K1 - Towards Precision Medicine

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Born in Scotland, Prof Ashley graduated with first class Honors in Physiology and Medicine from the University of Glasgow. He completed medical residency and a PhD in molecular physiology at the University of Oxford before moving to Stanford University where he trained in cardiology and advanced heart failure, joining the faculty in 2006. His group is focused on precision medicine. In 2010, he led the team that carried out the first clinical interpretation of a human genome. The paper published in the Lancet was the focus of over 300 news stories, became one of the most cited articles in clinical medicine that year, and was featured in the Genome Exhibition at the Smithsonian in DC. The team extended the approach in 2011 to a family of four and now routinely applies genome sequencing to the diagnosis of patients at Stanford hospital where Prof Ashley directs the Clinical Genome Service and the Center for Inherited Cardiovascular Disease. In 2014, Prof Ashley became co-chair of the steering committee of the NIH Undiagnosed Diseases Network. In 2013, Prof Ashley was recognized by the White House Office of Science and Technology Policy for his contributions to Personalized Medicine. He is recipient of the National Innovation Award from the American Heart Association as well as an NIH Director's New Innovator Award. He works with many Silicon Valley companies and investors. He is Principal Investigator of the MyHeart Counts cardiovascular health study, launched in collaboration with Apple in 2015. In 2016, he was part of the winning team of the $75m One Brave Idea competition funded by Google, the AHA and Astra Zeneca. Father to three young Americans, in his "spare" time, he tries to understand American football, plays the saxophone in a jazz quartet, and conducts research on the health benefits of single malt Scotch whisky.

The goal of the session is to introduce the concept of precision medicine and highlight some of the critical contributions made by clinical genomics to its origination and evolution. The session will briefly recap rapid advancements in genomic technology and illustrate the utility of genomics for clinical medicine using specific patient examples. Some of the essential algorithmic approaches to the interpretation of human genomes will be discussed. Areas where current short read technologies perform well are discussed, as well as areas where new approaches are required. In the context of precision and accuracy in genomics, newer technologies such as long read sequencing and new algorithms for improving test performance in complex areas of the genome will be introduced. The use of gold standards in genomics and the limitations of the human reference genome will be discussed. Finally, the session will highlight the near-term future of clinical genomics. Throughout the talk, illustrative patient examples are used including those from the Undiagnosed Diseases Network.

At the end of this session, participants should:
1. Understand what is meant by precision medicine and be able to provide examples;
2. Understand the opportunity and challenge represented by our ability to sequence whole human genomes at scale for clinical medicine;
3. Understand areas of need in the development of clinical genomics;
4. Understand the power and limitations of the human reference genome;
5. Understand the current state of the art in the application of clinical genomics to rare disease; and
6. Understand how genomics will move from rare disease to affect every patient in the healthcare system.