

**Victorian Centre for Biostatistics****SEMINAR****Thursday 28<sup>th</sup> March 2019****9.30am to 10.30am****Melbourne School of Population and Global Health****Room 515, 207 Bouverie St, Carlton****Effects of DNA variation on single-cell gene expression****Dr Davis McCarthy****St Vincent's Institute of Medical Research**

How can we understand the effects of DNA variation on gene expression in single cells? What sort of studies should we conduct and what computational tools do we need for them to succeed? Following a short introduction to the field of single-cell biology I will describe some of the opportunities available to improve expression quantitative trait locus mapping with single-cell data and to gain new views of the structure within clonal cell populations at unprecedented resolution. I'll also discuss the challenges inherent in scaling up single-cell data generation to assay cells from tens to hundreds of individuals. Throughout, I will provide details of the hierarchical and variational Bayes approaches we have used to tackle these problems. More specifically, I will demonstrate approaches for inferring clonal structure in a cell population by combining bulk exome and scRNA-seq data. I will present a two-step statistical method for the assignment of cells to a clonal tree structure, utilising detected mutations in sparse single-cell expression data.

**Dr Davis McCarthy** started in Bioinformatics as a UROP student (and Honours student, and RA) with Gordon Smyth in the WEHI Bioinformatics Division. He worked on differential expression methods for RNA-seq data, most notably the edgeR package. He completed a DPhil in Statistics in Oxford under the supervision of Peter Donnelly, before undertaking a postdoc in Oliver Stegle's group at the European Bioinformatics Institute in Cambridge. At EBI, Davis worked on single-cell methods development and on projects linking DNA variation to single-cell gene expression. He returned to Melbourne in late 2018 to start the Bioinformatics and Cellular Genomics group at St Vincent's Institute, joint with the Melbourne Integrative Genomics unit at the University of Melbourne.

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