Undiagnosed Diseases Network launches online application portal

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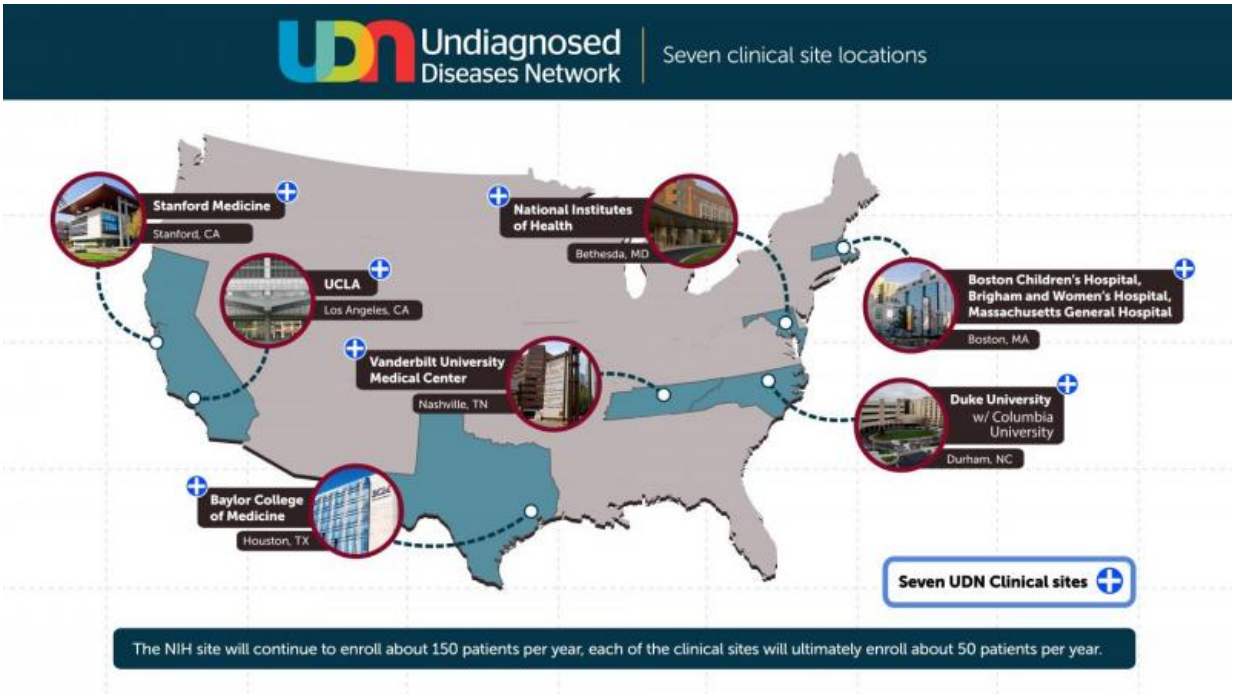

This map shows the location of the seven clinical sites located throughout the United States. Credit: Ernesto Del Aguila, NHGRI

The Undiagnosed Diseases Network (UDN), a clinical research initiative of the National Institutes of Health, has opened an online patient application portal called the UDN Gateway. Introduction of this application system sets the stage for the network to advance its core mission: to diagnose patients who suffer from conditions that even

skilled physicians have been unable to diagnose despite extensive clinical investigation. These diseases are difficult for doctors to diagnose because they are rarely seen, have not previously been described or are unrecognized forms of more common diseases.

The new system streamlines the application process. All applications for the UDN will go through the Gateway, rather than through individual clinical sites in the network. The Gateway replaces what had previously been a paper-and-mail application process for the NIH Undiagnosed Diseases Program (UDP), which is now part of the UDN.

"Although undiagnosed conditions present an array of challenges for clinicians, once identified, they may lead to treatments for individual patients. They also may lead to new, generalizable medical insights," said James M. Anderson, M.D., Ph.D., director of NIH's Division of Program Coordination, Planning, and Strategic Initiatives (DPCPSI), which provides financial support and joint leadership for the network via the NIH Common Fund. "The UDN Gateway will provide patients and their families access to the nation's leading diagnostic teams and sophisticated diagnostic tools."

The UDN grew out of the success of the Undiagnosed Diseases Program at the NIH Clinical Center in Bethesda, Maryland. Since its 2008 launch, the UDP has reviewed more than 3,100 applications from patients around the world. More than 800 patients have been enrolled for a one-week evaluation. While approximately 25 percent of those have received some level of clinical, molecular or biochemical diagnosis, many patients remain undiagnosed.

By adding six additional clinical sites to the original NIH UDP, the UDN will broaden its diagnostic expertise while expanding the opportunity for patients to participate. These additional clinical sites are:

- Baylor College of Medicine, Houston;
- Duke Medical Center, Durham, North Carolina, with Columbia University, New York City;
- Harvard Teaching Hospitals (Brigham and Women's Hospital, Boston Children's Hospital, Massachusetts General Hospital), Boston;
- Stanford Medical Center, Stanford, California; University of California at Los Angeles Medical Center; and
- Vanderbilt University Medical Center, Nashville, Tennessee.

By the summer of 2017, each new clinical site will accept about 50 patients per year. The network has also brought on board two DNA sequencing facilities. One is at the Baylor College of Medicine, and the other is at the HudsonAlpha Institute for Biotechnology in Huntsville, Alabama, with Illumina in San Diego.

The broader geographic distribution of sites in the UDN is intended to better serve patients. To support this collaboration on undiagnosed diseases, the UDN Coordinating Center at Harvard Medical School Department of Biomedical Informatics (DBMI) created the UDN Gateway as a centralized online application site.

"The Gateway is an important part of the infrastructure that we are establishing for the UDN," said Rachel Ramoni, D.M.D., Ph.D. "Our goal is to match 21st century medicine with 21st century technology by creating a comprehensive and streamlined online application process." Dr. Ramoni serves as executive director and a principal investigator of the UDN Coordinating Center at DBMI.

Those who are accepted will be seen by researchers and physicians from a wide array of medical specialties and may have their DNA sequenced to detect variations in genes that may underpin their disorders.

"The UDN aims to improve the level of diagnosis and care for patients with undiagnosed diseases," said Anastasia Wise, Ph.D., program director, NHGRI Division of Genomic Medicine and co-coordinator for the NIH Common Fund's Undiagnosed Diseases Network. "Based upon the experience of the NIH UDP, we know that the need and potential are great. The UDN Gateway will expand our ability to connect with patients who may benefit from the UDN. We want to make it as easy as possible for patients and their families to apply to participate in the network."

More information: www.genome.gov/27562472

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