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		Approved By:	**Brendan Mullen, Dr**

Advanced Molecular Diagnostics

The Advanced Molecular Diagnostic Laboratory provides molecular genetic testing for the purpose of identifying hereditary and sporadic genetic disorders. It serves as an international reference centre for molecular diagnostics. The laboratory is accredited by the Ontario Laboratory Accreditation (OLA) program, Quality Management Program – Laboratory Services and participates regularly in molecular genetic proficiency testing offered by a number of international providers including, the College of American Pathologists (CAP). Our mission is to provide the highest grade clinical testing service and to translate research findings into clinical practice using new technologies.

Testing is available for a large number of genetic disorders including specialty areas of hereditary breast, ovarian and colon cancers. As the number of disorders with known etiology increases new tests are continually being added to the test menu. The laboratory strives to meet this demand through introduction of new technologies and in-house development of cutting edge diagnostic tests. The laboratory is dedicated to teaching and education and to providing molecular training. Testing is performed by a team of experienced medical laboratory technologists with certification in molecular diagnostics and the laboratory is overseen by clinical molecular geneticists board certified by the American and Canadian Colleges of Medical Genetics.

Contact Details

Advanced Molecular Genetics
Mount Sinai Hospital
600 University Ave
6th Floor
Room 6-306
Toronto, Ontario
M5G 1X5

Telephone
416-586-4800 x**5974**

Reports: 416-586-4457
Fax: 416-586-5140
MolecularLab@mtsinai.on.ca

Hours of Operation
Monday – Friday
08:00 to 16:00

Certification:

The laboratory directors are certified in Clinical Molecular Genetics specialty by the Canadian College of Medical Geneticists (CCMG) or by the American Board of Medical Geneticists (ABMG).

Accreditation:

The molecular genetics laboratory at the Mount Sinai Hospital operates in compliance with the OLA, CAP, CCMG, ACMG and the Clinical & Laboratory Standards Institute (CLSI) guidelines.
The laboratory is regularly inspected and accredited by various regulatory bodies including Accreditation Canada.

The laboratory participates in external proficiency testing program, the results of which are available to health care providers upon request.

Referral:

The Molecular Genetics Laboratory accepts referrals from healthcare providers only. Patient requisitions must be signed by the referring health-care provider.

Consultation:

Administration		
Name	**Room Number**	**Extension**
Sharon Crafter, Manager, Diagnostic Medical Genetics	6-500	416.586.4800 x6379

Scientific Directors		
Name	**Room Number**	**Extension**
Dr. Jordan Lerner-Ellis, Director & Head, Advanced Molecular Diagnostics	OPG 8-400	416.586.4800 x4488
Dr. George Charames, Director & Head, Advanced Molecular Diagnostics	OPG 8-400	416.586.4800 x7733
Dr. Aaron Pollett, Provincial Lead, Pathology and Laboratory Medicine Program, Cancer Care Ontario; Co-Director, Division of Diagnostic Medical Genetics	6-500	416-586-4800 x6452

Consultation with a molecular certified individual is available for health care providers. Additional information on laboratory procedures, available tests, specimen requirements, and interpretation of results may be obtained by telephoning the laboratory.

Requisitions:

MOLECULAR PATHOLOGY REQUISITION is available through the **intranet:**

http://info2/departments/pathlabmed/

or internet:

https://www.mountsinai.on.ca/care/pathology/laboratory-forms-and-requisitions/molecular-pathology-requisition-fillable-aoda.pdf

Precautions:

Specimens are collected using Body Substance Precautions
Dispose of consumables and supplies according to appropriate policies and protocols.

Please refer to specimen requirements and shipping instructions below before sending specimens.

Labeling of Specimens:

The laboratory reserves the right to refuse improperly labeled specimens.
Specimens collected for laboratory testing must be labeled with a permanently attached label in the form of a computer generated label or written legibly in indelible ink.

Containers

All specimens for molecular testing must be labelled according to Ontario Laboratory Accreditation requirements.
All specimen containers require (see summary below in Table 1):

- Patient's full first and last name and
- at least one unique identifier such as Provincial Health Card Number, MRN, Clinic #, Chart #)

The patient's full name and unique identifier on the specimen container must completely match the patient's full name and unique identifier on the requisition.

