



BIOINFORMATICS SEMINAR

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Methods and Applications of Long-read Sequencing in Medical Genomics

Long-read sequencing technologies, such as those from 10X Genomics, Bionano Genomics, Pacific Biosciences and Oxford Nanopore, have now enabled the generation of sequenced fragments that are 10kb or longer, allowing for the interrogation of complex genomic regions that are otherwise inaccessible by short-read sequencing. Here I describe the development of novel computational approaches to best leverage these new generations of sequencing technology, and the application of these approaches in the study of human diseases.

BIOGRAPHY

Education

B.S. (Biochemistry & Molecular Biology), Peking University, 2000

M.S. (Tumor Biology), Mayo Clinic, 2002

Ph.D. (Microbiology & Computational Biology), University of Washington, 2005

Postgraduate Training

Postdoctoral Fellow, University of Pennsylvania, 2006-2008

Postdoctoral Fellow, The Children's Hospital of Philadelphia, 2008-2010

Research Expertise

The research in our laboratory focuses on the development of bioinformatics methods to improve our understanding of the genetic basis of human diseases, and the integration of electronic health records and genomic information to facilitate genomic medicine on scale. Current projects include the development of computational tools to call structural variants and DNA modifications from long-read sequencing data, the identification/prioritization of disease-relevant genetic mutations, the use of deep neural network to predict prognosis and optimize therapy for patients with cancer, the application of systems biology approaches on single-cell gene expression data, and the data mining on electronic health records to predict genetic syndromes and causal genes.

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