Essay on Next Generation Sequencing and Metagenomics

Bioinformatics course

Spring, 2013

In the curriculum for the master course *Bioinformatics* at Chalmers there are two topics *Next Generation Sequencing* and *Metagenomics* that will not be taught in regular lectures in Spring Semester 2013. Instead you will be asked to write a short essay (roughly 2+2 pages long) introducing the two topics and discussing their connection, i.e. how next generation sequencing is used as a key technology in metagenomics.

1. The essay should start with a short introduction to a couple of the main next generation sequencing platforms and some of the challenges that arise in the interpretation of data from these. Focus either on:

   resequencing, that is the sequencing of a genome with a previously sequenced reference genome. A major application of resequencing is identification of disease-causing mutations in the human genome.

   de novo sequencing, that is the characterization of a completely new genome for an organisms where there is no or little previously knowledge.

2. Metagenomics is a technique where microorganisms are studied by sequencing DNA directly from a sample. This means that single individual organisms are ignored. Instead, metagenomics investigates the entire genetic composition of a community of microorganisms. Samples typically studied in metagenomics includemicroorganisms in soil, bacteria in he human gut, for example from both healthy or sick patient(s). Discuss metagenomics, its dependence on next generation DNA sequencing and the challenges in analyzing the resulting metagenomic data. Identify also (at least) one application of metagenomics and next generation sequencing and give a brief summary of it (them).

Start your reading with:

- Loman et al, High-throughput bacterial genome sequencing: an embarrassment of choice, a world of opportunity, Nature Reviews Microbiology, 10, 2012.
- Hugenholtz & Tyson, Microbiology: Metagenomics, Nature Q&A Microbiology, Vol. 455, 2008.
- Horner et al, Bioinformatics approaches for genomics and post genomics applications of next-generation sequencing. Brief Bioinform. 2010 Mar; 11(2):181-97.
- Metzker, Sequencing technologies - the next generation, Nat Rev Genet, 2010, 11(1): 31-46.
- Mardis, The impact of next-generation sequencing technology on genetics, Trends in Genetics, Volume 24, Issue 3, March 2008, Pages 133-141.
- Pop & Salzberg, Bioinformatics challenges of new sequencing technology. Trends in Genetics, Volume 24, Issue 3, March 2008, Pages 142–149.