Glass Slides

All glass slides (stained and unstained sections) require:

- Patient's full first and last name and
- at least one unique identifier such as accession number, Provincial Health Card Number, MRN, Clinic #, Chart #)

Note: All glass slides must be labelled using lead pencil or indelible ink labels. The actual slide must be labelled, not the slide container.

The patient's full name and unique identifier on the glass slide must completely match the patient's full name and unique identifier on the requisition.

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SAMPLE TYPE	STANDARD REQUIREMENTS			
	NAME IDENTIFIER	**UNIQUE IDENTIFIER**	**OTHER**	**SAMPLE SPECIFIC**
BLOOD	**Patient's full first and last name** (OR coded name for confidential patients OR temporary ID for unknown patients)	At least one (preferably two) of the following assigned identifiers (in order of priority): 1. Provincial Health card number 2. Personal Identification Number (e.g. Federal, Military, RCMP, Refugee, immigration, Passport, etc.) 3. Facility Assigned Number (e.g. hospital # / Clinic # / Unit # / Account # / Accession #) 4. Date of Birth (DOB)	Collection date and time	Exact source for each sample submitted (not abbreviated to just a corresponding number/letter) as deemed necessary for accurate test reporting
PARAFFIN BLOCK	**Patient's full first and last name** (OR coded name for confidential patients OR temporary ID for unknown patients)	Facility Assigned Number (e.g. hospital # / Clinic # / Unit # / Account # / Accession #)	1. Tissue type/specimen source 2. Submitting Hospital/Lab Identification (name & address)	Exact source for each sample submitted (not abbreviated to just a corresponding number/letter) as deemed necessary for accurate test reporting
DNA	**Patient's full first and last name** (OR coded name for confidential patients OR temporary ID for unknown patients)	Facility Assigned Number (e.g. hospital # / Clinic # / Unit # / Account # / Accession #)	1. Tissue type/specimen source 2. Submitting Hospital/Lab identification (name & address)	

Summary of labeling requirements for Sample Containers & Blocks

Labeling - Requisition Requirements:

Requisitions received with specimens by the Advanced Molecular Diagnostics laboratory **MUST** include:

1. Patient's first & last name
2. Date of birth
3. Gender
4. One of the following:
    1. Provincial Health Card Number (PHN) & Province
    2. Hospital Patient Identification Number (MRN)
5. Full name of the referring physician and the CPSO number
6. Complete address for the referring physician including email and fax number
7. Full name of the genetic counsellor and contact information
8. Collection/procedure time, collection date and the name of the person who collected the specimen
9. Relevant clinical / family history, treatment, medication, special instructions, disease status
10. Location of specimen collection
11. Specimen type and origin – source/site, surgical number
12. Collection method
13. Reason for referral
14. Race & Ethnicity
15. IHC results (if applicable)

NOTE: A completed requisition must accompany each case.

Priority Definitions

Routine cases within each testing area are completed in chronological order. Samples received are monitored by the Senior Technologist and Lab Manager and samples are assigned for testing to a Technologist for completion within the allotted turn around time. Expedited cases move to the front of the queue in sequence with other expedited samples.

1. A case is given **EXPEDITED** status (indicated in the database):
   a) When a result is required for surgical decision making and/or chemotherapy. This must be indicated on the requisition form.
   b) At the Laboratory Director's discretion
2. All other samples are given ROUTINE "R" status.

Note: Verbal or written requests by the physician or genetic counsellor to have the status upgraded to EXPEDITED are accepted providing the test result is required for the criteria listed in 1a and 1b.

Acceptance Criteria for Specimens/Test Requests

The Advanced Molecular Diagnostics laboratory recognizes that the quality and accuracy of laboratory results can only be assured when specimens and test requests meet specific criteria for collection, labeling, and integrity. This policy applies to all specimens and test requests submitted to the laboratory.

NOTE: The laboratory only accepts isolated or extracted nucleic acids for which extraction or isolation is performed in an appropriately qualified laboratory. E.g. CAP or IQMH accredited.

When a Specimen does not meet the Criteria for Acceptance

Specimens and requisitions with major discrepancies/ deficiencies **(or samples where the information is illegible) may be rejected.** The submitting facility/physician will be contacted.

