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Scripps launches whole genome sequencing study to find root causes of idiopathic diseases

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Scripps Health announced today it has launched an innovative clinical research study that is using whole genome sequencing to help determine the causes of idiopathic human diseases -- those serious, rare and perplexing health conditions that defy a diagnosis or are unresponsive to standard treatments.

The use of whole genome sequencing is a novel approach to accurately diagnosing and treating rare medical cases. Whole genome sequencing provides information on all 6 billion data points in the human genome. This is more than 3,000 times more data than the current standard in sequencing technology – DNA arrays – which offers genotypic information on up to 1 million base pairs.

"We're now at a turning point in medicine, where we can look at a person's entire genetic code and examine their biologic underpinnings in a way that was never possible before," said Dr. Eric J. Topol, chief academic officer of Scripps Health and the study's principal investigator. "For patients who have searched in vain for the cause of their unexplained illnesses, whole genome sequencing could provide important and potentially life-saving answers."

The use of genome sequencing took a major leap forward last year, when doctors and scientists at the Medical College of Wisconsin and Children's Hospital of Wisconsin announced the first successful use of

the technique to diagnose and treat a patient. Four-year-old Nic Volker had an undiagnosed intestinal illness that doctors considered terminal. Genome sequencing revealed a single mutation responsible for Nic's disease, leading to the umbilical blood transplant that saved his life.

To date, only a select few institutions across America are using whole genome sequencing for purposes such as diagnosing patients, predicting disease risk and understanding health. The technology is gradually becoming more accessible to the medical community, as the cost and time involved in sequencing a whole human genome has substantially declined – though the time involved in interpreting results is still substantial.

The new Scripps study – dubbed IDIOM, short for Idiopathic Disease of Man – is believed to be the first whole genome sequencing study of adult idiopathic disease to date. Scripps plans to collaborate with other medical and research centers involved in different types of whole genome sequencing research by sharing key findings.

Scripps anticipates its new research will initially include up to six participants, with most to be referred by Scripps-affiliated physicians. Two of the study's participants are expected to be pediatric patients from Saint Francis Hospital in Tulsa, Okla. Data analysis will be provided by the Scripps Translational Science Institute (STSI), a major research initiative at Scripps Health involving a collaboration with The Scripps Research Institute.

"Making sense of the immense amount of data associated with the human genome is one of the biggest challenges with the application of whole genome sequencing in clinical settings," said Dr. Nicholas Schork, director of biostatistics and bioinformatics with STSI. "At Scripps, we have amassed what we believe is one of the premier teams of genome analyzers in the world, who have developed expertise in creating the

methodologies that are needed to sift through and dissect the genetic basis of remarkably complex human traits and diseases."

Scripps has assembled a panel of physicians from a broad range of medical specialties to review and select patient cases for the IDIOM study. The panel is expected to begin reviewing its first potential patient cases in late October. Selection of study participants will not be limited to any specific disease types, age ranges, gender or ethnicity.

To be eligible for the study, each patient and both of their parents must be available to provide saliva and blood samples. Scripps' IDIOM study investigators will consult with participants and their doctors on diagnosis and possible care options. While the study's aim is to find the root cause of each participant's idiopathic disease, patients in the study are not guaranteed that whole genome sequencing will resolve their case.

According to a recent Institute of Medicine (IOM) report, rare diseases collectively affect millions of Americans of all ages. Of the 5,000 to 8,000 diseases categorized as rare, some affect only a few individuals, while others affect thousands. Most of these diseases are genetic or have a genetic component. The National Institutes of Health (NIH) has recognized a one- to five-year timeframe to reach proper diagnosis for 33 percent of patients with rare disorders and more than five years for 15 percent of such patients.

"Studies such as IDIOM will not only help fill a significant knowledge gap with respect to the causes of rare diseases, they will also help 'test-drive' the use of whole genome sequencing in the clinical setting more broadly," said Dr. Cinnamon Bloss, an assistant professor and genomic scientist with STSI. "By reaching out to and including physicians in the process of selecting cases, interpreting and returning genomic results, we hope to help facilitate greater integration of genomics and medicine."

Provided by Scripps Health