## GENETICS IN YOUR PRACTICE

# Internist-Geneticist to Head NIH

The internal medicine community should be encouraged that President Obama recently tapped Dr. Francis S. Collins, an internist-geneticist, to lead the largest biomedical research enterprise on the planet. As director of the National Institutes of Health, Dr. Collins will play a major role in shaping our nation's basic science and translational research agenda.

BY MATTHEW R.G. TAYLOR, M.D., PH.D.

A substantial proportion of the research published in the major clinical journals read by internists are supported directly or indirectly by NIH dollars. This summer the NIH received roughly $10 billion additional money as part of President Obama's economic stimulus effort.

The presence of an internist-geneticist at the helm of the NIH is likely to foster more research on adult-onset conditions. Dr. Collins is best known for his contributions as a geneticist, and although most clinical geneticists are trained in pediatrics, Dr. Collins' career reflects his internal medicine roots. He has been centrally involved in discovering genes for conditions that are not exclusive to children, including Huntington's disease, neurofibromatosis, cystic fibrosis, and multiple endocrine neoplasia. Throughout much of the 1990s, he was director of the National Center for Human Genome Research, which coordinated the NIH's work on the International Human Genome Project. He was also involved in locating a gene for progeria, a disease of premature aging that causes teenagers to develop adult-onset diseases of senility.

The appointment of an internist-geneticist will highlight the importance of applying genetic principles to the diagnosis and management of adult-onset diseases. Genome-wide association studies ("2008: Year of the GWAS," Feb. 1, 2009, p. 28), with the prospect of widespread use of next-generation DNA sequencing just around the corner, are positioning scientists to dissect the genetics of complex diseases and to understand gene-environment interactions. These advances will increase our understanding of many common disorders of adulthood (obesity, hypertension, atherosclerosis, diabetes) and shed light on genetic variants that influence disease risk.

Indeed, Dr. Collins has predicted that genetic testing will continue to move beyond the realm of diagnosis and further into risk assessment. In a 1999 review, he presented a hypothetical scenario of a 23-year-old man visiting his primary care physician in the year 2010. Family history and molecular genetic testing for risk variants would be done to develop an individualized screening and prevention strategy. Although this tailored approach is not likely to be in widespread use by 2010, the successes of recent genome-wide association studies are important steps in that direction.

The idea that genetic testing could be widely used to assess the risk of future disease promises to expand the role of genetics into disease prevention. As proposed by Dr. Collins, the ability to employ genetic markers for risk assessment for future diseases or for predicting responses to medications, for example, would usher in an era of "personalized medicine." Thus a patient's own genetic makeup (family history and select genetic markers) would allow treatment and preventive efforts to be individually customized.

We are still several years away from implementing this in the clinic, since current studies are just starting to identify the genetic risk markers for common diseases. However, Dr. Collins' vision no longer appears as "futuristic" as it did when he championed this approach a decade ago. In early 2010, Dr. Collins will publish his latest in a series of genetics-related books, which will center on the movement of personalized medicine from research to the clinical arena. If his past works are any indication, the latest book will be thought provoking and challenge us to keep pace with the forces coming to bear on internal medicine and our health care system as a whole. ■

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# *Dr. Francis S. Collins Begins As NIH Director*

Dr. Francis S. Collins, former director of the National Human Genome Research Institute, become director of the National Institutes of Health on Aug. 17 after being approved unanimously by the Senate earlier in the month.

"The National Institutes of Health stands as a model when it comes to science and research," President Obama said when he nominated Dr. Collins for the post in July. "My administration is committed to promoting scientific integrity and pioneering scientific research, and I am confident that Dr. Francis Collins will lead the NIH to achieve these goals. Dr. Collins is one of the top scientists in the world, and his groundbreaking work has changed the very ways we consider our health and examine disease. I look forward to working with him in the months and years ahead."

Dr. Collins oversaw the federal Human Genome Project, which finished the complete mapping of the human genome in April 2003, at about the same time as a parallel private effort. Dr. Collins' research also has led to the discovery of several genes. He is interested in the intersection of science and faith and has written two books on the topic.

Dr. Collins received a bachelor's degree in chemistry from the University of Virginia, a Ph.D. in physical chemistry from Yale University, and a medical degree from the University of North Carolina. Before coming to NIH in 1993, he spent 9 years on the faculty of the University of Michigan, where he was an investigator at the Howard Hughes Medical Institute. He is a member of the Institute of Medicine and the National Academy of Sciences, and was awarded the Presidential Medal of Freedom in November 2007.

**—Joyce Frieden**

# NIH-Pharma Collaboration Urged to Advance Drug Pipeline

BY JOYCE FRIEDEN

WASHINGTON — The National Institutes of Health needs to partner more with the pharmaceutical industry in order to create a better pipeline for new drugs, Dr. Francis Collins said at the annual meeting of the Endocrine Society.

Dr. Collins, who recently began work as NIH director, said that with all the developments now occurring in genomic research, "pharmaceutical companies are a little overwhelmed about where to start" when it comes to figuring out which genes would make good targets for drug therapy.

"Academic investigators should get more intentionally involved in the translational process of going from basic research to drug development," Dr. Collins said before his nomination and confirmation as NIH director.

"There is an opportunity now, more than ever, to bring together academic investigators and the private sector to put together a really exciting version of a drug development pipeline," he said.

Such a collaboration "involves more of academics taking the front-end risk of developing promising compounds so they become attractive and licensable by the private sector."

Dr. Collins noted that many academic researchers are identifying promising targets for drugs, "but relatively few are taking that information and turning it into an assay … to see if there is something promising that might turn out to become a therapeutic."

Some targets start out looking promising, but when they get to a point where they need support for preclinical development, "that's where things often die," he said. "Congress just a few months ago put $24 million into the fiscal year 2009 budget to start this process in an NIH-funded way, and I hope the money will go up substantially in the next 5 years."

**'There is an opportunity now … to put together a really exciting version of a drug development pipeline.'**

DR. COLLINS

Conflicts of interest on the part of pharmaceutical companies "would have to be factored in," Dr. Collins said in an interview. Drugmakers' interest in commercialization would be a factor. "You want to start a project that is going to get somewhere," he said. "But there are companies across the board that are interested in almost any disease—even very rare ones—as long as it won't cost a fortune to get that drug approved. For the rare diseases, you may have to push things further down the pipeline with public money before the company says, 'Okay, I'll start with that one now,' but I don't know that that should discourage consideration of working on even a very rare disease in this pipeline."

"We have an opportunity to take [the new technologies] that have started to appear and apply them in a vigorous way to understand fundamentals of biology, That would include genomics and nanotechnology and a wide variety of approaches to epigenetics," he said.

"I would also think we need to take seriously the charge coming from the Congress and the Administration to provide useful information to guide health care reform. That would mean, in some instances, comparative effectiveness research, but we need to be careful not to lose the personalized aspects of individual [health] along the way."

Global health should also be an NIH focus, he said. "The U.S. is in a position to spread more soft power instead of hard power around the world; NIH ought to be able to play a useful role in that. And we should encourage the research community, including young investigators, and increase the diversity of our workforce, to make it vigorous and effective. I think if we focus on a few of those things … we can really get some stuff done." ■