Major discrepancies/deficiencies may include:

- Specimen container or slide is unlabeled
- Patient's first or last name is missing, incomplete or significantly misspelled on the specimen container/slide or requisition
- Patient's first or last name is completely different between specimen container/slide and requisition
- Unique identifier is missing, incomplete or incorrect on the specimen container/slide or requisition
- Unique identifier is completely different between specimen container/slide and requisition
- Specimen container leaked in transit
- Glass slide is received broken beyond repair
- Requisition is received without a corresponding specimen container/slide

Irretrievable specimens and requisitions with major or discrepancies/deficiencies will not be rejected.

Reasonable effort will be taken to resolve any discrepancies/ deficiencies.

Specific Specimen Collection Instructions

TEST	TYPE OF SPECIMEN	VOLUME REQUIRED	CONTAINER	STORAGE & TRANSPORT	SPECIAL INSTRUCTIONS
BREAST / COLON CANCER: **Expanded Breast and Ovarian Cancer Panel (20 genes):** *ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, FANCC, MLH1, MSH2, MSH6, NBN, PALB2, PMS2*, PTEN, RAD51C, RAD51D, STK11, TP53*	Blood	10 ml blood	EDTA tube (lavender top)	Room Temperature	Sample must be received within 24 – 48 hours. Overnight preferred.
Breast Cancer Surgical Panel (4 genes): *BRCA1, BRCA2, PALB2* **High Risk Hereditary Cancer Panel (16 genes):** *APC, ATM, BRCA1, BRCA2, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, PALB2, PMS2*, PTEN, STK11, TP53* **Lynch and Polyposis Panel (7 genes):** *APC, EPCAM, MLH1, MSH2, MSH6, MUTYH, PMS2** **Lynch Syndrome Panel (5 genes):** *EPCAM, MLH1, MSH2, MSH6, PMS2** **Polyposis Panel (2 genes):** *APC, MUTYH* **Gastrointestinal Cancer Panel (15 genes):** *APC, BMPR1A, CDH1, EPCAM, MLH1, MSH2, MSH6, MUTYH, PMS2*, POLE, POLD1, PTEN, SMAD4, STK11, TP53*	DNA	5 ug minimum. 10 ug requested	In Tris-HCl (TRIS) buffer*	Room Temperature	Sample must be received within 24 – 48 hours. Overnight preferred. Please make sure the tube is very securely sealed.

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TEST	TYPE OF SPECIMEN	VOLUME REQUIRED	CONTAINER	STORAGE & TRANSPORT	SPECIAL INSTRUCTIONS
Note: Gene panels include both sequencing and deletion duplication analysis with the exception of EPCAM, which is deletion duplication analysis only. *Does not include coverage of *PMS2* exon 15. This exon will be covered by Sanger sequencing for all patients with PMS2 deficient tumors ASHKENAZI JEWISH PANEL FAMILIAL (KNOWN) MUTATION TESTING					

TEST	TYPE OF SPECIMEN	VOLUME REQUIRED	CONTAINER	STORAGE & TRANSPORT	SPECIAL INSTRUCTIONS
COLON CANCER MICROSATELLITE INSTABILITY	10 um tissue sections for each normal and tumour tissue**	10 unstained and 1 H & E stained slide of the same section**	n/a	Room Temperature	Picric acid fixed tissue & decalcification processes made adversely affect DNA retrieval
TUMOUR MUTATION GENOTYPING EXTENDED RAS C-KIT, PDGFRA (GIST OR OTHER)	Paraffin block preferred (repres-entative tumour) or 10 um tissue sections **	10 unstained and 1 H & E stained slide of the same section**	n/a	Room Temperature	Picric acid fixed tissue & decalcification processes made adversely affect DNA retrieval
SOMATIC BRCA TESTING OVARY, FALLOPIAN TUBE, PRIMARY PERITONEAL CANCER	Paraffin block preferred (repres-entative tumour) or 10 um tissue sections **	10 unstained and 1 H & E stained slide of the same section**	n/a	Room Temperature	Picric acid fixed tissue & decalcification processes made adversely affect DNA retrieval
SARCOMA PHARMACOGENOMIC SCREENING BY NGS (DNA) FUSION PANEL (RNA)	Paraffin block (repres-entative tumour)	n/a	n/a	Room Temperature	Picric acid fixed tissue & decalcification processes made adversely affect DNA retrieval
POLYCYSTIC KIDNEY DISEASE PKD1 GENE SEQUENCING PKD2 GENE SEQUENCING	Blood	10 - 20 ml blood	EDTA tube (lavender top)	Room Temperature	Sample must be received within 24-48 hours. Overnight preferred.

