Fluorescence in situ hybridization (FISH) is a useful tool for assessment of specific chromosomes or chromosomal regions of interest.

Specifically, FISH probes may be designed to detect copy number (loss, gain, and amplification) of loci known to be associated with disease, such as amplification of ERBB2 (HER-2/neu) in breast cancer and hyperdiploidy in pediatric acute lymphocytic leukemia (ALL). Alternatively, FISH probes may be designed to detect rearrangements of certain genes of interest that are known to cause disease, such as the translocation between chromosomes 9 and 22 resulting in BCR/ABL1 gene fusion in patients with chronic myelogenous leukemia (CML).

In addition to our extensive hematology/oncology/lymphoma FISH offerings, our laboratory provides one of the most comprehensive solid tumor FISH menus available. For some testing indications, such as a suspected diagnosis of lipoblastoma, initial testing may include only a single FISH probe (e.g., a probe that targets rearrangement of PLAG1). For other testing indications, we have developed indication-specific probe panels based on current literature (e.g., a ten probe panel for loci of interest in multiple myeloma; an eight probe panel for loci of interest for myelodysplastic syndrome). As we continually expand our menu, please contact the laboratory or refer to the web page for the most up-to-date FISH probe offerings.

Unlike many other tests, FISH allows for a rapid analysis of the region(s) of interest, enabling providers to have diagnostic results within hours of specimen collection for critical patients. For example, the presence of a translocation between chromosomes 15 and 17, resulting in fusion of the PML and RARA genes, is diagnostic for the M3 subtype of acute myeloid leukemia (AML) called acute promyelocytic leukemia (APL). Since the treatment of APL differs from the treatment of patients with other subtypes of AML, our laboratory has recently validated a PML/RARA dual fusion probe that reduces the hybridization time from 4 to 1.5 hours without compromising assay performance, thereby reducing the amount of time from specimen collection to communication of FISH results by approximately 50%. We look forward to more rapidly meeting your patients’ needs with this probe and welcome any questions that you may have regarding the improved process.
New Look, Same Promise

We hope that you have become familiar with our University’s new logo, released in late 2014. This logo has been included on our print materials for several months, but we have more recently transitioned our test collection kits to incorporate this new look. Also, our team has been working diligently over the past several months to generate test-specific brochures for each of our hereditary cancer tests. These brochures were developed as a tool for providers to share with patients when testing options are being discussed. Please contact our marketing specialist if your facility is in need of new test collection kits or if your patients would benefit from the newly developed informational brochures for our hereditary cancer tests.

In the months after Dr. Sanger’s passing, our team has remained firmly committed to our laboratory’s mission: to provide accurate and affordable genetic testing services. During this time of transition, our team continues under the leadership of Dr. Tanner Hagelstrom, Interim Director, and Drs. Bhavana Dave and Jennifer Sanmann, Associate Directors. We value your trust in our expertise and welcome questions or suggestions about how our laboratory can better meet the needs of you, our medical colleagues, and the patients that we work together to serve.

The Human Genetics Laboratory Team
University of Nebraska Medical Center

est. 1974

MISSION: To provide accurate and affordable genetic testing and interpretation for the patients we serve.
“As a laboratory genetic counselor, I am a resource and advocate for your patients as they seek testing services.”

Rachel Barbar, M.S., licensed and certified genetic counselor

Comprehensive Genetic Testing for Patients Experiencing Pregnancy Loss

At least 10% to 15% of all recognized pregnancies are miscarried. Chromosome abnormalities cause more than 50% of first trimester losses and 15% of second trimester losses.1 In many situations, identifying the underlying genetic etiology for a pregnancy loss may help to resolve misconceptions about the cause of the loss. In addition, it may help to alleviate feelings of guilt and blame that are common in women experiencing a loss and, in most instances, will allow for accurate calculation of recurrence risks to determine appropriate fertility and future pregnancy management. The approach to genetic testing depends largely on the patient’s history and current pregnancy state. Please refer to the table below for the recommended testing algorithms for patients experiencing pregnancy loss.

<table>
<thead>
<tr>
<th>Patient History</th>
<th>Our Recommended Testing</th>
<th>Rationale</th>
</tr>
</thead>
<tbody>
<tr>
<td>Woman with a history of recurrent pregnancy loss not pregnant</td>
<td>Chromosome Analysis performed on peripheral blood for woman and her partner</td>
<td>Chromosome Analysis identifies a chromosomal rearrangement, such as a balanced translocation, in 5-8% of individuals with recurrent loss.2,3</td>
</tr>
<tr>
<td>Woman with a history of recurrent pregnancy loss currently experiencing a miscarriage</td>
<td>Pregnancy Loss Microarray performed on fetal tissue</td>
<td>Pregnancy Loss Microarray offers an increased detection rate and greater likelihood of obtaining results as compared to traditional chromosome analysis.4</td>
</tr>
<tr>
<td>Woman currently experiencing a stillbirth</td>
<td>Pregnancy Loss Microarray performed on fetal tissue</td>
<td>Pregnancy Loss Microarray offers an increased detection rate and greater likelihood of obtaining results as compared to traditional chromosome analysis.4</td>
</tr>
<tr>
<td>Woman without a history of recurrent pregnancy loss currently experiencing a miscarriage</td>
<td>Pregnancy Loss Microarray performed on fetal tissue</td>
<td>Testing may be appropriate for women experiencing their first or second loss, particularly when the family or pregnancy history is suspicious for a chromosomal cause of pregnancy loss.</td>
</tr>
</tbody>
</table>

![Genetic Breakdown of Pregnancy Loss](Image)

Genetic Counselors Link Laboratory and Providers

Genetic testing is rapidly evolving and is being used by an increasingly large number of health care providers for diagnosis and medical management. Our laboratory team includes licensed and board-certified genetic counselors who assist providers and their patients as they navigate the genetic testing process.

