

London Health Sciences Centre Molecular Genetics Laboratories
Requisition for DNA Testing

DACC002 REV 20170815



Molecular Genetics Laboratory

Victoria Hospital, Room B10-123A,
800 Commissioners Road East, Ph: (519) 685-8122
London Ontario, N6A 5W9 Fax: (519) 685-8279

Reason for Referral:

- ☐ Diagnostic Testing –
 - ☐ Affected
 - ☐ Unaffected
 - ☐ Carrier testing/Known Family Mutation
Name of Index Case in the Family: _____
D.O.B. __yy__ / __mm__ / __dd__
(include copy of report)
Relationship to this patient: _____
- Gene: _____ Mutation: _____
- RefSeq:NM _____

- ☐ Prenatal Diagnosis
- ☐ DNA Banking
- ☐ RNA Banking
- ☐ Referral to an outside Laboratory – must specify lab:

AUTHORIZED SIGNATURE IS REQUIRED

INCOMPLETE REQUESTS WILL BE BANKED

Patient's Name: _____

Birthdate: __yyyy__ / __mm__ / __dd__

Sex: _____

Patient's Address: _____

Health Card #: _____

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): _____

____yy__ / __mm__ / __dd__

AUTHORIZED SIGNATURE (required):

Referring physician: (Please Print)

Name: _____

Address: _____

Tel () _____ Fax () _____
e-mail address: _____

Signature (required) _____

CC report to:

Name: _____

Address: _____

Tel () _____ Fax () _____

Sample Collection: Date Drawn: __yyyy__ / __mm__ / __dd__

- ☐ EDTA blood (lavender top)(min. 2 ml 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:

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Patient's Name: _____

Birthdate: __yy__ / __mm__ / __dd__

NGS Panels (includes deletion/duplication analysis)

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

☐ Hereditary Cancer - Comprehensive (30)

APC (incl. 5'UTR), ATM, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, CTNNA1, EPCAM, GREM1, MLH1 (incl. 5'UTR), MSH2, MSH6, MUTYH, NBN, 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, 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	☐ GREM1	☐ MLH1 (incl. 5'UTR)	☐ MSH2
☐ MSH6	☐ MUTYH	☐ NBN	☐ 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						

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 (isoform1 & 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 (isoform1 & 2), ☐ SUCLA2, ☐ SUCLG1, ☐ TK2, ☐ TWNK (C10orf2), ☐ TYMP

Cancer Hotspot Panel v2 (IonAmpliseq) (Tumour FFPE samples only)

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

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			

Lysosomal Storage Disorders *(Available for research purposes only)*

☐ **Lysosomal Storage Disorders (50)**

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

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 *(Available for research purposes only)*

☐ **Urea Cycle Disorders Panel (13)**

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

Urea Cycle Disorders- Individual selections

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

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