TEST	TYPE OF SPECIMEN	VOLUME REQUIRED	CONTAINER	STORAGE & TRANSPORT	SPECIAL INSTRUCTIONS
PKHD1 GENE SEQUENCING PKD1 DELETION DUPLICATION (MLPA) PKD2 DELETION DUPLICATION (MLPA) PKHD1 DELETION DUPLICATION (MLPA) FAMILIAL MUTATION TESTING (PKD1, PKD2, PKHD1)					
MOLECULAR HEMATOPATHOLOGY TESTING FACTOR V LEIDEN & FACTOR II PROTHROMBIN HEMOCHROMATOSIS GENOTYPE	Blood	10 ml blood	EDTA tube (lavender top)	Room Temperature	n/a

*** NOTE:** DNA for testing is to be submitted in TRIS-HCL or H2O only (no EDTA). However, blood is the preferred specimen type and DNA received that does not meet laboratory standard will be rejected or will be issued as inconclusive reports.

DNA sample should contain a minimum of 5 mM Tris-HCl with a pH between 8 and 9.
We recommend to add Tris-HCl pH 8.5 to the sample, resulting in a 5-10 mM concentration.

**Instructions for the selection and cutting of paraffin sections from Tumour blocks

1. Choose the one block with the greatest amoutn/area of the highest grade carcinoma, morphologically consistent with the submitting diagnosis.
2. Neutral buffered formalin is the preferred fixative. Alternative fixatives are NOT recommended.
3. Wrap paraffin block in clean parafilm® and cool in the freezer.
4. Thoroughly clean the microtome and blade holder with mineral oil soaked gauze.
5. Wipe down thoroughly with a clean kim-wipe®.
6. Clean the space that holds the blade with filter paper folded in a "Z" pattern, cleaning the front and back of the blade holder as well as the blade space. Use a new blade for each block.

   Note: Keep old blades for non-PCR use. Change gloves (after cleaning microtome but before inserting new blade) for each case. Cleaning of microtome will take approximately 8-10 minutes between each block.
7. Without trimming any tissue, align the block as close as possible to the blade. Do this as gradually as necessary to avoid cutting away tissue.
8. Cut 1-2 5um sections from the face of the block to remove any surface contaminants, and discard.

9. Cut 10 um curls and transfer to labelled 1.5 ml tube using a clean 200 ul pipette tip. For specimens 5-6 cm^2 use one 1 curl, 1-4 cm^2 use 2 curls, intact needle core biopsies use 4 curls, fragmented needle core biopsies use 6 curls.
10. Use charged glass slides.
11. Prepare 15 5 um serial unstained slides with one 5 um serial section on each slide.
12. Ensure sections on each slide are oriented similarly.
13. After taking sections for PCR, cut 1 5um section for a post-PCR H&E to confirm the presence of tumour in the test sections.
14. Allow the slides to air dry. Do not place the slides in a dryer or hot plate.
15. Do not place cover slips on the unstained slides.
16. Once the slides are dry, insert them into slide carriers.
17. Rewrap block in parafilm and re-file in appropriate location.

MOHLTC ONTARIO Guidelines: **These do not apply to specimens or patients received from out-of-province or country**

RISK CATEGORIES FOR INDIVIDUALS ELIGIBLE FOR SCREENING FOR A GENETIC SUSCEPTIBILITY TO BREAST OR OVARIAN CANCERS

Testing for Affected Individuals with Breast or Ovarian Cancer

At least one case of cancer:

1. Ashkenazi Jewish and breast cancer <50 years, or ovarian cancer at any age.
   Note: testing limited to ethnic specific mutations, unless other criteria given in this list are met.
2. Breast cancer <35 years of age.
3. Male breast cancer.
4. Invasive serous ovarian cancer at any age.

At least 2 cases of cancer on the same side of the family:

5. Breast cancer <60 years, and a first or second-degree relative with ovarian cancer or male breast cancer.
6. Breast and ovarian cancer in the same individual, or bilateral breast cancer with the first case <50 years.
7. Two cases of breast cancer, both <50 years, in first or second-degree relatives.
8. Two cases of ovarian cancer, any age, in first or second-degree relatives.
9. Ashkenazi Jewish and breast cancer at any age, and any family history of breast or ovarian cancer.
   Note: testing limited to ethnic specific mutations, unless other criteria given in this list are met.

