

Requisition for DNA Testing

Patient Identifier:

Please use the dedicated requisitions for the following test requests:

- Hereditary Cancer Panel (HCP)
- Epilepsy
- Atypical Hemolytic Uremic Syndrome
- Hematologic Oncology
- Biochemical Genetics

Requisitions can be found at <https://www.lhsc.on.ca/palm/forms/requisitions.html#moldx>

Sample Requirements can be found at <https://www.lhsc.on.ca/palm/molecular/specimen.html#main-content>

NGS PANELS (INCLUDES DELETION/DUPLICATION ANALYSIS)

Charcot Marie Tooth

Charcot Marie Tooth, HNPP - Comprehensive (88)

AARS, ABHD12, AIFM1, ARHGEF10, ARHGEF28, ATP1A1, ATP7A, BAG3, BSCL2, C1orf194, CNTNAP1, DCTN1, DCTN2, DGAT2, DHTKD1, DNAJB2, DNMT2, DNMT1, DRP2, DYNC1H1, EGR2, FBLN5, FGD4, FIG4, GARS, GDAP1, GJB1, GNB4, HARS, HINT1, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, JAG1, KARS, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MARS, MCM3AP, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, NAGLU, NDRG1, NEFH, NEFL, PDK3, PDKX, PLEKHG5, PMP2, PMP22, PNKP, PRPS1, PRX, PTRH2, RAB7A, RAB7A, SBF1, SBF2, SCO2, SELRC1, SEPT9, SETX, SGPL1, SH3TC2, SIGMAR1, SLC12A6, SLC9A3R1, SORD, SPG11, SPTLC1, SURF1, TFG, TRIM2, TRPV4, TTR, VCP, VRK1, WARS, YARS

Charcot Marie Tooth - Individual selections (Individual selections available for out-of-province requests only except SPTLC1 and TTR)

AARS	ABHD12	AIFM1	ARHGEF10	ARHGEF28	ATP1A1	ATP7A	BAG3
BSCL2	C1orf194	CNTNAP1	DCTN1	DCTN2	DGAT2	DHTKD1	DNAJB2
DNMT2	DNMT1	DRP2	DYNC1H1	EGR2	FBLN5	FGD4	FIG4
GARS	GDAP1	GJB1	GNB4	HARS	HINT1	HSPB1	HSPB3
HSPB8	IGHMBP2	INF2	JAG1	KARS	KIF1B	KIF5A	LITAF
LMNA	LRSAM1	MARS	MCM3AP	MFN2	MME	MORC2	MPV17
MPZ	MTMR2	NAGLU	NDRG1	NEFH	NEFL	PDK3	PDKX
PLEKHG5	PMP2	PMP22	PNKP	PRPS1	PRX	PTRH2	RAB7A
SBF1	SBF2	SCO2	SELRC1	SEPT9	SETX	SGPL1	SH3TC2
SIGMAR1	SLC12A6	SLC9A3R1	SORD	SPG11	SPTLC1	SURF1	TFG
TRIM2	TRPV4	TTR	VCP	VRK1	WARS	YARS	

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 (Individual selections available for out-of-province requests only)

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					

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Requisition for DNA Testing

Patient Identifier:

Lysosomal Storage Disorders

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

Urea Cycle Disorders Panel (13)

AGR1, ASL, ASS1, CA5A, CPS1, GLUD1, GLUL, NAGS, OTC, SLC25A2, SLC25A13, SLC25A15, SLC7A7

Urea Cycle Disorders - Individual selections

ARG1	ASL	ASS1	CA5A	CPS1	GLUD1	GLUL	NAGS
OTC	SLC25A2	SLC25A13	SLC25A15	SLC7A7			

SINGLE GENES BY NGS (INCLUDES DELETION/DUPLICATION ANALYSIS)

ACADM - Medium Chain Acyl CoA Dehydrogenase (MCAD)

GJB2 (CX26) / GJB6 (CX30) - Recessive Deafness

MECP2 - RETT Syndrome

NOTCH3 - CADASIL

SCN4A - Paramyotonia Congenita

SPTLC1 - Hereditary Sensory Neuropathy

TTR - Amyloidosis

NPC (NPC1/NPC2) - Niemann-Pick Disease

ARG1 - Arginase Deficiency

ARSA - Metachromatic Leukodystrophy

CTNS - Cystinosis

CLN2-TPP1 - Batten Disease

CLN3 - Batten Disease

OTC - Ornithine Transcarbamylase

TARGETED ASSAYS

CFTR - Cystic Fibrosis -70 mutation screen (Mass Array)

DPYD Genotyping - c.1905+1G>A, c.1679T>G, c.2846A>T and c.1236G>A / c.1129-5923C>G (Mass Array)

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)