Design and implementation of an information technology infrastructure for clinical genomics reporting and decision support

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This talk will discuss the design and implementation of an information technology infrastructure to support clinical use of genomics, specifically in the area of oncology. The “last mile” in taking processed variant calls and producing a useful, annotated clinical report with therapeutic recommendations is critical for enabling the clinical use of genomics data. Generating, delivering, and updating a clinical genomic sequencing report that a physician can use in routine care has proven to be a challenge, driven by reliance on outdated software tools. To bring genomics data to point of care, healthcare providers must be able to handle three types of data with very different properties and requirements: Evolving biomedical knowledge, complex medical data, and high-volume genomics data. Syapse has created a software platform, based on semantic computing principles, that enables the integration of all three data types for the purposes of clinical reporting. The author here will present the results of clinical pilots focused on maintenance of an evolving variant interpretation knowledge base, sharing of variant interpretation across institutions, and the use of this knowledge base to automate clinical genomic report generation and updating, as knowledge changes. The interpretation of identified variants through a shared knowledge base, automated assembly of annotated clinical reports and delivery of reports to clinicians at point of care will be described. Results of a pilot program to integrate these technologies into practice within an integrated community health care system will be presented. Physician adoption of a web-based, interactive clinical genome report format that incorporates clinical care guidelines with genomics report data will be discussed.

Biography

Jonathan Hirsch is the Founder and President of Syapse, a software company transforming healthcare by bringing omics into routine medical use. He is a member of the UCSF Technology Advisory Group, and a member of the Steering Committee of Free the Data, an effort to crowdfund the interpretation of cancer genes. Earlier in his career, he worked in Neuroscience Commercial Development at Abbott Laboratories, where he developed strategies to fund drug development through partnerships and private equity financing. His research at the Center for Molecular Neurobiology at the University of Chicago helped establish the effect of exercise on promoting hippocampal neurogenesis and combating Alzheimer’s disease. He received an MSc in Neuroscience from Stanford University, and an AB in Biology and Political Philosophy from the University of Chicago.

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