



MICHIGAN MEDICINE  
UNIVERSITY OF MICHIGAN

LABORATORIES



2024

# Molecular Services



New molecular division director, **Annette S. Kim, M.D., Ph.D.** and her team.

[mlabs.umich.edu](http://mlabs.umich.edu)  
800.862.7284

Michigan Medicine Laboratories (MLabs) is a CLIA-certified/CAP accredited laboratory offering state of the art molecular testing services that support diagnosis, prognosis and therapeutic guidance for your patients.

Our highly experienced faculty, ABMGG board-certified geneticist, medical technologist and bioinformaticians are dedicated to delivering the highest quality laboratory results to meet the needs of today's patients in a cost effective and personalized manner.

**142k**

SQUARE FOOT  
BRAND NEW STATE  
OF THE ART FACILITY

**50+**

YEARS OF  
EXPERIENCE

**30+**

MOLECULAR, PEDIATRIC  
& GENETIC COUNSELORS  
AND CONSULTANTS



The combined molecular (both oncology and germline) and cytogenetic laboratories of Michigan Medicine Laboratories was rebranded as the Division of Diagnostic Genetics and Genomics (DGG) in 2023 under new division director, Annette S. Kim, M.D., Ph.D., Henry Clay Bryant Professor of Pathology. This rebranding is to celebrate a new stage in the history of genetic and genomic medicine with new state-of-the-art technologies and assays under development.

# MOLECULAR ONCOLOGY & GENETICS

## CONSULTANTS



**Eman Abdulfatah**  
M.D.



**Elizabeth Ames**  
M.D., Ph.D. Board  
Certified Geneticist



**Bryan Betz**  
Ph.D.



**Scott Bresler**  
M.D., Ph.D.



**Noah Brown**  
M.D.



**May Chan**  
M.D.



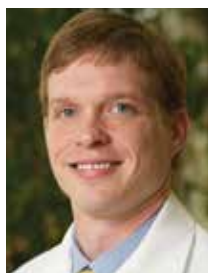
**Arul M. Chinnaiyan**  
M.D., Ph.D.



**Kyle Conway**  
M.D., J.D.



**Thomas Giordano**  
M.D., Ph.D.



**Paul Harms**  
M.D., Ph.D.



**Nora Joseph**  
M.D.



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**Annette Kim**  
M.D. Ph.D. Director  
for Molecular and  
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**David Manthei**  
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**Aiko Otsubo**  
Ph.D.



**Lina Shao**  
Ph.D. Board Certified  
Geneticist



**Thomas E. Wilson**  
M.D., Ph.D.



**Chan Yang**  
M.D., Ph.D. Board  
Certified Geneticist

## SERVICES

### MOLECULAR ONCOLOGY

Performing over 20,000 assays annually with an average turn-around-time of 5 days, our Molecular Oncology Laboratory specializes in offering cutting-edge molecular tests to aid in cancer diagnosis, selection of therapy, determination of prognosis, and monitoring of residual disease. An expanding menu of tests are offered in disease-specific service lines including a comprehensive array of both single gene assays and next-generation sequencing (NGS) panel.

3%  
QNS RATE

#### SOLID TUMOR MOLECULAR ONCOLOGY

Both single gene assays and robust next generation sequencing panels to provide excellent sensitivity and broad coverage while also preserving the ability to test small biopsies and challenging specimens with low neoplastic cell content.

68  
MOLECULAR  
ONCOLOGY  
TESTS

#### HEMATOLYMPHOID MOLECULAR ONCOLOGY

Single gene assays as well as a comprehensive Myeloid NGS Panel to detect clinically-relevant somatic mutations as well as gene fusions/rearrangements that can be cryptic by conventional cytogenetic methods and germline mutations associated with a predisposition to a myeloid neoplasm that might not be otherwise suspected (e.g. DDX41, ANKRD26, GATA2, ETV6, etc.). Myeloid NGS is also validated on a variety of specimen types including formalin-fixed, paraffin-embedded (FFPE) tissue.

