

Presentation title: Detecting mutations using next-generation sequencing

Date: Monday 4 July 2011

Abstract

As part of the International Cancer Genome Consortium the Queensland Centre for Medical Genomics has established laboratory and computational infrastructure and expertise to analyse whole human genomes for the presence of DNA and RNA mutations and epigenetic marks underlying cancer. This talk will describe and discuss various strategies for identifying DNA mutations from short read sequence data, including SNPs, indels, copy number changes, structural aberrations and rearrangements. I will present the analysis approaches that have been implemented at the QCMG and how we use these to distinguish germline variants from somatic mutations potentially driving tumourigenesis.