Gene Up! New genes added to hereditary cancer panels

The Human Genetics Laboratory (HGL) routinely expands its indication-specific gene panels when new gene-disease associations are identified. To reflect the advancements in scientific knowledge associated with hereditary cancer syndromes, our team updated several hereditary cancer panels this fall. These updates increased the number of genes evaluated in two focused panels (Breast/Ovarian/Uterine and Colorectal) and in the Comprehensive panel. The additions of DICER1 and SMARCA4 have expanded our panel for individuals with a personal or a family history of breast, ovarian, and/or uterine cancer from 26 to 28 genes. Similarly, three additional genes, BLM, MSH3, and NTHL1, have been added to our colorectal hereditary cancer panel, expanding this assay to include interrogation of 23 genes. All five of these new genes are also available in the Comprehensive63 panel.

Testing comprehensiveness in every hereditary cancer panel is exceedingly important to the HGL. To this end, our hereditary cancer tests interrogate for both the sequence level variation and copy number changes of all genes included in the study. Likewise, gene selection plays a critical role in maximizing diagnostic yield without identifying a large number of genetic changes with uncertain clinical significance. Basic science research is implicating genes in cancer predisposition at an incredible rate, which may provide exciting candidate genes for patients and families affected by cancers that are apparently hereditary in nature. However, significant scientific rigor is required to demonstrate the clinical utility of testing for these genes, both from the perspective of increased cancer risk and from the perspective of medical management and decision-making processes. To balance the rapid rate of discovery with meaningful clinical utility, our team curates the literature and established guidelines to gather sufficient evidence to support the addition of new genes based on well-established disease associations.

Combining both analyses into a single panel is a thorough approach that significantly increases the diagnostic yield relative to performing sequencing or copy number studies alone.

Clinicians may order genetic testing for individuals who have or who are at-risk to have a predisposition to early onset tumors. Depending on the patient’s personal and family history, ordering clinicians may select from one of the indication-specific hereditary cancer panels available through the HGL. Alternatively, they may create a custom panel that mixes and matches genes from the Comprehensive63 to ensure analysis of all genes that are clinically indicated. As previously noted, both sequence level variants and copy number variants are included in each standard and custom panel.
Happy Holidays from Our Team to Yours

In health care, we share our patients’ successes and setbacks on a daily basis, both directly and, in our case as laboratorians, indirectly. However, this season offers us an opportunity to reflect on the past year from a 10,000 foot view, celebrating our major achievements and identifying opportunities for continued improvement. In 2018, the Human Genetics Laboratory broadened our scope of genetic testing in multiple areas of the laboratory, including fluorescence in situ hybridization (FISH) and molecular genetics, to keep pace with the ever-changing needs of patients. Our FISH portfolio was enhanced to include probes that target newly identified markers associated with leukemia, lymphoma, and solid tumors. Among other molecular test expansions, clinical exome sequencing was launched in the spring, which allows for improved diagnostic yield for patients who have often been on diagnostic odysseys. In addition, we contributed to several research projects aimed at improving our understanding of the genetic basis of disease, which we hope will positively impact clinical care in years to come. Finally, we are very proud to have provided genetics education to students and clinicians across Nebraska and the world.

We are sincerely grateful to be your genetic testing laboratory and look forward to a continued partnership. Our team has several exciting developments on the horizon and is eager to share these with you in 2019. As the genetic testing needs of your patients evolve, we appreciate your feedback and suggestions about our testing services and portfolio. Please feel welcome to reach out to us through a phone call, an email, or our client survey (accessible via our website: unmc.edu/geneticslab).

Wishing you joy and health in the coming year!

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and The Human Genetics Laboratory Team

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.
Happy Holidays

Beyond the Laboratory

As an academic unit at UNMC, education plays an important role in our mission. Locally, we participate in the Nebraska Science Festival and several other conferences and meetings throughout the Omaha-area, allowing us opportunities to personally connect with health care providers, researchers, and the public. Our staff is routinely involved in local and national field-specific committees and organizations to extend the scope of our educational presence. We also exhibit at the American College of Medical Genetics (ACMG) and National Society of Genetic Counselors (NSGC) annual meetings. If you attend one of those meetings in 2019, please stop by our booth for a visit!