### MOLECULAR GENETICS

Molecular Genetics Laboratory offers comprehensive clinical testing for germline genetic diseases and is committed to assisting clinicians in identifying genetic aberrations to facilitate clinical diagnosis, management, and genetic counseling. Molecular Genetics Laboratory offers a variety of germline NGS (sequencing and del/dup) tests for single-gene and cancer panels including: Breast and Ovarian, Colorectal, Pancreatic, Endometrial/Uterine, Stomach, and Prostate. In addition, the laboratory offers SNP Chromosomal Microarray; screening and diagnostic testing for cystic fibrosis and spinal muscular atrophy; methylation analysis for Prader-Willi/Angelman syndrome, Beckwith-Wiedemann & Russel-Silver syndrome, and other imprinting disorders; and Fragile X tests.

8  
PANELS

80+  
GENETIC  
TESTS

# SELECT TEST MENU

## Molecular Oncology

### MYELOID NEOPLASMS (AML, MDS, MPN)

#### Myeloid NGS Panel\*

NPM1 Mutation (PCR)  
FLT3 Mutation (PCR)  
CEBPA Mutation (Sanger)  
IDH1 and IDH2 Mutation (Sanger)  
KIT D816V Mutation (PCR)  
KIT Mutation for AML - Exons 8, 17 (Sanger)  
PML/RARA t(15;17) Translocation  
Quantitative (PCR)  
JAK2 V617F Mutation (PCR)  
JAK2 Exon 12 Mutation (PCR)  
CALR Mutation (PCR)  
MPL Mutation (PCR)  
BCR/ABL1 Analysis, Quantitative (PCR)  
BCR/ABL1 Kinase Domain Mutation (Sanger)

### BREAST CANCER

HER2 (FISH)  
PIK3CA Mutation (NGS)  
PD-L1 (IHC)

### LYMPHOPROLIFERATIVE DISORDERS

B Cell Clonality (PCR)  
(IGK & IGH Gene Rearrangement)  
B Cell Clonality (PCR)  
(IGK Gene Rearrangement)  
B Cell Clonality (PCR)  
(IGH Gene Rearrangement)  
T Cell Clonality (PCR)  
(TRG & TRB Gene Rearrangement)  
T Cell Clonality (PCR)  
(TRG Gene Rearrangement)  
T Cell Clonality (PCR)  
(TRB Gene Rearrangement)  
IGH/BCL2 t(14;18) Translocation (PCR)  
IGH/BCL2 t(14;18) Translocation (FISH)  
BCL6 (3q27) Rearrangement (FISH)  
MYC (8q24) Rearrangement (FISH)  
MALT1 (18q21) Rearrangement (FISH)  
MYD88 (L265P) Mutation (PCR)  
BRAF V600E/V600K Mutations (PCR)

## COLORECTAL AND ENDOMETRIAL CANCER

### Colorectal Cancer NGS Panel\*

MLH1 Promoter Methylation (PCR)  
KRAS Mutation (NGS)  
NRAS Mutation (NGS)  
BRAF V600E/V600K Mutations (PCR)  
Microsatellite Instability Analysis (PCR)  
UGT1A1 Promoter Genotyping (PCR)  
POLE Mutation (Sanger)

### GASTROINTESTINAL STROMAL TUMOR

KIT Mutation - Exons 9,11,13,17 (Sanger)  
PDGFRA Mutation for GIST (Sanger)

### GENITOURINARY TUMOR

FGFR Mutation/Translocation (NGS)  
BRAF (7q34) Rearrangement (FISH)  
ERG Rearrangement (FISH)  
TFE3 (Xp11,2) Rearrangement (FISH)  
for Renal Cell CA & Other Tumors  
TFEB (6p21) Rearrangement (FISH) for  
Renal Cell Carcinoma  
UroVysion™ (FISH) (Bladder Cancer)  
TERT Promoter Mutation (PCR)