At least 3 cases of cancer on the same side of the family:

10. Three or more cases of breast or ovarian cancer at any age.

Testing for Unaffected Individuals (this should be done only if affected individuals are unavailable e.g. deceased)

11. Relative of individual with known BRCA1 or BRCA2 mutation.
    Note: specific family mutation only tested.
12. Ashkenazi Jewish and first or second-degree relative of individual with: breast cancer <50 years, or-ovarian cancer at any age, or-male breast cancer, or-breast cancer, any age, with positive family history of breast or ovarian cancer. *Note: testing limited to ethnic specific mutations, unless meet other criteria*
13. A pedigree strongly suggestive of hereditary breast/ovarian cancer, i.e. risk of carrying a mutation for the individual being tested is >10%.

MOHLTC ONTARIO Guidelines: **These do not apply to specimens or patients received from out-of-province or country**

RISK CATEGORIES FOR INDIVIDUALS ELIGIBLE FOR SCREENING FOR A GENETIC SUSCEPTIBILITY TO COLON CANCER

Testing for Hereditary Non-Polyposis Colon Cancer (HNPCC)

If a tumour sample is unavailable, germline testing may proceed on the youngest, living, affected individual from families meeting criteria 1 & 2 ONLY.

1. Affected and unaffected individuals from families with a known HNPCC causing mutation.
2. Affected individual from Amsterdam I and II families. Family must meet all of the following criteria: Three affected relatives with any combination of colorectal, endometrial, small bowel, ureter, transitional cell kidney cancer (urothelial), sebaceous adenoma/carcinoma and/or keratoacanthoma. One should be a first-degree relative of the other two. At least 2 successive generations should be affected. At least 1 diagnosis must be before age 50. Tumour type should be confirmed by review of pathology or other medical records.
3. Affected individuals from families with: Three affected individuals, one with colorectal cancer, and the other two with any combination of colorectal, endometrial, small bowel, ureter, sebaceous adenoma/carcinoma, ovarian, pancreatic, kidney (transitional cell cancer only), gastric, primary brain or primary hepatobiliary cancer. Two of the three family members must be in a first-degree relationship. At least one diagnosis under the age of 50. FAP should be excluded. Tumours should be verified by pathological examination.
4. Individual affected with CRC and a second primary HNPCC-associated cancer (as listed in #3). This includes synchronous and metachronous colorectal cancers. At least one primary cancer must be diagnosed under age 55. Families are eligible with or without family history of HNPCC-associated cancer, and tumours should be verified by pathological examination.
5. Individual diagnosed with CRC under the age of 35. Families are eligible with or without family history of HNPCC-associated cancer, and tumours should be verified by pathological examination.
6. One case of CRC<50, with a 1st or 2nd degree relative with one of the following HNPCC-related cancers diagnosed <50; colorectal, endometrial, small bowel, ureter, urothelial, sebaceous adenoma/carcinoma or keratoacanthoma.
7. Individuals with immunodeficient tumours (regardless of family history) as follows: MSH2 deficient tumour +/- MSH6 deficiency (sequence and MLPA of MSH2 gene only). MSH6 (only) deficient tumour (sequence and MLPA of MSH6 gene only). MLH1 deficient tumour in individual < age 60 (sequence and MLPA of MLH1 gene only).

Testing for Familial Adenomatous Polyposis (FAP)

Families eligible for testing:

1. Affected and unaffected individuals from families with a known FAP causing mutation.
2. Individuals with clinical confirmed FAP (100 or more adenomas).
3. Individuals with putative attenuated FAP, that is, 10 or more histologically confirmed adenomas. Cumulative pathology and endoscopy reports are required to confirm that the total number and histology are appropriate. A referral with less than 10 adenomas (including hyperplastic polyps) will be excluded.
   Testing for HNPCC will precede APC testing if individuals meet HNPCC testing criteria.

MSH Criteria for Testing Polycystic Kidney Disease

ADPKD

A family history of polycystic (hereditary) kidney disease and:

1) The presence of three or more (unilateral or bilateral) renal cysts on ultrasound in an individual 15-39 years of age (Pei 2009).

   OR

   The presence of a total of more than ten cysts on MRI in an individual between the ages of 16-40 years of age (Pei 2015).