Laboratory genetic counselors participate in the entirety of the testing process, beginning with a review of the ordered tests at specimen receipt and extending beyond the laboratory to provider education. In addition, they are available to recommend indication-specific testing strategies, to review clinical information for improved result interpretation, to notify providers of urgent results, and to discuss test results and pertinent management issues with ordering providers. Please call 402.559.8400 to speak with a laboratory genetic counselor regarding general testing questions or patient-specific inquiries.

Rachel Barbar, M.S., LCGC has been a genetic counselor at UNMC since 2011. She came to Omaha from the University of Wisconsin - Madison, where she received her Master of Science in Genetic Counseling. She is originally from Curlew, Iowa, and received her Bachelor of Science in Integrative Physiology from the University of Iowa. Rachel enjoys collaborating with her laboratory and clinical colleagues to provide access to medical genetic services for patients and support to their health care providers.

Dani Bishay, M.S., LCGC has been a genetic counselor at UNMC since 2009. Originally from Chadron, Nebraska, she received her Bachelor of Science in Biology from University of Nebraska-Lincoln and trained as a genetic counselor at University of Arkansas for Medical Sciences, receiving her Master of Science in Genetic Counseling in 2009. Dani enjoys both the challenge of investigating novel genetic variants and the variety of clinical experiences her job affords.

Our laboratory is also pleased to announce that Kathryn Matthews, M.S., joined our team in September after completing her Masters of Science in Genetic Counseling at the University of Oklahoma Health Sciences Center.

Genetic Testing for Male Infertility

Genetic testing may be useful for men who exhibit non-obstructive azoospermia or oligospermia, as current literature suggests approximately 7% of infertile men have a structural or numerical chromosome anomaly.1–2 Specifically, chromosome abnormalities are found in 3-5% of men with oligospermia, 10-19% of men with non-obstructive azoospermia, and less than 1% of men with a normal sperm count.2–3 Although most often phenotypically normal, an additional 10% of men with azoospermia and severe oligospermia harbor microdeletions in the azoospermia factor region (AZF) of the Y chromosome.2–4

To ensure comprehensive testing, our Male Infertility Panel includes two assays: Chromosome Analysis and Y Chromosome Microdeletion (YCMD) studies. The combination of these tests can detect structural and numerical chromosomal anomalies (sex chromosome aneuploidies, translocations, and inversions) and microdeletions in the Y chromosome including the three azoospermic regions (AZFa, AZFb, and AZFc).

ADVANTAGES OF TESTING:

– Influences Clinical Management

• Men with deletions in AZFa, AZFb, or AZFb+c are poor candidates for sperm retrieval procedures.5
• Men with deletions of AZFc causing azoospermia have a 50% success rate for sperm retrieval.2
• Men who carry structural chromosome rearrangements and their partners may be offered prenatal genetic testing, preimplantation genetic screening, or alternate reproductive strategies.

– Determine Risks to Offspring

• Male offspring of men with AZFc deletions conceived following sperm retrieval will inherit their father’s microdeletion and will have a high risk for infertility.2,4
• Men with structural chromosome abnormalities may have an increased risk for miscarriage and children with chromosome abnormalities or congenital defects.

Results are generally available in 10-14 days. For comprehensive testing, please send 3-5 ml of whole blood in both a sodium heparin tube and an EDTA tube. Collection kits are available at no cost to our clients by contacting Nicole Hackendahl, our marketing specialist, at 402.559.6935.
Molecular Genetics Fellowship Established

In addition to a fellowship in the subspecialty of clinical cytogenetics, the American Board of Medical Genetics and Genomics (ABMGG) recently approved the Human Genetics Laboratory’s request to train individuals holding an M.D. or a Ph.D. in the subspecialty of molecular genetics. This fellowship is typically completed over 1-2 years, during which time fellows participate in all aspects of assay development, sample preparation, analysis, and reporting in the molecular genetics area of the laboratory.

International Collaborators Visit the Laboratory

Our laboratory has developed a long-term collaborative effort with Dr. Alvin Soon Tiong Lim from Singapore General Hospital and his colleagues at the neighboring KK Women’s and Children’s Hospital. Dr. Lim and five of his colleagues from SingHealth visited the laboratory earlier this year to discuss career path development for laboratory staff, competency assessment, staff education, updated test offerings, and assay development. This week-long collaboration provided a great opportunity to exchange ideas and to foster the long and mutually beneficial relationship that we’ve had over the past two decades with members of the SingHealth team.

Visit us online at www.unmc.edu/geneticslab

Renée Fordyce-Boyer Announces Retirement

Throughout her years in the lab, Renée has provided expertise in a multitude of areas, including specimen processing, chromosome analysis, regulatory guidelines and compliance, and billing practices. In addition, Renée has developed tremendous professional relationships with you, our laboratory clients, over the past 38 years.

We will miss Renée’s experience, leadership, and wisdom in the laboratory but hope that you will help us to send Renée into retirement with a shower of gratitude and well wishes.

Thank you, Renée, for your invaluable contributions to patients and providers, as well as the field of laboratory medicine, over the past 40 years!

Diane Pickering, M.S., CG(ASCP)CM, former supervisor in the microarray section of the laboratory, has begun the transition to laboratory manager in preparation for Renée’s retirement. We are confident that Diane’s 26 years of experience with our team will make this a seamless transition for our clients.

After celebrating her 40 year anniversary with the UNMC team, our laboratory manager, Renée Fordyce-Boyer, M.S., CG(ASCP)CM, announced that she will be retiring in January.
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