### NEURO-ONCOLOGY

#### Solid Tumor NGS Panel\*

BRAF (7q34) Rearrangement (FISH)  
IDH1 and IDH2 Mutations for Glioma (NGS)  
1p/19q Deletion (FISH)  
BRAF V600E/V600K Mutations (PCR)  
MGMT Promoter Methylation (PCR)  
TERT Promoter Mutation (PCR)  
Neuropathology Methylation Array  
Cancer Cytogenomics Array

### LUNG CANCER

#### Lung Cancer NGS Panel\*

EGFR Mutation (NGS)  
BRAF V600E/V600K Mutations (PCR)  
KRAS Mutation (NGS)  
ALK Rearrangement for NSCLC (FISH)  
ROS1(6q22) Rearrangement (FISH)  
RET (10q11) Rearrangement (FISH)  
PD-L1 (IHC)  
MET Amplification (FISH)

## MELANOMA

### Melanoma NGS Panel\*

BRAF (7q34) Rearrangement (FISH)  
BRAF V600E/V600K Mutations (PCR)  
KIT Mutation for Melanoma -  
Exons 11,13,17 (Sanger)  
NRAS Mutation (NGS)  
Chromosomal (Microarray) for Melanoma  
Multiprobe (FISH) for Melanoma  
TERT Promoter Mutation (PCR)  
Cancer Cytogenomics Array

### PROSTATE CANCER

Prostate Cancer Antigen 3 (PCA3)

### BONE AND SOFT TISSUE

SYT/SSX Translocation (PCR)  
PAX/FOXO1 Translocation (PCR)  
EWSR1/WT1 Translocation (PCR)  
EWSR1/ATF1 Translocation (PCR)  
EWSR1/FLI1 & EWSR1/ERG Translocation (PCR)  
EWSR1 (22q12) Rearrangement (FISH)  
MDM2 Amplification (FISH)  
CIC (19q13) Rearrangement (FISH)  
DDIT3 (12q13) Rearrangement (FISH)  
PDGFB (22q13) Rearrangement (FISH)  
USP6 (17p13) Rearrangement (FISH)

### THYROID CANCER

BRAF V600 E/V600K Mutations (PCR)  
BRAF (7q34) Rearrangement (FISH)  
TERT Promoter Mutation (PCR)

### MISCELLANEOUS

#### Solid Tumor NGS Panel\*

Biliary Tract Malignancy (FISH)  
Bone Marrow Transplant Engraftment  
Analysis (PCR)  
HER2 (FISH)  
UGT1A1 Promoter Genotyping (PCR)  
Mesothelioma (FISH)  
PD-L1 (IHC)

\*NGS Panels include: mutations (35 genes), amplification (19 genes) and gene fusion (21 genes)

TEST LISTING CONTINUED ON BACK

## SELECT TEST MENU CONTINUED

### Genetics (Germline)

#### AUTISM SPECTRUM DISORDERS/ INTELLECTUAL DISABILITY

Chromosomal Microarray Analysis  
Fragile X Syndrome Mutation  
Prader-Willi /Angelman Syndrome

CDKL5 Gene Sequencing  
GDI1 Gene Sequencing  
MBD5 Gene Sequencing  
MEF2C Gene Sequencing  
NLGN3 Gene Sequencing  
NLGN4X Gene Sequencing  
SHANK2 Gene Sequencing  
SHANK3 Gene Sequencing  
SLC9A6 Gene Sequencing

TCF4 Gene Sequencing  
UBE3A Gene Sequencing

#### MECP2 (RETT SYNDROME)

MECP2 Gene Sequencing  
MECP2 Deletion/Duplication  
MECP2 Targeted Sequencing Familial

#### PTEN HARMARTOMA TUMOR SYNDROME

PTEN Gene Sequencing  
PTEN Deletion/Duplication  
PTEN Targeted Sequencing Familial

