, ARSB, ASAH1, CLN3, CLN5, CLN6, CLN8, CLN9, CTNS, CTSA, CTSD, CTSK, DNAJC5, FUCA1, GAA, GALC, GALNS, GBA, GLA, GLB1, GM2A, GNPTAB, GNPTG, GNS, GRN, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, LAMP2, LIPA, MAN2B1, MANBA, MCOLN1, MFSD8., NAGA, NAGLU, NEU1, NPC1, NPC2, PPT1, PSAP, SGSH, SLC17A5, SMPD1, SUMF1, TPP1

Lysosomal Storage Disorders – Individual selections

☐ AGA	☐ ARSA	☐ ARSB	☐ ASAH1	☐ CLN3	☐ CLN5	☐ CLN6	☐ CLN8
☐ CTNS	☐ CTSA	☐ CTSD	☐ CTSK	☐ DNAJC5	☐ FUCA1	☐ GAA	☐ GALC
☐ GALNS	☐ GBA	☐ GLA	☐ GLB1	☐ GM2A	☐ GNPTAB	☐ GNPTG	☐ GNS
☐ GRN	☐ GUSB	☐ HEXA	☐ HEXB	☐ HGSNAT	☐ HYAL1	☐ IDS	☐ IDUA
☐ LAMP2	☐ LIPA	☐ MAN2B1	☐ MANBA	☐ MCOLN1	☐ MFSD8	☐ NAGA	☐ NAGLU
☐ NEU1	☐ NPC1	☐ NPC2	☐ PPT1	☐ PSAP	☐ SGSH	☐ SLC17A5	☐ SMPD1
☐ SUMF1	☐ TPP1						

Urea Cycle Disorders

☐ **Urea Cycle Disorders Panel (13)**
ARG, ASL, ASS1, CA5A, CPS1, GLUD1, GLUL, NAGS, OTC, SLC25A2, SLC25A13, SLC25A15, SLC7A7

Urea Cycle Disorders – Individual selections

☐ ARG	☐ ASL	☐ ASS1	☐ CA5A	☐ CPS1	☐ GLUD1	☐ GLUL	☐ NAGS
☐ OTC	☐ SLC25A2	☐ SLC25A13	☐ SLC25A15	☐ SLC7A7			

Mitochondrial Genome and Depletion/Integrity Panel

☐ **Mitochondrial Genome and Depletion/Integrity Panel (56)**

Mitochondrial encoded genes: ATP6, ATP8, COX1, COX2, COX3, CYTB, ND1, ND2, ND3, ND4, ND4L, ND5, ND6, RNR1, RNR2, TRNA, TRNC, TRND, TRNE, TRNF, TRNG, TRNH, TRNI, TRNK, TRNL1, TRNL2, TRNM, TRNN, TRNP, TRNQ, TRNR, TRNS, TRNS2, TRNT, TRNV, TRNW, TRNY

Nuclear encoded genes: APTX, DGUOK, DNA2, FBXL4, GFER, MGME1, MPV17, OPA1, OPA3 (isoform A & B), POLG, POLG2, RRM2B, SLC25A4, SPG7 (isoform 1 & 2), SUCLA2, SUCLG1, TK2, TWNK, TYMP

Mitochondrial Genome and Depletion and Integrity – Individual selections

Mitochondrial encoded genes:

☐ ATP6	☐ ATP8	☐ COX1	☐ COX2	☐ COX3	☐ CYTB	☐ ND1	☐ ND2
☐ ND3	☐ ND4	☐ ND4L	☐ ND5	☐ ND6	☐ RNR1	☐ RNR2	☐ TRNA
☐ TRNC	☐ TRND	☐ TRNE	☐ TRNF	☐ TRNG	☐ TRNH	☐ TRNI	☐ TRNK
☐ TRNL1	☐ TRNL2	☐ TRNM	☐ TRNN	☐ TRNP	☐ TRNQ	☐ TRNR	☐ TRNS
☐ TRNS2	☐ TRNT	☐ TRNV	☐ TRNW	☐ TRNY			

Nuclear encoded genes:

☐ APTX	☐ DGUOK	☐ DNA2	☐ FBXL4	☐ GFER	☐ MGME1	☐ MPV17	☐ OPA1
☐ OPA3 (isoform A & B)	☐ POLG	☐ POLG2	☐ RRM2B	☐ SLC25A4	☐ SPG7 (isoform A & B)	☐ SUCLA2	☐ SUCLG1
☐ TK2	☐ TWNK (C10 or f2)	☐ TYMP					

Cancer Hotspot Panel v2 (Ion Ampliseq)

(Tumour FFPE samples only)

☐ ERAS (KRAS, NRAS) ☐ BRAF ☐ EGFR ☐ FULL screen (2800 COSMIC mutations, research only)

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Single Genes by NGS (includes deletion/duplication analysis)

- ☐ **ACADM** – Medium Chain Acyl CoA Dehydrogenase (MCAD)
- ☐ **GJB2 (CX26) / GJB6 (CX30)** – Recessive Deafness
- ☐ **MECP2** – RETT Syndrome
- ☐ **MEN1** – Multiple Endocrine Neoplasia Type 1
- ☐ **NOTCH3** – CADASIL
- ☐ **TP53** – Li-Fraumeni Syndrome
- ☐ **RET** – Multiple Endocrine Neoplasia Type 2 / FMTC
- ☐ **SCN4A** – Paramyotonia Congenita
- ☐ **SPTLC1** – Hereditary Sensory Neuropathy
- ☐ **NPC1** – Niemann-Pick Disease 1
- ☐ **NPC2** – Niemann-Pick Disease 2
- ☐ **ARSA** – Metachromatic Leukodystrophy
- ☐ **CTNS** – Cystinosis
- ☐ **CLN2** – Batten Disease
- ☐ **CLN3** – Batten Disease
- ☐ **OTC** – Ornithine Transcarbamylase
- ☐ **ARG1** – Arginase Deficiency

