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Production-scale Deployment of Next-generation Sequencing Instruments

Status	Current
Competition	Development of New Technologies Competition
Sector	Development of New Technologies
Genome Centre	Genome British Columbia
Project Leader	Marco Marra, Steven Jones & Robert Holt

Project Description

The genome of an organism represents its entire hereditary information stored in the form of a DNA code. Recent developments in DNA sequencing technologies are changing the way scientists collect and interpret genomic data. At the Genome Sciences Centre (GSC), we are incorporating technology advances that are positively impacting the cost, quality and quantity of genomic data. For example, the human genome project cost $3 billion and took about 15 years to complete. Amazingly, and only a short time after the completion of the human genome sequencing project, the latest technology advances are aimed at soon making the “$1000 genome” a possibility. The revolutionary advances represented by the latest technology are not only applicable to sequencing entire genomes, but also provide critical infrastructure for researchers to understand genetic-based diseases such as cancer, diabetes, cardiovascular disease and mental illness, as well as current environmental issues such as bioremediation and forest pest infestations. For example, with this new technology, we can for the very first time imagine re-sequencing human genomes, and using this ability to compare genome regions in healthy versus diseased individuals to identify mutated genes that cause disease. We can also sequence populations of microorganisms in soil or water samples, or the genomes of species of agricultural importance. This type of information can be applied to more effective disease treatment, prevention strategies, or personalized drug therapies in the case of humans, and more effective pest control in agriculture or forestry, or safer methods to clean up oil contamination using naturally occurring microbes, to name only a few applications.

This technology differs greatly from the current sequencing machines and methods and involves using new chemicals, developing innovative methods, creating validated protocols, and developing new computational approaches to process and interpret the information produced by the new machines. This has been a major focus at the GSC since we acquired an early access sequencing machine (called a Genome Analyzer) from Illumina in November, 2006. We have worked extensively with Illumina to generate a substantial amount of high quality sequencing data (over 30 billion base pairs of sequence), and have been extremely impressed by the quantity of high quality data that can be generated at low cost and in relatively short time frames with this technology. To lend some perspective to the time and cost savings involved with “next generation sequencers”, these instruments can generate over 1 billion base pairs of sequence

data every three days; whereas the current generation of devices generate only ~3,000,000 base pairs every three days. This difference is profound - one Illumina sequencing device has the potential to determine, in a single year, an amount of DNA sequence approximately equivalent to three times the total amount of sequence that had been generated worldwide using hundreds of machines up to January 2007. In addition to this remarkable increase in sequencing rate, the new machines are also 300 times more cost effective.

This proposal outlines a two year plan that will capitalize on the capacity of these next generation sequencing devices and develop applications which will be of significance to a broad spectrum of biological and medical studies.

We are ideally positioned to perform this work at the GSC due to our expertise in the areas of genomics, proteomics and bioinformatics. We have established a high-throughput genome sequencing facility as part of the Genome BC/Genome Canada technology platform, where we support cutting edge local, national and international projects. To remain internationally competitive, it is important for us to continue to pioneer efforts in technology development and to develop novel protocols and software in the areas listed above. This technology development is directly relevant to Genome Canada’s mandate and will help to maintain Canada as a world leader in genomics research.