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Metabolomics adds function to precision diagnosis

DNA sequencing provides a candidate list of genes/mutations which account for an individual's family disorder, personal disorder and risk for medical disorders. It is not uncommon to identify DNA variants (VUS) which are difficult to interpret with regard to disease causation, since such interpretation relies heavily on disease data bases (HMGD/clinvar/private). The measurement of blood analytes (750 reality of 1500 detected) examines biochemical function on an individual basis. Three examples of metabolomics utility for DNA sequence interpretation will be presented. These include: R/O VUS as disease causative; accurately diagnosing inborn error of metabolism both known and newly discovered and; identifying pathways and specific gene mutations in twin studies that were undetected by standard bioinformatics DNA based search tools. These results have led us to utilize both metabolomics and whole genome sequence to achieve precision diagnosis and direct therapy interactions.

Biography

C Thomas Caskey is a Professor at the Department of Molecular & Human Genetics, Baylor College of Medicine. In 1994, he became Sr. VP of Drug & Vaccine Development at Merck. He is a Member of NAS, NAM and Royal Society of Canada. He is currently directing a program of precision medicine with Young Presidents Organization (YPO). He is a Consultant at Human Longevity and Member of Board of Metabolon. His current research focuses on the application of whole genome sequence and metabolomics of individuals for disease risk and its prevention.

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