Targeted Assays

- ☐ **BCR-ABL** – CML minimal residual disease (quantitative RT-PCR)
- ☐ **B-Cell Clonality (Lymphoma)** (fragment analysis)
- ☐ **T-Cell Clonality (Lymphoma)** (fragment analysis)
- ☐ **CFTR – Cystic Fibrosis -70 mutation screen** (Mass Array)
- ☐ **JAK2-p.V617F** – Myeloproliferative disorders (quantitative PCR)
- ☐ **F2** – Prothrombin G20210A (F2:c.97G>A) (Mass Array)
- ☐ **F5** – Factor V Leiden (F5:p.R534Q) (Mass Array)
- ☐ **HFE** – Hemochromatosis p.C282Y and p.H63D (Mass Array)
- ☐ **MCC/Identity testing** – Maternal cell contamination/ tissue contamination studies (fragment analysis)

NGS Panels for Research Purposes Only (includes deletion/duplication analysis)

Epilepsy (Available for research purposes only)

- ☐ **Epilepsy – Comprehensive (69)**
  ALDH7A1, AMT, ARX, ASAH1, ATP1A2, ATP1A3, CDKL5, CERS1, CHD2, CHRNA7, CNTNAP2, CSTB, DNM1, DOCK7, EPM2A, FOLR1, FOXG1, GAMT, GATM, GLDC, GOSR2, GRIN2A, GRIN2B, HCN1, KCNC1, KCNJ10, KCNJ11, KCNQ2, KCNQ3, KCNT1, KCTD7, LMNB2, MBD5, MECP2, MEF2C, MOCS1, NECAP1, NEU1, NHLRC1, NRXN1, PCDH19, PHGDH, PLCB1, PNKP, PNPO, POLG, PRICKLE2, PRRT2, PSAT1, PSPH, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC2A1, SLC6A8, SLC9A6, SPTAN1, STXBP1, SUOX, SYNGAP1, TBC1D24, TCF4, TSC1, TSC2, UBE3A, ZEB2
- ☐ **Epilepsy – Management Impact (16)**
  ALDH7A1, AMT, FOLR1, GAMT, GATM, GLDC, MOCS1, PHGDH, PNPO, POLG, PSAT1, PSPH' SCN1A, SLC2A1, SLC6A8, SUOX
- ☐ **Epilepsy – Progressive myoclonic (12)**
  ASAH1, CERS1, CSTB, EPM2A, GOSR2, KCNC1, KCTD7, LMNB2, NEU1, NHLRC1, PRICKLE2, SCARB2
- ☐ **Epilepsy – Severe Phenotype (19)**
  ARX, CDKL5, CHD2, DNM1, DOCK7, GRIN2A, GRIN2B, HCN1, KCNQ2, MECP2, MEF2C, NECAP1, PCDH19, SCN1B, SCN2A, SCN8A, SPTAN1, STXBP1, SYNGAP1
- ☐ **Epilepsy – Syndromes of Infancy (22)**
  ATP1A2, ATP1A3, CHRNA7, CNTNAP2, FOXG1, KCNJ10, KCNJ11, KCNQ3, KCNT1, MBD5, NRXN1, PLCB1, PNKP, PRRT2, SCN9A, SLC9A6, TBC1D24, TCF4, TSC1, TSC2, UBE3A, ZEB2

Epilepsy – Individual selections

☐ ALDH7A1	☐ AMT	☐ ARX	☐ ASAH1	☐ ATP1A2	☐ ATP1A3	☐ CDKL5	☐ CERS1
☐ CHD2	☐ CHRNA7	☐ CNTNAP2	☐ CSTB	☐ DNM1	☐ DOCK7	☐ EPM2A	☐ FOLR1
☐ FOXG1	☐ GAMT	☐ GATM	☐ GLDC	☐ GOSR2	☐ GRIN2A	☐ GRIN2B	☐ HCN1
☐ KCNC1	☐ KCNJ10	☐ KCNJ11	☐ KCNQ2	☐ KCNQ3	☐ KCNT1	☐ KCTD7	☐ LMNB2
☐ MBD5	☐ MECP2	☐ MEF2C	☐ MOCS1	☐ NECAP1	☐ NEU1	☐ NHLRC1	☐ NRXN1
☐ PCDH19	☐ PHGDH	☐ PLCB1	☐ PNKP	☐ PNPO	☐ POLG	☐ PRICKLE2	☐ PRRT2
☐ PSAT1	☐ PSPH	☐ SCARB2	☐ SCN1A	☐ SCN1B	☐ SCN2A	☐ SCN8A	☐ SCN9A
☐ SLC2A1	☐ SLC6A8	☐ SLC9A6	☐ SPTAN1	☐ STXBP1	☐ SUOX	☐ SYNGAP1	☐ TBC1D24
☐ TCF4	☐ TSC1	☐ TSC2	☐ UBE3A	☐ ZEB2			

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Hyperferritinemia (Available for research purposes only)

☐ **Hyperferritinemia Panel (15)**
ALAS2, B2M, CDAN1, CP, FTH1, FTL, HAMP, HFE, HFE2, SEC23B, SLC25A38, SLC40A1, STEAP3, TF, TFR2

Hyperferritinemia – Individual selections

☐ ALAS2	☐ B2M	☐ CDAN1	☐ CP	☐ FTH1	☐ FTL	☐ HAMP	☐ HFE
☐ HFE2	☐ SEC23B	☐ SLC25A38	☐ SLC40A1	☐ STEAP3	☐ TF	☐ TFR2	

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