

2025 Molecular Services

Noah Brown, M.D. Assoc. Professor, Pathology Director, Molecular Diagnostics

Bryan Betz, Ph.D. Assoc. Professor, Molecular Diagnostics, Molecular Genetics, Molecular Pathology Technical Director, Division of Molecular and Genomic Pathology 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.





30⁺ MOLECULAR, PEDIATRIC & GENETIC COUNSELORS AND CONSULTANTS

DIAGNOSTIC GENETICS AND GENOMICS EXPERTS



Annette Kim M.D. Ph.D. Director, Molecular and Genomic Pathology



Noah Brown M.D. Director, Molecular Diagnostics



Bryan Betz Ph.D. Techincal Director, Molecular and Genomic Pathology



Daniel Hovelson Ph.D. Director, DGG Informatics



Marcin Cieslik Ph.D. Director, **Bioinformatics for** Division of DGG



Eman Abdulfatah MBBCh. MSc



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



Robert Bell M.D.



Daniel Boyer M.D., Ph.D.



Scott Bresler M.D., Ph.D.



May Chan



Kyle Conway M.D., J.D.



Paul Harms M.D., Ph.D.



Nora Joseph M.D.



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



Jaeseung Kim Ph.D.

Thomas E. Wilson

M.D., Ph.D.





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



Suguna Narayan M.D., Ph.D.



Aiko Otsubo Ph.D.



Lina Shao M.D., Ph.D., FACMG **Board Certified** Geneticist



Christina Sloan-Heggen M.D., Ph.D.





Navin Mahadevan M.D., Ph.D.



SERVICES

MOLECULAR ONCOLOGY

Performing over 17,000+ assays annually with an average turn-around-time of 7.7 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.

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.

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 formalinfixed, 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.



80⁺ GENETIC TESTS



68 MOLECULAR ONCOLOGY 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)

BREAST CANCER

HER2 (FISH) PIK3CA Mutation (NGS) PD-LI (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 (3g27) Rearrangement (FISH) MYC (8g24) Rearrangement (FISH) MALT1 (18g21) 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

Comprehensive Solid Tumor Fusion Panel 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-LI (IHC)

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

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 SHANK2 Gene Sequencing SHANK3 Gene Sequencing SLC9A6 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

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