



presents Guest Speaker:

## **Inanc Birol**

**Genome Sciences Centre, BC Cancer Agency  
School of Computing Science, Simon Fraser University**

### *Haploid Assembly of Diploid Genomes*

Most assembly algorithms in use have an implicit assumption that a sequenced nucleic acid has a haploid-like structure, or at worst that the sequence representing a genomic location is present with a high similarity in the read data. This assumption holds nicely in the case of model organisms, which are usually inbred to reduce haplotypic diversity, and is even not challenged significantly in the case of human samples because of an evolutionarily recent population bottleneck our species experienced. However, when an organism of interest has a diploid genome with a pronounced distance between haplotypes, or when an environmental sample is investigated that represents a collection of similar species, the assumption fails. Furthermore, read lengths of popular high throughput sequencing technologies, and even the spatial associations brought by paired end reads are often not enough to completely de-phase the assembled sequences. In this talk, I will discuss the particular challenges one faces during de novo assembly of such datasets, and describe how we approach to their solution within the ABySS framework. ABySS is a short read assembly tool, particularly developed to address large scale assembly problems. I will introduce how it handles distance information to extend and scaffold contigs, and how it represents haplotypic information in its output. I will demonstrate its performance on the assembly of the mountain pine beetle genome.

Introductory speaker (10 mins):

**Olena Morozova**, Marra lab, GSC

*A wide spectrum of somatic mutation in high-risk neuroblastoma*

**Thursday, October 13, 2011, 6:00 pm**

Gordon and Leslie Diamond Family Theatre,  
BC Cancer Research Centre,  
675 West 10th Avenue



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