2) The presence of two or more cysts in each kidney on ultrasound in an individual 40-59 years of age (Pei 2009).

3) Cysts in other organs including the liver, seminal vesicles, pancreas, and arachnoid membrane, or extra renal abnormalities including intracranial aneurysms and dolichoectasias, dilatation of the aortic root and absence of other manifestations suggestive of a different renal cystic disease. **Please note that this criterion alone is not an indication for genetic testing for ADPKD.*

No Family history of ADPKD and

4) The presence of bilateral renal enlargement and cysts with or without the presence of hepatic cysts and absence of other manifestations suggestive of a different renal cystic disease (Chapman).

ARPKD

5) Increased renal size, echogenicity and poor corticomedullary differentiation (CMD)

6) Oligohydramnios or anhydramnios

7) Imaging findings consistent with biliary ductal ectasia

8) Clinical/laboratory signs of congenital hepatic fibrosis that leads to portal hypertension and may be indicated by hepatosplenomegaly and/or esophageal varices

9) Hepatobiliary pathology demonstrating a characteristic developmental biliary ductal plate abnormality and resultant congenital hepatic fibrosis

PKD

10) An affected family member with a known *PKD1, PKD2* or *PKHD1* variant(s). Note testing is restricted to only the familial variant(s). Testing in children is not recommended in the absence of clinical symptoms.

Please note: The above criteria are not inclusion/exclusion criteria but may be used along with additional patient medical history to determine if genetic testing is appropriate. Detailed and accurate medical and family history will assist in variant interpretation. Please include all additional information on the requisitions under DISEASE TYPE.

Testing of Minors:
"Presently, in the absence of a regulatory-approved disease-specific therapy, screening at-risk pediatric subjects (18 years of age) may not be justified. The decision to screen should take into account clinical circumstances and parent/carer discussion regarding the potential benefits, risks, and limitations of ultrasound diagnosis in this population. In children, a tailored approach for screening may be appropriate, and include blood pressure measurement with or without renal ultrasound imaging (Ragan 2015)."

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Specimen Packaging and Transport

Specimens should reach the Advanced Molecular Diagnostics laboratory receiving area (Room 6-306) by 3:30 p.m. for processing.

All specimens must be packaged carefully to avoid breakage or leakage of the specimen.

- Ensure all container lids are tightly secured before packaging.
- Ensure specimens are labeled according to protocol above.
- Place tubes / containers into **individual** 6" x 9" biohazard zipper closing bags. Please use **one requisition per specimen** and place it in the bag's outside pouch.
- **DO NOT roll specimen in the requisition or attach specimen to the requisition using tape, elastics, staples or patient demographic labels.**
- Send slides in slide mailers – one slide per track

After Hours Specimen Packaging and Transport

SHIPPING INSTRUCTIONS: Collect and ship samples at room temperature on the same day. Samples should be received within 24 to 48 hours.

For assistance or questions, please call Advanced Molecular Diagnostics at 416.586.4800 x5974 or molecularlab@sinaihelathsystem.ca

Specimens should be packaged in compliance with IATA P.I. 650 shipping standards and the Transportation of Dangerous Goods Act summarized below:

- **A water tight primary receptacle (e.g. blood specimen tube)**
- **A water tight secondary receptacle with sufficient absorbent material to absorb the fluid in the primary receptacle**
- **An outer package of adequate strength for its intended use**

The outside of the package should have a label (see below) indicating '*Non-biohazardous, Non toxic material*' and

of no commercial value. Use the following as a label:

Advanced Molecular Diagnostics
Mount Sinai Hospital
Pathology & Laboratory Medicine,
600 University Ave.,
Rm. 6-306A
Toronto, Ontario
M5G 1X5
Canada

DIAGNOSTIC SPECIMEN

Non-biohazardous, Non toxic material and of no commercial value

Due to issues related to the validation of our test methods, the availability of reagents for DNA extraction and the amount and/or quality of DNA obtained from other tissues, MSH **cannot** generally accept other tissue types (e.g., buccal swabs, fresh tissue or blood spot cards) for DNA extraction and testing. Contact the laboratory manager or director for cases where collection of blood or Formalin fixed tissue samples for DNA extraction is not feasible.

Packaging instructions

Samples should be packaged in a way as to maintain patient confidentiality and prevent leakage and/or contamination to couriers and porters.