Ordering Updates

Complying with Signature Requirements for Diagnostic Tests:

In accordance with Medicare guidelines, a physician signature is required for all diagnostic test orders. Similarly, other payors follow suit. There are several different options to comply with these requirements when ordering laboratory testing:

1. Submission of a signed requisition either electronically through an EMR or handwritten by the ordering provider. Ideally, the signature should be above a printed or typed name.

2. If the requisition is unsigned there must be medical documentation by the ordering physician that he/she intended the diagnostic test be performed.

3. A signature attestation from the ordering provider/author of the medical record can be submitted. Our laboratory will contact your facility for documentation, typically via fax, as needed.
Oncology FISH

Ph-like Acute lymphoblastic leukemia (ALL), also known as BCR-ABL-like ALL, a subtype of precursor B-cell ALL, is characterized by a range of genomic alterations and is associated with adverse outcomes. The genetic changes found in Ph-like ALL trigger a select number of defined signaling pathways that can potentially be targeted by tyrosine-kinase inhibitors. Precise identification of the genetic alterations in patients with Ph-like ALL may thus help therapeutic stratification and likely improve the outcome. Currently, the Human Genetics Laboratory offers FISH probes for two chromosomal regions, the Xp22.3 flanking the CRLF2 gene and the 5q32 region flanking the PDGFRB gene. Validation studies for more FISH probes related to Ph-like ALL are in progress, and an expanded FISH panel for Ph-like ALL will soon be available. Please check our website for updates in early 2019.

Our goal is to request and obtain prior authorizations as quickly and as accurately as possible for you and your patients. In an effort to save you and your staff the time required to navigate the appeals process, we are requesting that your clinic notes include the following documentation. While the reasons for testing may be obvious to us, insurance requires your patient’s clinical presentations to be clearly stated and not implied or inferred.

Genetic Testing Supporting Documentation:
Authorization of genetic testing, especially molecular genetics, is complex. Frequently payors manually review letters of medical necessity (LMN) and other supporting documentation when making authorization decisions. Many denials are due to lack of supporting documentation. Below are key components to a successful authorization.

- Indicate how the test results will be utilized in the medical management of your patient. Include specifics.
- Mention if test results will significantly improve a patient’s treatment outcome. Include diagnostic or therapeutic interventions.
- Document your patient’s current clinical scenario, history, physical examination, and conventional diagnostic test results.
- Indicate if your patient was seen by a geneticist or genetic counselor.
- Include a pedigree and/or family history when appropriate.

For questions related to client services, please contact our team at clientservhgl@unmc.edu.

If you have more specific questions about the status of a preauthorization, our team can be contacted at preauthhgl@unmc.edu.

Billing questions? We have answers! Email billinghgl@unmc.edu.

“Because treatment decisions are increasingly driven by genetics, the Human Genetics Laboratory continues to be driven to meet those needs through test development.”

Jennifer Sanmann, PhD, FACMG
Family, Friends, Team

The average American spends 90,000 hours at work over their lifetime, and that’s a daunting prospect for those who don’t create bonds with coworkers. Thankfully, our story stands in stark contrast.

Many of our staff have worked in the laboratory for more than 15 years; some more than 25. For others, well, let’s just say that they’re probably approaching the aforementioned 90K mark. Together, we’ve walked, biked, curled, golfed, traveled, and gathered at each other’s homes. Through all of these activities, we’ve established and strengthened bonds that pour into the trust and cohesiveness necessary to fulfill our laboratory’s mission to its fullest potential. It is a privilege to have the opportunity to serve you and your patients through such exciting work – genetics! And, the journey is just that much sweeter doing it with the friends that we’ve made along the way!

WE VALUE YOUR INPUT!
Access a client satisfaction survey on our home page

Visit us online at www.unmc.edu/geneticslab

Genetic Reporting: At Your Fingertips

Did you know that genetic reports generated by our ABMGG-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. The options to have finalized reports faxed immediately upon sign-out and/or printed and delivered by postal mail also continue to be offered.
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At HGL it’s in our genes.

EDUCATIONAL OUTREACH OPPORTUNITIES

APR 2-6, 2019 | SEATTLE, WA
AMERICAN COLLEGE OF MEDICAL GENETICS AND GENOMICS (ACMG) ANNUAL MEETING

NOV 5-8, 2019 | SALT LAKE CITY, UT
NATIONAL SOCIETY OF GENETIC COUNSELORS (NSGC) ANNUAL CONFERENCE