Trillium Health Partners
Better Together

Genetics Laboratory - Credit Valley Hospital
2200 Eglinton Ave West, Room 2H144
Mississauga, ON L5M 2N1
Tel: (905) 813-1100 ext. 6288
Fax: (905) 813-3854

CONSTITUTIONAL / GERMLINE / HEREDITARY GENETIC TESTING REQUISITION

~ Visit community collection lab for blood draw. ~

PATIENT DEMOGRAPHICS			
Last Name:		Health Card #:	Version Code:
First Name:		Date of Birth (DD/MM/YYYY):	
Address:		Legal Sex: ☐ Male ☐ Female ☐ Non-Binary	
City:		☐ Unknown ☐ X	
Province:		Phone Number:	
Postal Code:		Email Address:	

REFERRING PROVIDER		**COPIES TO**	
Last Name:		Last Name:	
First Name:		First Name:	
CPSO #:	OHIP Billing #:	CPSO #:	OHIP Billing #:
Address:		Address:	
City/Prov:	Postal Code:	City/Prov:	Postal Code:
Phone:	Fax:	Phone:	Fax:
Signature (required):	Date:		

SPECIMEN REQUIREMENTS

SPECIMEN COLLECTION DATE (DD/MM/YYYY): _____ TIME (HH:MM): _____

Molecular	**Cytogenetics and Microarray**
☐ Blood *(5-10mL EDTA, room temp) (1mL for neonates)*	☐ Blood *(3mL NaHep; room temp) (1mL for neonates)*
☐ Extracted DNA *Source: _____ (2µg total; min. 70ng/uL)*	
☐ Amniotic Fluid *Gestational Age:*	
☐ Cultured Amniocytes *Gestational Age:*	
☐ Products of Conception *(No Formalin; Placenta not accepted) Gestational Age (if known):*	
☐ Skin Biopsy *Source:*	
☐ Tissue (Fresh) *Source:*	

CLINICAL INFORMATION	**FAMILY HISTORY**
☐ Pregnant? LMP: _____ ☐ Egg Donor?	Index Case: ☐ Y ☐ N
TESTING STATUS	Relationship to Index case: _____
☐ Routine	Name/MRN/DOB of Index Case:_____
☐ Expedited Reason *(required)*: _____ Surgery Date *(if applicable)*: _____	Relevant Personal/Family History: *Please attach family reports*

THP LAB USE ONLY	
Date / Time Received:_____	
Specimen Details:_____ Initials:_____	FOR THP LABEL ONLY
Comments: _____	
RQ#:_____	

Genetics Laboratory - Credit Valley Hospital
2200 Eglinton Ave West, Room 2H144
Mississauga, ON L5M 2N1
Tel: (905) 813-1100 ext. 6288
Fax: (905) 813-3854

CYTOGENETICS

REASON FOR REFERRAL for Karyotype Analysis (G-banding)	
☐ Ambiguous genitalia	☐ Stillbirth
☐ Amenorrhea	☐ Neonatal death
☐ Turner syndrome	☐ Ring Chromosome 20 study (epilepsy patients)
☐ Klinefelter syndrome	☐ Family History of Chromosomal Rearrangement
☐ Infertility	*Specify:* _____
☐ Recurrent Miscarriages (≥3)	*Please attach family reports*

MOLECULAR DIAGNOSTIC TESTING

ANEUPLOIDY TESTING / SEX CHROMOSOME DETERMINATION

☐ Ambiguous genitalia / Sex Chromosome Determination

☐ Fetal Demise / Products of Conception

☐ Postnatal *Specify Chromosome of interest (13, 18, 21, X, Y):* _____

☐ Prenatal

☐ Maternal Cell Contamination (MCC) studies

BLOOD DISORDERS

☐ Hemochromatosis (*HFE*) Common Variants

☐ Thrombophilia Common Variant Panel (Factor II, Factor V)

COPD or LIVER DISEASE

☐ Alpha-1 Antitrypsin *SERPINA1* (A1AT) Whole Gene Sequencing ☐ Lung Disease ☐ Liver Disease ☐ Asymptomatic

Patient's Serum A1AT Activity: _____g/L ☐ Carrier Testing ☐ Other:_____

FAMILIAL HYPERCHOLESTEROLEMIA

☐ **Carrier Testing / Known Family Mutation of Familial Hypercholesterolemia (FH)** *(DNA not accepted; EDTA Blood only)*

(Send copy of report if testing not completed at THP)

THP Lab Report #:
Name of Index Case in the Family:
Relationship to this Patient:

Variant Details:

Gene:
Variant Identified:
Reference Sequence NM#:

☐ **Familial Hypercholesterolemia Panel** *(DNA not accepted; EDTA Blood only)*

Individual must meet one or more of the following. Select all that apply:

☐ 1. Confirmed FH disease-causing pathogenic/likely pathogenic variant in a close (1st or 2nd degree) blood relative.

