

## BiRC Seminar - open to all

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## Title: Making sense of population next-generation sequence data

Time: Friday 9 March, 2012, 14:15 - 15:00

Place: BiRC, Aud. 223, Building 1110, C. F. Møllers Allé 8

## Abstract:

Sequencing of the first reference genomes for many species, including humans, was completed at great expense using automated Sanger sequencing to provide high quality long read DNA sequence. Sanger sequencing has now largely been superseded by next generation sequencing, which refers to a set of technologies and sample preparation techniques which have in common their ability to produce short read sequence data in a very high throughput manner at relatively low cost. This technology is ideally suited to re-sequence 1000s of individuals from multiple sub-populations in each species in order to provide a complete catalogue of genomic variation in that species, such as is being undertaken by the 1000 genomes sequencing consortium, and to search for trait associated variation not captured by genotyping arrays.

However, next generation sequence data is noisy, with the rate of base-calling error greatly exceeding the rate at which novel mutations occur. We have developed a suite of tools which model next generation sequence data at the population level for inference from next generation sequence data, including: detection and genotyping of indels; detection and genotyping of copy number variation; association of copy number variation with phenotype from exome sequence data; identification of highly differentiated SNP and indel sites between populations. I will present data which show that our methods have substantially greater sensitivity than other methods, at the same false discovery rate. Moreover, our methods have been designed to work at arbitrary levels of ploidy, making them ideally suited to applications in crop science.

After the seminar there will be beer/soda/coffee and chips in the coffee room on the 4<sup>th</sup> floor.

http://birc.au.dk/activities/seminar-series/

