



UW Cytogenetics and Molecular Genetics Services

Wisconsin State Laboratory of Hygiene

University of Wisconsin-Madison

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CONTACT INFORMATION

UW CYTOGENETICS & MOLECULAR GENETICS SERVICES LABORATORY

465 Henry Mall
Madison, WI 53706

Phone: (608) 262-0402

Fax: (608) 265-7818

Email: cytogenetics@slh.wisc.edu

www.slh.wisc.edu/clinical/cytogenetics

LABORATORY DIRECTOR

Vanessa Horner, Ph.D., FACMG

ASST. LABORATORY DIRECTOR

Kaitlin Lenhart, Ph.D.

ONCOLOGY SUPERVISOR

Eric Johnson, B.S. CG(ASCP)

PRENATAL SUPERVISOR

Kim Oxendine, B.S. CG(ASCP)

MOLECULAR SUPERVISORS

Leah Frater-Rubsam, Ph.D.

Richel Mayberry, M.S., MT(ASCP)CG, MB

GENETIC COUNSELOR

Kimberly Anderson, M.S. CGC

Contents

[Test Menu Updates](#)

[Test Reporting Updates](#)

[Specimen Shipper Kits](#)

[Variant Reanalysis Requests](#)

[Employee Spotlight](#)

Test Menu Updates

New FISH probes sets available for ordering

Effective immediately, the UW Cytogenetics and Molecular Genetics Services Laboratory is offering three new FISH probes sets:

MECOM: The *MECOM* break-apart DNA probe set is used to detect rearrangements of the *MECOM* gene including translocations and deletions. Rearrangements of the *MECOM* gene are associated with acute myeloid leukemia (AML) with *inv(3)(q21.3q26.2)* or *t(3;3)(q21.3q26.2)*, or myelodysplastic syndrome (MDS).

NUP98: The *NUP98* break-apart DNA probe set is used to detect rearrangements of the *NUP98* gene including translocations and deletions. Rearrangements of the *NUP98* gene are associated with high risk AML.

MYH11-CBFB: The *MYH11-CBFB* dual fusion DNA probe set is used to detect rearrangements between the *MYH11* and *CBFB* genes and deletions of 16q22. The *MYH11-CBFB* fusion is associated with *inv(16)(p13q22)*, *t(16;16)(p13;q22)*, and *del(16)(q22)* in acute myeloid leukemia (AML) or myelodysplastic syndromes (MDS).

Test Reporting Updates

New Chromosomal Microarray Analysis (CMA) reporting guidelines

In late 2019, the American College of Medical Genetics and Genomics (ACMG), in collaboration with the Clinical Genome Resource (ClinGen), published updated technical standards for the interpretation and reporting of constitutional copy number variants (CNVs) (PMID: 31690835). These updated CNV interpretation guidelines utilize a semi-quantitative point based scoring metric for CNV classification. The goal of this evidence-based classification system is to increase consistency and transparency across clinical laboratories. The UW Cytogenetics and Molecular Genetics Services Laboratory has implemented these guidelines, and subsequently updated our report format to be consistent with the recommendations made by ACMG and ClinGen. If you have any questions regarding our updated reporting guidelines, please contact our laboratory (608-262-0402).

Specimen Shipper Kits

Specimen shipper kits available

The Wisconsin State Laboratory of Hygiene strives to make testing as accessible as possible. One approach to reducing barriers to genetic testing is to offer readily available shipper kits. The UW Cytogenetics and Molecular Genetics Services Laboratory will provide specimen shipper kits to clinicians upon request. The kit includes everything required to ship human blood specimens within the United States, including a pre-paid return mailer envelope. The laboratory will ship kits to clinicians or directly to patients if requested. This service is provided free of charge. Shipper kits may be requested by phone (608-262-0402) or email (cytogenetics@slh.wisc.edu).

Variant Reanalysis Requests

Variant reanalysis request process

The UW Cytogenetics and Molecular Genetics Services Laboratory will reanalyze reported variants of uncertain significance identified through microarray or exome sequencing upon request. Reanalysis requests can be made at least one year after the time of testing. An amended report will be issued to the original ordering provider within 1-4 weeks of the request. Reanalysis requests may be made by faxing (608-265-7818) or emailing (cytogenetics@slh.wisc.edu) the laboratory a completed Variant Reanalysis Request Form. The Variant Reanalysis Request Form can be found on our website (www.slh.wisc.edu/clinical/cytogenetics) or requested by phone (608-262-0402).

Employee Spotlight

UW Cytogenetics and Molecular Genetics Services Laboratory's Newest Assistant Director

In September of 2019, the laboratory welcomed its newest addition to the team. Kaitlin Lenhart, Ph.D. has joined Director Vanessa Horner, Ph.D., FACMG as Assistant Director of Cytogenetics and Molecular Genetics Services.

Dr. Lenhart received her Ph.D. in Pathology and Laboratory Medicine from the University of North Carolina – Chapel Hill (*go Heels!*), where she studied genetic and cellular mechanisms of skeletal muscle disorders. She then carried out postdoctoral training at UNC's McAllister Heart Institute, performing genomic studies for the Heart Healthy Lenoir project, an initiative to reduce blood pressure and improve overall health of an impoverished group of individuals living in rural North Carolina. Dr. Lenhart then went on to complete a Laboratory Genetics and Genomics (LGG) Training Fellowship at the University of Colorado Denver – Anschutz Medical Campus.

Dr. Lenhart has been a wonderful addition to our team and we are excited to have her with us at the UW Cytogenetics and Molecular Genetics Services Laboratory!

