Enhanced Testing Portfolio and Improved Strategy for Multiple Myeloma FISH Testing

Fluorescence in situ hybridization (FISH) plays a critical role in the assessment of patients with a clinical diagnosis of multiple myeloma, as certain cytogenetic abnormalities confer a poorer prognosis. Specifically, t(4;14), t(14;16), and t(14;20) are translocations that are designated as high risk markers; gain of 1q, often observed with deletion of 1p, deletion of 17p (TP53), and nonhyperdiploidy are also markers of increased concern in this cohort of patients. As a result, treatment may vary for patients with multiple myeloma based on the cytogenetic abnormalities detected by targeted FISH analysis.

Our laboratory continues to perform CD138 cell sorting on samples received with a suspected diagnosis of multiple myeloma or a related testing indication. We also recently validated a t(14;20) dual fusion probe in the laboratory to detect IGH/MAFB fusion, which is associated with high risk disease, and added probes for 1p/1q to our testing portfolio (Table 1).

To ensure that the most prognostically-relevant information is available to clinicians, we have developed a testing algorithm for patients with a known, suspected, or associated diagnosis of multiple myeloma (Figure 1). Probes associated with the highest risk disease will be performed initially, followed by probes associated with lower risk markers as the number of CD138+ cells available for study allows.


FIGURE 1. Testing algorithm for bone marrow samples from patients with a known, suspected, or associated diagnosis of multiple myeloma

DAY 1

<table>
<thead>
<tr>
<th>Probes</th>
</tr>
</thead>
<tbody>
<tr>
<td>6q23</td>
</tr>
<tr>
<td>CDKN2C (1p32.3) / CKS1B (1q21.2) new probe</td>
</tr>
<tr>
<td>D13S319 [13q14] / 13q34</td>
</tr>
<tr>
<td>Hyperdiploidy</td>
</tr>
<tr>
<td>9 centromere</td>
</tr>
<tr>
<td>11 centromere 15q22</td>
</tr>
<tr>
<td>IGH [14q32 abnormalities]</td>
</tr>
<tr>
<td>IGH / CCND1 [t(11;14)]</td>
</tr>
<tr>
<td>IGH / FGFR3 [t(4;14)]</td>
</tr>
<tr>
<td>IGH / MAF [t(14;16)]</td>
</tr>
<tr>
<td>IGH / MAFB [t(14;20)] new probe</td>
</tr>
<tr>
<td>TP53 [17p13.1]</td>
</tr>
</tbody>
</table>

DAY 2

- if an IGH disruption is observed:
  - t(14;16), t(14;20), t(4;14), t(11;14)
  - 13q9 MM/MYB
- if no IGH disruption is observed:
  - 13q9 MM/MYB
Committed to Success

Greetings from the Human Genetics Laboratory! Our team has enjoyed many exciting developments over the past few months. We have added several new members to our cytogenetics, molecular genetics, and client services teams to meet ongoing needs; and we look forward to their contributions for years to come. In addition, we continue to survey the needs of clinicians in order to enhance our testing portfolio to meet those needs. In particular, clinical exomes can provide an answer for many patients whose underlying genetic condition has previously gone unidentified, and our molecular team has been working diligently on this project. Please be on the lookout in the coming months for this exciting testing development!

As the need for genetic testing continues to grow and reach a broader range of medical providers, our team recognizes the complexities of the genetic testing process, such as insurance inquiries and specimen collection requirements. We hope that you have had an opportunity to interact with our client services team over the past year to help you with the logistical details associated with testing. If not, please don’t hesitate to reach out for all of your billing, insurance and preauthorization, and specimen-related questions by calling 402-559-5070 (option 3) or by emailing clientservhgl@unmc.edu.

The Human Genetics Laboratory Team
University of Nebraska Medical Center

est. 1974

MISSION: To be leaders in providing exceptional genetic testing and interpretation for patients, families, and communities we serve through extraordinary patient care, premier educational programs, and innovative research.
Dr. Sanmann Named Director of Laboratory

Jennifer Sanmann, Ph.D., has been named the director of the Human Genetics Laboratory.

The appointment is the culmination of a journey that began in 2005, when Dr. Sanmann joined MMI as a technologist and enrolled at UNMC as a graduate student.

“When I came on board, I was unsure about my future career path but could not be more grateful for the opportunities and people here at UNMC, particularly my friend and mentor, the late Dr. Warren Sanger, who introduced me to the field of clinical laboratory medicine,” she said. “His encouragement and passion got me hooked on genetics, and the mission and people at MMI have kept me energized ever since.”

With genetic testing advancing at a remarkable pace, it is an exciting time for the lab, Dr. Sanmann said.

“The number of patients whose care we can positively affect through testing is growing exponentially,” she said. “It was the impact on patients’ lives that drew me into the field of clinical laboratory medicine, and it is this impact that keeps me focused on excellence. However, for me, the well-known saying ‘there’s no place like Nebraska’ really rings true. I am incredibly excited about the opportunity to do this work in a place where I truly feel at home alongside a remarkable laboratory team.”

