

Dec. 6, 2005
For immediate release

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Laboratory of Human Molecular Genetics Receives Patent for Ground-Breaking DNA Probes

Imagine a microscopic submarine swimming through the body, searching for defects in DNA that lead to disease. And imagine that submarine identifying the tiniest strands of DNA in such a way that doctors can offer personalized treatment for cancer and inherited genetic disorders.

This is not science fiction. In fact, doctors at the University of Missouri have received a United States patent (*US Patent # 6,828,097: Single copy genomic probes and method of generating the same*) for DNA probes that can very specifically detect a piece of genetic material to determine the exact cause of diseases.

Cancer and inherited genetic malformations are caused by mutations in our body's chromosomes. But we've never been able to see all the genetic nuances of exactly how those chromosomes are different – until now.

Using the human genome sequence, the Laboratory of Human Molecular Genetics at the University of Missouri School of Medicine in Kansas City, Mo., developed ground-breaking technology that does just that. Developed by lab co-directors, Joan H.M. Knoll, PhD, a cytogeneticist, and Peter K. Rogan, PhD, a molecular geneticist, utilize a novel technology called single copy genomic fluorescence in situ hybridization (or scFISH).

Drs. Knoll and Rogan hope to make these tiny probes widely available for research and clinical applications. In the future, two patients with seemingly similar illnesses, leukemia for instance, could be offered more specific treatments for their specific conditions.

“There are certain diseases we want to be able to dissect better because we know so patients with the same diagnosis don't respond as well to certain treatments,” explains Dr. Knoll. “These probes allow us to now drill down to the DNA level and see that cause of the disease may actually be slightly different between patients with the same diagnosis. Such findings will make it possible to direct patients to the right specialist who may eventually be able to prescribe personalized treatments.”

Currently, there are commercial DNA probes available to diagnose many genetic diseases and conditions, but these probes do not provide the same level of detail as single copy probes. “Single copy probes are much smaller and much more densely represented on a chromosome,” Dr. Knoll explains. “Therefore, we can look at much smaller targets and be more precise in our diagnosis. Another advantage of Single Copy Genomic Probes is that we can search for very rare medical conditions.”

Drs. Knoll and Rogan already have tested the probes and published scientific papers in the *American Journal of Medical Genetics* and in *Genome Research*, both high-impact, respected journals in the field of human genetics.

In these papers, the doctors reported on their experience using Single Copy Genomic Probes to detect a wide variety of congenital disorders. With funding from the National Cancer Institute, they also have used the probes to diagnose acquired diseases such as chronic and acute myelogenous leukemia.

There are potentially similar applications for the thousands of other diseases and conditions that affect the human body, but the possibilities don't stop there. The National Science Foundation has funded the development of an automated microscopy system which will accelerate Single Copy Genomic Probe analysis. “These probes can help us target the specific DNA information that affects health, not just in humans, but in animals as well,” Dr. Rogan explains.

The doctors are interested in making Single Copy Genomic Probes widely available for research and clinical applications and are seeking to license the technology to interested companies. “These probes have the potential to help us answer some of medicine's most intriguing questions, and ultimately save lives,” Dr. Rogan says.

Additional funding for the research using Single Copy Genomic Probes has been provided by the Hall Foundation, the Katherine B. Richardson Trust and the Paul Patton Charitable Trust.

“Over the past 10 years, our children's hospital has emerged as a leader in research, and the work Drs. Knoll and Rogan are doing is an integral part of that effort,” says Randall L. O'Donnell, PhD, president and chief executive officer. “We are very proud that the research they are doing here could have this kind of potential impact on the future of medicine, and on our ability to provide the best and most advanced medical care possible for children.”

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