☐ 2. Extremely high LDL-cholesterol level of ≥8.5 mmol/L at any age.

☐ 3. High LDL with additional features: Personal history of untreated elevated LDL cholesterol level (not due to secondary causes). **Specify: _____ mmol/L**

- ☐ Untreated LDL-cholesterol level ≥5.0 mmol/L for age 40 years and over
- ☐ Untreated LDL-cholesterol level ≥4.5 mmol/L for age 18 to 39 years
- ☐ Untreated LDL-cholesterol level ≥3.5 mmol/L for age under 18 years

AND at least one of the following:

- ☐ Tendon xanthomas and/or corneal arcus in proband
- ☐ First-degree relative (FDR) with high LDL-cholesterol level (not due to secondary causes)
- ☐ Proband or FDR with early onset ASCVD (men under 55 years; women under 65 years)
- ☐ Limited family history information (e.g. adopted)

☐ 4. Clinical judgement: Criteria above not met, but suspicion remains. Describe:_____

If baseline/untreated LDL cholesterol is unknown, an imputed level can be derived using the CardioRiskCalculator:
https://www.circl.ubc.ca/cardiorisk-calculator.html

FH Sequence and copy number analysis of the following genes: *ABCG8, ABCG5, APOE, APOB , LDLR, LDLRAP1, LIPA, PCSK9*

Genetics Laboratory - Credit Valley Hospital
2200 Eglinton Ave West, Room 2H144
Mississauga, ON L5M 2N1
Tel: (905) 813-1100 ext. 6288
Fax: (905) 813-3854

MOLECULAR FAMILIAL HEREDITARY CANCER VARIANT TESTING

FAMILIAL VARIANT	
Gene: Variant: Reference Sequence: NM#	Index Case (Name & MRN / Date of Birth): _____ Relationship to this Patient: _____ THP Report Number: _____ *If index case testing performed elsewhere, please attach a copy of the original result (all pages).*
Gene: Variant: Reference Sequence: NM#	

MOLECULAR HEREDITARY CANCER PREDISPOSITION NGS PANELS

Only available for oncology-initiated testing for hereditary cancer predisposition. Select the appropriate risk category.

Breast Cancer[1] (HBOP1)		**Category** *(Lab Use Only)*
☐	Personal history of breast cancer ≤45 years of age.	1
☐	Personal history of breast cancer ≤50 years of age with limited family structure (ie. Adoption; few close relatives assigned female at birth).	2
☐	Personal history of breast cancer ≤50 years of age with a second primary breast cancer.	3
☐	Personal history of triple negative invasive breast cancer ≤60 years of age.	4
☐	Personal history of male breast cancer at any age.	5
☐	Personal history of breast cancer at any age where germline status may determine eligibility for approved targeted therapy, regardless of other hereditary cancer syndrome risk factors.	General-3
Ovarian Cancer[1] (HBOP1)		
☐	Personal history of invasive epithelial ovarian cancer (any grade), epithelial fallopian tube or peritoneal cancer at any age.	6
Prostate Cancer[1] (HBOP1)		
☐	Personal history of metastatic prostate cancer.	1
☐	Documented personal history of high risk, locally advanced, prostate cancer. High-risk prostate cancer can be confirmed with evidence of one or more of the following features: T3 or higher staging; Grade Group 4 or 5 (Gleason Score 8 to 10), Lymph node involvement, PSA ≥20.	2
Pancreatic Cancer[2] (PANC1)		
☐	Personal history of pancreatic adenocarcinoma regardless of age or family history.	1

Regions of Interest:
[1] *ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53*
[2] *ATM, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, STK11, TP53*

Gene panels include both sequencing and deletion/duplication analysis. (Exception: GREM1 and EPCAM analysis for large deletion/duplication only).