#### BREAST AND OVARIAN CANCER

BRCA1 and BRCA2 Gene Sequencing  
BRCA1 and BRCA2 Targeted  
Sequencing, Familial  
BRCA1 and BRCA2 Deletion/Duplication  
BRCA Ashkenazi Jewish Founder Mutations  
BRCA Mutation Panel  
Hereditary Breast and Ovarian Cancer (HBOC)  
Comprehensive Germline NGS Panel  
Hereditary Breast and Ovarian Cancer (HBOC)  
High-Moderate Risk Germline NGS Panel

PTEN Gene Sequencing  
PTEN Deletion/Duplication  
TP53 Gene Sequencing  
TP53 Deletion/Duplication

#### COLORECTAL CANCER

Colorectal Cancer Germline NGS Panel  
MSH2 Gene Sequencing  
MLH1 Promoter Methylation (PCR)

#### CYSTIC FIBROSIS

Cystic Fibrosis Carrier Screening  
Cystic Fibrosis Full Gene Sequencing  
Cystic Fibrosis Deletion/Duplication  
Cystic Fibrosis Diagnostic Mutation Detection  
Cystic Fibrosis Targeted Sequencing Familial

#### HEARING LOSS

GJB2 (Connexin 26) Mutation Analysis  
GJB2 (Connexin 26) Targeted Sequencing  
Familial  
GJB6 (Connexin 30) Deletion Analysis  
WFS1 (Wolfram Syndrome) Gene  
Sequencing

#### NOONAN SYNDROME

PTPN11 Gene Sequencing  
KRAS Gene Sequencing  
RAF1 Gene Sequencing  
SOS1 Gene Sequencing

#### FRAGILE X SYNDROME

Fragile X Syndrome Mutation Detection

#### LI-FRAUMENI SYNDROME

TP53 Gene Sequencing  
TP53 Deletion/Duplication

#### NEUROFIBROMATOSIS

NF1 Gene Sequencing

#### SPINAL MUSCULAR ATROPHY

SNM1 and SNM2 Deletion/Duplication

#### MISCELLANEOUS

Apolipoprotein E Genotyping  
Factor V Leiden Mutation  
Hereditary Hemochromatosis Mutation  
Prothrombin 20210 Mutation

#### CANCER GERMLINE NGS PANELS

##### Colorectal Cancer Germline NGS Panel

**19 Genes:** MLH1, MSH2, MSH6, MUTYH, PMS2, EPCAM, APC, TP53, PTEN, STK11, SMAD4, BMPR1A, CDH1, CHEK2, GREM1, POLD1, POLE, ATM, AXIN2

##### Endometrial/Uterine Cancer Germline NGS Panel

**13 Genes:** BRCA1, BRAC2, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, PMS2, PTEN, TP53, STK11, POLD1

##### Hereditary Breast and Ovarian Cancer Comprehensive Germline NGS Panel

**21 Genes:** ATM, BARD1, BRAC1, BRAC2, BRIP1, CDH1, STK11, CHEK2, EPCAM, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, PTEN, RAD51C, RAD51D, TP53, FANCC, XRCC2

##### Hereditary Breast and Ovarian Cancer High-Moderate Risk Germline NGS Panel

**9 Genes:** ATM, BRAC1, BRAC2, BRIP1, CDH1, CHEK2, PALB2, PTEN, TP53

##### Pancreatic Cancer Germline NGS Panel

**18 Genes:** APC, ATM, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, STK11, TP53, CDK4, BMPR1A, SMAD4, VHL, XRCC2

##### Prostate Cancer Germline NGS Panel

**6 Genes:** BRCA1, BRCA2, CHEK2, HOXB13, NBN, TP53

##### Stomach Cancer Germline NGS Panel

**11 Genes:** MLH1, MSH2, MSH6, EPCAM, PMS2, APC, TP53, STK11, CDH1, BMPR1A, SMAD4



Expertise Delivered  
Professionally