In announcing her appointment, MMI Director Karoly Mirnics, M.D., Ph.D., said he had been impressed with Dr. Sanmann’s knowledge, thoughtfulness, dedication, responsiveness to challenges, management skills, cheerful attitude and resilience as she served as the lab’s interim director.

“I am convinced that she is the perfect person to lead our laboratory,” he said. “With Jennifer at the helm, the best days of the Human Genetics Lab are yet to come.”

Dr. Sanmann says leading the lab will require vision and versatility from herself and her staff.

“As with all other areas of health care, we are asked continually to deliver more for less in half the time,” she said. “In addition, the field of genetics is evolving rapidly, and undoubtedly genetic testing will look very different in 5, 10, and 20 years. Success will require us to push beyond current constraints and past experiences and to readily transform our approach to clinical genetic testing.”
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 402.559.5070 and ask to speak with a laboratory genetic counselor regarding general testing questions or patient-specific inquiries.

Hope Chipman, M.S., LCGC has been a genetic counselor at UNMC since 2006 but joined the laboratory team in 2016. Originally from Northfield, Minnesota, she received her Bachelor of Science in Biology from Iowa State University and trained as a genetic counselor at Sarah Lawrence College in Bronxville, New York, receiving her Master of Science in Human Genetics in 2006. Hope enjoys the discovery of unique findings that help to provide answers to patients with challenging medical diagnoses.

Joanna Spaulding, M.S., LCGC recently joined UNMC in June, moving back to the Midwest after several years of clinical practice in Southern California. Originally from Kansas City, she graduated from Valparaiso University in Indiana in 2007 with a Bachelor of Science in Biology; she went on to obtain her Master of Science degree through the genetic counseling training program at the University of Colorado Denver in 2009. Joanna enjoys educating others about the diversity of genetic disease and the importance of genetic testing in both clinical and research settings.

Hereditary Cancer Testing

Hereditary cancer testing remains a vital part of the services offered through the Human Genetics Laboratory, as this information can be critical to inform cancer screening and prevention strategies for individuals at increased risk. Our testing portfolio ranges from small, disease-specific panels to a comprehensive panel that includes many genes associated with a known increased risk for various types of cancer. Regardless of which hereditary cancer panel is best for your patient, these panels include both sequencing-based assessment as well as high resolution deletion/duplication analysis to ensure detection across the spectrum of mutations that may be present. Our client services team is able to assist with the insurance authorization, and our laboratory provides specimen collection kits and informational patient brochures to streamline the process for providers. Please visit our website for complete information about the gene content and specimen requirements for our hereditary cancer panels.
Outreach and Education

2017 marks the second year our laboratory has exhibited at the NE SciFest at Omaha’s Durham Museum where participants learned about genetics by using gummy worm “chromosomes” to create a karyotype (a systematic arrangement of chromosomes in descending order of size). In our interactive booth, technologists discussed how this activity relates to the real-life karyotyping performed in our laboratory to diagnose various cancers and other genetic disorders. Additionally, our laboratory was among a select group invited to bring a private, mini SciFest to patients within the pediatric oncology unit on The Nebraska Medical Center campus. This special event provided our team the rare opportunity to connect personally with patients about how we use cytogenetic analysis to identify extra, missing, or atypical chromosomes.

A Week of Harry Potter Fun

Our group celebrated Medical Laboratory Professionals Week (April 24-28) with a Harry Potter theme. Staff were sorted into four teams of Hogwarts houses and earned points through participation in wizard wand and tie making, Quidditch, daily hunts to find the escaped Golden Snitch, Potions Class, and an interactive who-dunnit mystery. Festivities concluded with a judged potluck feast and everyone’s favorite activity - a laboratory safety quiz. Securing a second place win in the Association of Genetic Technologists (AGT) photo contest, our week of team building and magic was deemed victorious!

Genetic Reporting: At Your Fingertips

Did you know that genetic reports generated by our FACMG-boarded directors could be at your fingertips moments after sign-out?

HIPAA-compliant, secure emailed reporting is available to all providers. Simply request emailed reporting by contacting our laboratory by telephone (402-559-5070) or email (humangenetics@unmc.edu).

Once emailed reporting has been added to your provider profile, an email will be generated upon completion of testing. Then, follow the instructions within the email to create a log in and password needed to retrieve our reports. Still offered are options to have finalized reports faxed immediately upon sign-out and/or printed and delivered by postal mail.
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5 Genetic Reporting
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EDUCATIONAL OUTREACH OPPORTUNITIES

SEPT 13-16, 2017 | COLUMBUS, OH
NATIONAL SOCIETY OF GENETIC COUNSELORS (NSGC) ANNUAL CONFERENCE

APR 10-14, 2018 | CHARLOTTE, NC
AMERICAN COLLEGE OF MEDICAL GENETICS AND GENOMICS (ACMG) ANNUAL MEETING