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Scientists deploy data analysis to identify subtypes of common disease

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A new publication from scientists at the Icahn School of Medicine at Mount Sinai offers a glimpse of precision medicine in action, with a massive data analysis project that identified clinically and genetically distinct subtypes of patients with type 2 diabetes. This work not only points to the possibility for more tailored diagnosis and treatment of type 2 diabetes in the future, but also reveals a novel approach that can be applied to virtually any disease.

The paper, published today in *Science Translational Medicine*, describes a complex network analysis of electronic medical records (EMRs) and genotype data for more than 11,000 patients. Patients were grouped into three distinct subtypes based on EMR data, followed by genomic analysis pinpointing common genetic variants representative of each subtype. These subtypes were associated with different clinical characteristics. Patients were more likely to suffer diabetic nephropathy and retinopathy in subtype 1; cancer and cardiovascular disease in subtype 2; and neurological disease, allergies, and HIV infections in subtype 3. For each subtype, the researchers discovered unique genetic variants in hundreds of genes.

"This project demonstrates the very real promise of precision medicine to improve healthcare by tailoring diagnosis and treatment to each patient, as well as by learning from each patient," said Joel Dudley, PhD, senior author on the paper and Director of Biomedical Informatics at the Icahn School of Medicine at Mount Sinai. "It is absolutely encouraging that we were able to paint a much higher-resolution understanding for a

common and complex disease that has long stymied the biomedical community with its heterogeneity. I look forward to seeing what we can accomplish for other patient populations."

Type 2 diabetes has quickly become a leading cause of death, and the World Health Organization estimates that 8% of the global adult population has the disease. The medical community has struggled to diagnose and treat type 2 diabetes because it presents with many different symptoms and a wide range of associated complications. It has long been thought that the disease, like cancer, could be treated more successfully if patients could be grouped into clinically distinct subtypes with more specific prognoses.

"Our approach demonstrates the potential to unlock clinically meaningful patient population subgroups from the wealth of information that is accumulating in electronic medical record systems. The unique genetic component of this study yielded high-priority variants for follow-up study in patients with type 2 diabetes," said Dr. Ronald Tamler, co-author of the study and Director of the Mount Sinai Clinical Diabetes Institute, within the Mount Sinai Health System. "The team's results suggest an attractive alternative to the kind of large-scale, narrow phenotype studies that have produced limited success in explaining common, complex disease."

More information: "Identification of type 2 diabetes subgroups through topological analysis of patient similarity," stm.sciencemag.org/lookup/doi/ … scitranslmed.aaa9364

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