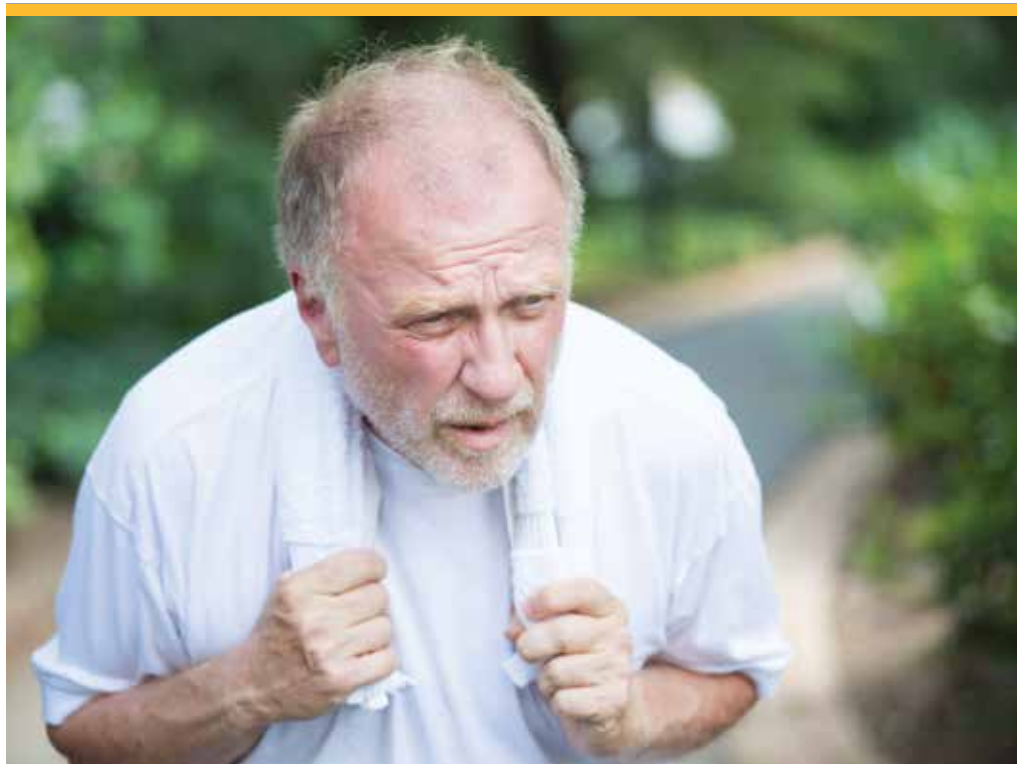


UCLA's Undiagnosed Diseases Network offers advanced diagnostic services at no cost



Patients who suffer a variety of seemingly unrelated and chronic symptoms — such as a heart problem with an autoimmune disorder, or learning difficulties with low muscle tone and hearing issues — often see a variety of specialists and undergo a number of tests in trying to determine a diagnosis. But many are left with no definitive answers after a diagnostic process that can last months or even years.

While rare diseases are individually uncommon, by definition affecting fewer than 200,000 people nationally, in aggregate they affect between 25 and 30 million people in the United States — close to 10 percent of the population.

In some cases, genetic testing provides answers and leads to a diagnosis for these patients. But for many, paying for a genomic sequencing test and appointments with specialists across a variety of disciplines is out of reach. Thanks to a new program, funded with \$43 million from the National Institutes of Health, more patients may be able to benefit from advanced diagnostic services.

As part of the nationwide Undiagnosed Diseases Network, UCLA's Department of Human Genetics and other clinical departments are now seeing select patients at no cost to them or to their insurance. These patients undergo a complete clinical evaluation with genetic testing in an attempt to solve their medical mystery.

Bedside to bench approach means bringing findings back to the lab

The goal is for patients accepted to UCLA's Undiagnosed Diseases Network to leave their weeklong visit with a diagnosis in hand. "Just identifying what's going on is very empowering," says Katrina Dipple, MD, PhD, co-principal investigator in UCLA's Undiagnosed Diseases Network.

But that's not where the work ends for participating investigators. After the clinical visit, they'll bring their findings back to the lab to conduct basic and translational research. "Many diagnoses won't have a treatment attached to them early on, but that is the long-term goal," Dr. Dipple says. "That's what the whole network is about."

With a research basis — and at no cost to the patient or insurance — and access to a nationwide network of experts, "we just have so much more power to solve each patient's diagnostic dilemma," Dr. Dipple continues. "Our goal is to shorten each patient's diagnostic odyssey so that we can find the cause and treatment for their disease."

Applying to the program

The Undiagnosed Diseases Network incorporates six clinical sites across the country, including UCLA. Patients from 1 month to 99 years old can apply to the program online at www.genetics.ucla.edu/udn.

As part of the application, they'll be asked to provide basic demographic information, as well as their medical history and results from previous clinical evaluations. No application will be considered without a referral letter from a primary-care physician that includes a summary of the patient's medical issues along with previous diagnoses and other clinical information.

Once the network's coordinating center has reviewed the application, they will forward it to one of the participating clinical sites, chosen by the site's specialty or geographic proximity to the patient. The UCLA team will review all applications they receive and accept patients on a rolling basis — 35 people the first year, and 50 the second and third years — who they believe they can help find a diagnosis.

Working to reach a diagnosis

Patients accepted to UCLA's Undiagnosed Diseases Network will first undergo genetic testing. For pediatric patients, the genomes of both parents will also be sequenced, and if there are other affected family members, the program may sequence those as well.

With the patients' genetic profile compiled, UCLA's investigators will arrange a five- to seven-day clinical visit, scheduling additional tests and consultations with immunologists, neurologists, cardiologists and other specialists as warranted. Before the visit, the UCLA team will present the case to the larger national network so other experts can weigh in.

The goal at the conclusion of the weeklong visit is to give patients and their families a diagnosis with a complete summary of findings before returning to local providers for follow-up care. Genetic counselors will work with the families to help them understand test results and how the diagnosis will impact their lives, and a patient advocate will help them connect to people with similar conditions.

Participating Physicians

Katrina Dipple, MD, PhD

Co-principal Investigator, UCLA Undiagnosed Diseases Network
Professor of Pediatrics and Human Genetics

Stanley Nelson, MD

Co-principal Investigator, UCLA Undiagnosed Diseases Network
Professor of Human Genetics and Psychiatry

Christina Palmer, CGC, PhD

Co-principal Investigator, UCLA Undiagnosed Diseases Network
Professor-in-Residence, Human Genetics and Psychiatry

Eric Vilain, MD, PhD

Co-principal Investigator, UCLA Undiagnosed Diseases Network
Chair of the Medical Genetics Clinic
Professor of Pediatrics, Urology and Human Genetics

Contact Information

Department of Human Genetics
David Geffen School
of Medicine at UCLA
Gonda Neuroscience and Genetics
Research Center
695 Charles E. Young Drive South
Los Angeles, CA 90095
www.genetics.ucla.edu/udn

Study coordinators:

Naghmeh Dorrani
(310) 825-8084 phone
ndorrani@mednet.ucla.edu

Matthew Herzog
(310) 794-6395 phone
mherzog@mednet.ucla.edu