



U.S. Department of Health and Human Services

NIH News
National Institutes of Health

National Human Genome Research Institute (NHGRI)
<http://www.genome.gov>

NIH Office of Rare Diseases (ORD)
rarediseases.info.nih.gov

NIH Clinical Center (CC)
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NIH Launches Undiagnosed Diseases Program *Clinical Researchers to Tackle the Most Puzzling Medical Cases*

The National Institutes of Health (NIH) today announced a new clinical research program that will aim to provide answers to patients with mysterious conditions that have long eluded diagnosis. Called the Undiagnosed Diseases Program, the trans-NIH initiative will focus on the most puzzling medical cases referred to the NIH Clinical Center in Bethesda, Md., by physicians across the nation.

“A small number of patients suffer from symptoms that do not correspond to known conditions, making their care and treatment extraordinarily difficult. However, the history of biomedical research has taught us that careful study of baffling cases can provide new insights into the mechanisms of disease — both rare and common,” said NIH Director Elias A. Zerhouni, M.D., who has made a point during his six-year tenure at NIH of encouraging trans-NIH initiatives. “The goal of NIH’s Undiagnosed Diseases Program is two-pronged: to improve disease management for individual patients and to advance medical knowledge in general.”

The new program, which got under way over the past month, is the culmination of efforts by William A. Gahl, M.D., Ph.D., clinical director at the National Human Genome Research Institute (NHGRI), part of the NIH; John I. Gallin, M.D., director of the NIH Clinical Center; and Stephen Groft, Pharm.D., director of the NIH Office of Rare Diseases (ORD). With the program infrastructure now in place, the program is ready to accept patients, the first of which is expected to be seen in July 2008.

“The NIH Clinical Center, the nation’s clinical research hospital, provides an extraordinary environment for excellence in both patient care and collaborative clinical investigation,” said Dr. Gallin. “This new program will capitalize on a rich set of skills already at the Clinical Center to help patients with unusual medical conditions. These patients partner with us in clinical research to identify new diseases or new treatment.”

To evaluate each patient enrolled in the new program, NIH will enlist the expertise of more than 25 of its senior attending physicians, whose specialties include endocrinology, immunology, oncology, dermatology, dentistry, cardiology and genetics. Dr. Gahl, who is an expert on rare genetic diseases, will serve as director of the new program.

“We have developed a stringent referral process to ensure this program deals with those cases that have truly confounded medical experts,” Dr. Gahl said. “We will be very

selective when it comes to patient eligibility. Our focus is strictly on conditions that have not been diagnosed.”

To be considered for this NIH pilot program, a patient must be referred by a physician and provide all medical records and diagnostic test results requested by NIH. Patients who meet the program’s criteria — as many as 100 each year — will then be asked to undergo additional evaluation during a visit to the NIH Clinical Center that may take up to a week.

Two nurse practitioners will manage patient recruitment and logistics for the new program, which will utilize existing facilities and staff already at the NIH Clinical Center, NHGRI and ORD. Funding for the program includes \$280,000 per year from the ORD.

In organizing the Undiagnosed Diseases Program, NIH has reached out to patient advocacy groups that often serve as a source of information and support for people struggling with mysterious ailments. “We hope to build upon our strong working relationships with many patient advocacy groups. These organizations provide a crucial link in our nation’s efforts to improve human health through biomedical research,” said Dr. Groft. “We hope that this new partnership of NIH researchers, advocacy groups and patients will give hope for many Americans who now face troubling medical symptoms with no clear diagnosis.”

For more information about the Undiagnosed Diseases Program, go to: <http://rarediseases.info.nih.gov/Undiagnosed>. Physicians and patients with specific inquiries may call the NIH Clinical Center clinical information research line, at 1-866-444-8806.

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The NIH Clinical Center (CC) is the clinical research hospital for the National Institutes of Health. Through clinical research, physician-investigators translate laboratory discoveries into better treatments, therapies and interventions to improve the nation's health. For more information, visit <http://clinicalcenter.nih.gov>.

The NIH Office of Rare Diseases stimulates and coordinates research on rare diseases and supports research to respond to the needs of patients, healthcare providers and the research communities involved in the care, treatment, and evaluation of products for the prevention, diagnosis, or treatment of these conditions. For more information about ORD and its programs, visit rarediseases.info.nih.gov.

The Office of the Director, the central office at NIH, is responsible for setting policy for NIH, which includes 27 Institutes and Centers. This involves planning, managing, and coordinating the programs and activities of all NIH components. The Office of the Director also includes program offices which are responsible for stimulating specific areas of research throughout NIH. Additional information is available at <http://www.nih.gov/icd/od/index.htm>.

NHGRI is one of the 27 institutes and centers at the NIH, an agency of the Department of Health and Human Services. The NHGRI Division of Intramural Research develops and implements technology to understand, diagnose and treat genomic and genetic diseases. Additional information about NHGRI can be found at its Web site, www.genome.gov.

The National Institutes of Health — “The Nation’s Medical Research Agency” — includes 27 institutes and centers, and is a component of the U.S. Department of Health and Human Services. It is the primary federal agency for conducting and supporting basic, clinical and translational medical research, and it investigates the causes, treatments and cures for both common and rare diseases. For more, visit www.nih.gov.

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