Please note:

The quality and quantity of extracted DNA will be affected if samples are collected in the wrong containers, stored at extreme temperatures or delayed in transit. This may result in the failure of the test.

The interpretation of results depends on the diagnosis of affected individuals, identification of samples and biological relationships of the individuals being correct.

For advice on referral to Clinical Genetics and general advice on patients please contact *molecularlab@mtsinai.on.ca*

Lab Storage of DNA

DNA samples will be stored indefinitely. Clinical residual samples may be used for other clinical laboratory purposes including test development, validation and as positive controls for routine diagnostic testing. The laboratory number will be used as identifier for all clinical laboratory purposes and patient confidentiality will be maintained and secured from unauthorized access for all such samples. Use of samples for research purposes and publication will be conducted under institutional Research Ethics Board oversight.

REFERENCES:

1. Canadian College of Medical Geneticists, Molecular Genetics Guidelines, http://www.ccmg-ccgm.org/_includes/ckfinder/userfiles/files/Policies%2C%20Reports%20and%20Position%20Statements/Practice%20Guidelines/PractGuide_MOLEC_MolecGenTest_Sept2006.pdf
2. Ontario Laboratory Accreditation Requirements, Quality Management System – Laboratory Services, Version 5.1, 2011
3. Hall, J., Hamerton, J., Hoar, D., Korneluk, R., Ray, P., Rosenblatt, D. & Wood, S. (1991) Canadian College of Medical Geneticists: Policy statement concerning DNA banking and molecular genetic diagnosis. *Clinical and Investigative Medicine 14 (4)*, 363-365.
4. The Canadian College of Medical Geneticists (2002) *CCMG Molecular Genetics Guidelines.* http://www.ccmg-ccgm.org.
5. Harris PC, Torres VE. Polycystic Kidney Disease, Autosomal Dominant. 2002 Jan 10 [Updated 2015 Jun 11]. In: Pagon RA, Adam MP, Ardinger HH, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2016. Available from: http://www.ncbi.nlm.nih.gov/books/NBK1246/

6. Chapman AB, Devuyst O, Eckardt KU, Gansevoort RT, Harris T, Horie S, Kasiske, BL, Odland D, Pei Y, Perrone RD, Pirson Y, Schrier RW, Torra R, Torres VE, Watnick T, Wheeler DC; Conference Participants. Autosomal-dominant polycystic kidney disease (ADPKD): executive summary from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. Kidney Int. 2015 Jul;88(1):17-27. doi:10.1038/ki.2015.59. Epub 2015 Mar 18. PubMed PMID: 25786098

7. Rangan GK, Lee VW, Alexander SI, Patel C, Tunnicliffe DJ, Vladica P. KHA-CARI Autosomal Dominant Polycystic Kidney Disease Guideline: Screening for Polycystic Kidney Disease. Semin Nephrol. 2015 Nov;35(6):557-564.e6. doi: 10.1016/j.semnephrol.2015.10.004. Review. PubMed PMID: 26718159.

8. Sweeney WE, Avner ED. Polycystic Kidney Disease, Autosomal Recessive. 2001 Jul 19 [Updated 2014 Mar 6]. In: Pagon RA, Adam MP, Ardinger HH, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2016. Available from: http://www.ncbi.nlm.nih.gov/books/NBK1326/

9. Pei Y, Obaji J, Dupuis A, Paterson AD, Magistroni R, Dicks E, Parfrey P,

Cramer B, Coto E, Torra R, San Millan JL, Gibson R, Breuning M, Peters D, Ravine D. Unified criteria for ultrasonographic diagnosis of ADPKD. J Am Soc Nephrol. 2009 Jan;20(1):205-12. doi: 10.1681/ASN.2008050507. Epub 2008 Oct 22. PubMed PMID: 18945943; PubMed Central PMCID: PMC2615723.

10. Pei Y, Hwang YH, Conklin J, Sundsbak JL, Heyer CM, Chan W, Wang K, He N, Rattansingh A, Atri M, Harris PC, Haider MA. Imaging-based diagnosis of autosomal dominant polycystic kidney disease. J Am Soc Nephrol. 2015 Mar;26(3):746-53. doi: 10.1681/ASN.2014030297. Epub 2014 Jul 29. PubMed PMID: 25074509; PubMed Central PMCID: PMC4341484.