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Using cancer's fingerprint, researchers clinch a diagnosis

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Rochester scientists are using a gene test to diagnose a difficult-to-detect form of bone marrow cancer – an example of how academic medical centers are applying new technology in ways that play to their strengths to achieve better patient care.

In cancer care, gene tests are used broadly to predict the course of disease or to select the best treatment. The University of Rochester School of Medicine & Dentistry group, however, has shown there's a niche role for genomics, in this case using it to differentiate a type of early leukemia from other conditions.

"To be able to tell somebody: 'We know what disease you have,' is hugely important," said W. Richard Burack, M.D., Ph.D., director of Hematopathology at UR Medicine and the James P. Wilmot Cancer Center, overseeing gene testing for all types of cancer. "This is an example of precision medicine that sets us apart from many other institutions."

The test is called a comparative genomic hybridization with single nucleotide polymorphism (CGH-SNP) analysis. The UR is licensed by New York State to use the CGH-SNP microarray testing platform.

Scientists are uniquely applying CGH-SNP tests to suspected cases of myelodysplastic syndrome. MDS occurs when the bone marrow does not mature properly and can't generate enough healthy blood cells. Rochester has a long history of excellence in MDS research, led by Professor

Emeritus John M. Bennett, M.D., a widely recognized pioneer in the study of MDS who served as the founder and first chair of The Myelodysplastic Syndromes Foundation.

Known as pre-leukemia, MDS is often confused with other illnesses. Symptoms such as weakness, tiredness and unexplained bruising signal a low blood count, but low counts are also associated with anemia, lupus, infection, exposure to toxins, or even poor nutrition. Routine blood work can suggest the possibility of MDS, but approximately 50 percent of cases indicate vague cell abnormalities that need further clarification.

Patients with suspected MDS usually endure an invasive bone marrow biopsy. But those results often come back as indeterminate, requiring a long "watch and wait" period before the diagnosis can be confirmed and treatment can begin, Burack said.

M. Anwar Iqbal, Ph.D., associate professor of Pathology and Laboratory Medicine and director of UR Medicine's Microarray CGH Laboratory, understood the clinical problem and designed a study to apply the CGH-SNP test to 84 bone marrow samples from patients with abnormal blood work, comparing them to 22 normal samples.

The genetic analysis of MDS is trickier than with other cancers, because instead of having just one genetic lesion, innumerable changes in the chromosomes are associated with MDS. Often the DNA errors look different in each patient, and Wilmot pathologists have become experts at discerning which genetic changes are involved in each patient's cancer.

Among the 84 samples, Iqbal's CGH-SNP analysis produced positive results for MDS in 54 cases (64 percent). Importantly, in four cases doctors would not have established an MDS diagnosis without the CGH-SNP test. The test also revealed MDS-related genetic abnormalities that

would have otherwise gone undetected in three additional patients; and in 16 cases scientists learned more information about the genetic complexity of MDS.

Further studies are needed, but the microarray analysis seems to add a significant benefit that can alter medical decision-making, researchers concluded in a novel report. They were invited to present their data March 4, 2014, at the 103rd annual meeting of United States and Canadian Academy of Pathology (USCAP), the world's largest gathering of pathologists.

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