Once a futuristic concept, the routine use of genetic information to advance patient care is at our doorstep. The cost of full genome sequencing is decreasing rapidly, and the expertise needed to interpret genetic sequence information and translate that to patient care is developing. Lastly, patients and physicians are beginning to create a demand for this information, creating a convergence of factors that provide an impetus to develop personalized medicine programs.

A recent case demonstrated the potential of personalized medicine. Medical College of Wisconsin researchers and a team of Medical College physicians at Children’s Hospital of Wisconsin were among the first to use genetic sequencing to help in the diagnosis of a perplexing and severe chronic illness in a young patient. Their effort revealed 16,124 variations unique to this patient’s genome sequence. Examining the information, the team identified a unique, never previously reported mutation in 1 gene that is highly conserved and caused the child’s illness, a previously undocumented inflammatory bowel disease.

Medical genomics is a definitive tool in the emerging era of personalized medicine, which began with the completion of the Human Genome Project in 2003. That effort took 13 years and cost $2.7 billion. Today, complete genome sequencing of individuals is available for $50,000, and costs are falling fast. Howard Jacob, PhD, the Warren P. Knowles Professor in Human and Molecular Genetics, Professor of Physiology and Pediatrics, and Director of the College’s Human and Molecular Genetics Center (HMGC), predicts the price will be less than $1,000 by 2014. The Medical College, through generous financial support from the philanthropy, is in the process of purchasing two new sequencers for the HMGC that will markedly reduce the time and cost of a whole genome sequencing while greatly increasing the accuracy of the results.

The logical progression is for genome sequencing to be integrated into the primary care physician’s toolbox. Reviewing a patient’s profile could reveal to the physician diseases or conditions to which that patient is predisposed, and appropriate interventions could begin. For example, the physician could recommend lifestyle changes that might delay onset of the genetically indicated disease, or prescribe medication to control or prevent the disease’s development. Alternatively, physicians would be able to predict an individual’s response to pharmaceuticals, increasing effectiveness of drug choice and dose while reducing adverse reactions.

Cultivating such clinical applications requires further dedication to the science behind them. The Medical College is building this knowledge base through the work of interdisciplinary centers and programs.

College faculty in the HMGC analyze the genomic sequence to better understand diseases. Center researchers oversee projects related to diabetes, cervical cancer, cardiovascular disease, cystic fibrosis and more. The HMGC includes the Individualized Medicine Institute, led by Ulrich Broeckel, MD, Associate Professor of Pediatrics. Institute research includes comparing DNA from healthy individuals to DNA from individuals with particular diseases, searching for the genetic basis of that disease. Functional analysis is performed to understand the mechanisms that then contribute to the disease’s development.

The College’s Biotechnology and Bioengineering Center (BBC), directed by Andrew Greene, PhD, Professor of Physiology, contributes to personalized medicine through technology development.

In 2009, the BBC, with the HMGC, was selected by the National Human Genome Research Institute as home to the Wisconsin Medical Journal • 2010 • Volume 109, No. 5
the storage capacity of a typical PC. Multiply this by the over 2000 patients in an average primary care practice and the need for significantly increased computer memory and processing power is clear.

Additionally, physicians will require training to understand genetic information and gain proficiency at explaining the implications of genetic predispositions to patients during a typical 10-minute appointment.

The means to making personalized medicine viable in clinical care is within reach. Research, like that conducted at the Medical College of Wisconsin and our partner institutions, is key to ensuring related technologies are effective, dependable, and accessible.

Center of Excellence in Genomics Science (WCEGS), 1 of only 10 such centers in the nation pursuing novel technologies to advance genetics research. A collaboration between the Medical College, University of Wisconsin-Madison and Marquette University, the Center is co-directed at the Medical College by Michael Olivier, PhD, Professor of Physiology, and at UW-Madison by Lloyd M. Smith, PhD, Director of the Genome Center of Wisconsin. The WCEGS team is developing technologies for analyzing proteins that bind to DNA. Understanding how individual cells use the information encoded in the genome is critical to translating genetic sequence data to clinical care.

Although personalized medicine suggests many health care improvements and benefits to patients, it also presents major challenges.

Health care professionals and policymakers will encounter a host of bioethical issues. Physicians will have to weigh the consequences of fully disclosing to patients their profile’s results, including their risk for an incurable disease. Safeguarding information must also be addressed, lest individuals face discrimination from potential employers or insurers. Changes in legislation are beginning to reduce the risk of discrimination.

Technically problematic is that just one genomic or proteomic sequencing profile generates a terabyte of data, which is equal to 1024 gigabytes—far greater than the storage capacity of a typical PC. Multiply this by the over 2000 patients in an average primary care practice and the need for significantly increased computer memory and processing power is clear.

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