PMedIC Seminar Series

Applying metabolomics in the characterization of rare disease: Experiences from the NIH Undiagnosed Diseases Network

Wednesday, June 9, 2021
3:00 pm – 4:00 p.m.
Zoom

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The NIH Common Fund's Undiagnosed Diseases Network (UDN) was established to accelerate the diagnosis and clinical management of rare or previously unrecognized diseases, and to advance research in disease mechanisms. The UDN is composed of multiple clinical sites around the United States and multiple research cores, including DNA sequencing, model organisms and metabolomics. As the Metabolomics Core for Phase I of the UDN, our role was to provide comprehensive untargeted measurements to identify qualitative and quantitative changes of metabolites (metabolomics) and lipids (lipidomics) in biofluids from probands to assist in the evaluation and/or identification of the causes of rare and undiagnosed diseases. In contrast to identifying inborn errors of metabolism, the metabolic changes associated with rare diseases may be more subtle, consisting of complex patterns of minor changes of a large number of analytes rather than a few significant outliers. In addition, due to the rare nature of these disorders, the number of individuals with a given phenotype is usually limited to one or just a few, precluding the use of the balanced study designs typically used in metabolomics. In this presentation, we will discuss our experiences in applying metabolomics and lipidomics measurements in the study of individuals with rare disease and the associated unique challenges.

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