Partner Follow-Up Full Gene Testing
Gene of Interest:

AVAILABLE TESTS FOR *THP GENETICS CLINIC* ONLY

DNA BANKING	**MATERNAL Region-Specific Analysis (RSA) Studies**
☐ Short-Term Storage (2 years) ☐ Long-Term Storage (25 years)	☐ Microarray RSA *Specify Chromosome(s) of Interest:* _____

Trillium Health Partners
Better Together

Genetics Laboratory - Credit Valley Hospital
2200 Eglinton Ave West, Room 2H144
Mississauga, ON L5M 2N1
Tel: (905) 813-1100 ext. 6288
Fax: (905) 813-3854

MICROARRAY

DIAGNOSTIC TESTING
The 'Phenotypic Description' section below MUST be completed for accurate interpretation.

☐ Developmental Delay
☐ Intellectual Disability
☐ Two or more congenital anomalies

☐ Uniparental Disomy (UPD) Region-Specific Analysis (RSA) Studies
Specify Chromosome(s) of Interest: _____
Note: BOTH parents and proband samples are required for UPD studies

FAMILY HISTORY

☐ Parents ≥ 3 miscarriages
☐ Consanguinity

List Family Health Conditions (describe relationship with proband): _____

PHENOTYPIC DESCRIPTION (Clinical Symptoms)

Behavior, Cognition and Development
☐ Global development delay
☐ Gross motor delay
☐ Fine motor delay
☐ Language delay
☐ Learning disability
☐ Intellectual disability
  ☐ Mild
  ☐ Moderate
  ☐ Severe
☐ Attention deficit hyperactivity disorder
☐ Autism Spectrum Disorder
☐ Psychiatric disorders
☐ Other: _____

Neurological
☐ Hypotonia
☐ Seizures
☐ Ataxia
☐ Dystonia
☐ Chorea
☐ Spasticity
☐ Cerebral palsy
☐ Neural tube defect
☐ Central Nervous System Abnormality *specify below*
☐ Other: _____

Growth Parameters

	Less than	Greater than
Weight for age:	☐ 3rd %	☐ 97th %
Height for age:	☐ 3rd %	☐ 97th %

☐ Hemihypertrophy
☐ Other: _____

Cardiac
☐ Atrial septal defect
☐ Ventricular septal defect
☐ Atrioventricular canal defect
☐ Coarctation of aorta
☐ Tetralogy of Fallot
☐ Other: _____

Craniofacial
☐ Craniosynostosis
☐ Cleft lip
☐ Cleft palate
☐ Facial dysmorphism
☐ Macrocephaly
☐ Microcephaly
☐ Micrognathia
☐ Retrognathia
☐ Other: _____

Eye Defects
☐ Blindness
☐ Coloboma
☐ Epicanthus
☐ Hypertelorism
☐ Eyelid abnormality *specify below*
☐ Other: _____

Ear Defects
☐ Deafness
☐ Preauricular
☐ Low-set ears
☐ Outer ear abnormality *specify below*
☐ Inner ear abnormality *specify below*
☐ Other: _____

Genitourinary
☐ Kidney malformation *specify below*
☐ Hydronephrosis
☐ Ambiguous genitalia
☐ Hypospadias
☐ Cryptorchidism
☐ Other: _____

Respiratory
☐ Diaphragmatic hernia
☐ Lung abnormality *specify below*
☐ Other: _____

Musculoskeletal
☐ Upper limb abnormality
☐ Lower limb abnormality
☐ Camptodactyly
  ☐ Fingers ☐ Toes
☐ Syndactyly
  ☐ Fingers ☐ Toes
☐ Polydactyly
  ☐ Fingers ☐ Toes
☐ Preaxial
☐ Postaxial
☐ Oligodactyly
☐ Clinodactyly
☐ Contractures
☐ Scoliosis
☐ Vertebral Anomaly
☐ Club foot
☐ Other: _____

Gastrointestinal
☐ Esophageal atresia
☐ Tracheoesophageal fistula
☐ Gastroschisis
☐ Omphalocele
☐ Pyloric stenosis
☐ Other: _____

Cutaneous
☐ Hyperpigmentation
☐ Hypopigmentation
☐ Other: _____

PRENATAL AND PERINATAL HISTORY

☐ Intrauterine growth restriction
☐ Oligohydramnios
☐ Polyhydramnios
☐ Premature birth
☐ EFTS/NIPT Screen Positive *Result:* _____
☐ Fetal soft markers in obstetric ultrasound *Specify:* _____
☐ Fetal structural abnormality

FOLLOW-UP FAMILY TESTING

Proband Report Lab #: _____ Family ID #: _____ Relationship to Proband: _____
Indicate if a family member will not be providing bloodwork to prevent test delay: _____
Follow-Up test methodology will be performed based on lab protocol.