BCCGN
British Columbia Clinical Genomics Network

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BCCGN Newsletter

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IN THIS ISSUE:

BCCGN News and Updates
- Preparing Physicians for Advances in genomics

Research in Focus
- Pharmacogenomics in cancer

Technology in Focus
- Massively Parallel DNA Sequencing

BCCGN Activities
- Education
- Publications
- Member Awards
- Announcements
- Events Calendar

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BCCGN News and Updates

Preparing Physicians for Advances in Genomics

BCCGN recently met with the Ministry of Health Services to discuss planning for the clinical applications of genomics that will impact the practice of medicine over the next few years. Array genomic hybridization, mutation testing for many specific rare genetic diseases, molecular analysis of cancer tissues and occasional pharmacogenetic testing are already used in regular clinical practice in Canada. In the next several years, whole genome sequencing will probably replace most other genetic tests as a result of technological advances that are rapidly reducing the cost of genomic sequencing. This will make it possible (but perhaps not desirable) for a person's complete genetic sequence to be determined once and the data stored for life, to be used as needed to ask particular diagnostic or therapeutic questions that subsequently arise. This will present new challenges as the cost of the associated data interpretation and genetic counselling will be far greater than the cost of testing.

The pressure on healthcare payers and providers to use genomic technologies appropriately and to provide accurate and timely information to patients on the basis of genomic testing is already growing, and this pressure is likely to increase greatly over the next few years. If we are to maximize the benefits, minimize risks, and achieve cost effectiveness we must develop a comprehensive strategy for appropriate use of clinical genomics based on current and developing evidence. Such a strategy must include educating physicians, nurses, genetic counsellors, other healthcare providers, administrators and policymakers as well as the general public about genomics.

Research in Focus - Pharmacogenomics in Cancer

Cancer survival in children has improved dramatically over the years and today over 80% of treated children survive. However, many survivors are left with lifelong consequences of severe adverse drug reactions as a result of their chemotherapy treatment. Dr. Rod Rassekh, a pediatric onologist at BC Children's Hospital asked the question, "Why does one child suffer severe hearing loss with Cisplatin (a highly effective anti-tumor agent), while the other does not?" To find the answer he teamed up with Dr. Colin Ross, a research scientist in the Department of Medical Genetics at UBC. This meeting paved the way for the development of a huge collaborative effort across Canada which was financed by pharmaceutical and genomic companies as well as federal and provincial granting agencies. The goal of this study was to use the latest genomic technology to identify specific mutations that cause severe Cisplatin-induced hearing loss. This would then provide an opportunity for alternative therapy with different drugs and/or doses to be used with the same success but without the damaging side-effects.

Drs. Rassekh and Ross initiated a series of experiments using a platform supported by the BC

Clinical Genomics Network, genome-wide SNP Genotyping. After collecting hundreds of DNA samples from control and cancer patients from across Canada, they used this high technology to identify 2 genes which function to inactivate the effects of Cisplatin in the ear. The results were astonishing in that they were able to demonstrate the mutation with almost 100% accuracy. This research team is now working towards the development of a predictive screening test for use in the clinic.

Technology in Focus - Massively Parallel DNA Sequencing

Modern DNA sequencing platforms allow researchers to identify genetic changes that may cause conditions such as intellectual disability or cancer. Variations in DNA sequences also contribute to our predisposition or resistance to various common diseases and our responses to drug therapy. Today's massively-parallel sequencing platforms are 10,000 times faster and produce equivalent amounts of data more than 10,000 times less expensively than the DNA sequencing technology used just a decade ago. Although several different massively-parallel sequencing technologies are available, they all are characterized by performing hundreds of thousands or millions of sequencing reactions simultaneously with extremely small volumes of reagents and monitoring the results in real time, usually by digital microphotography of fluorescent signals. Analysis of the enormous amounts of data produced by these machines requires huge amount of computer processing, storage and great bioinformatics expertise.

The BCCGN, in collaboration with the Michael Smith Genome Sciences Centre, provides clinical investigators access to massively-parallel sequencing technologies and the expertise needed to use them effectively to identify disease genes in mendelian disorders and cancer, define potential therapeutic targets, improve diagnostic precision, and understand differences in treatment responses. You can read more about this exciting technology on bccgn.ca where you can also download a fact sheet about massively-parallel genomic sequencing

BCCGN Activities

Education - BCCGN 2009 conference videos are now available on the education page of our website! So even if you missed the conference you can still benefit from our experts. These presentations give explanations of how latest genomic technologies are currently being used in clinical research.

Publications

►BCCGN article "What Can Genomics Do For Healthcare?" was published in the September 2009 issue of *BC Medical Journal*. This article highlights the use of **SNP Genotyping** in clinical research.
►BCCGN advisor Marco Marra was published in Nature 461, 809-813, 8th October 2009 regarding "Mutational evolution in a lobular breast tumour profiled at single nucleotide resolution", Shah et al.
►BCCGN consultant Tracy Tucker and colleagues published a review in Am J Hum Genet. 2009 Aug; 85(2):142-54. "Massively parallel sequencing: the next big thing in genetic medicine".
►Members of the Network published on "Perceptions of genetic discrimination among people at risk for Huntington's disease: a cross sectional survey" in the BMJ 2009 Jun 9; 338; Yvonne Bombard et al.

Member Awards

►On September 16, 2009 BCCGN co-founder Michael Hayden MD PhD received the Order of British Columbia award which recognizes provincial leaders who have served with the greatest distinction and excelled in a field of endeavor benefiting the people of BC and beyond.
►Jan Friedman MD PhD received the MedGen Teaching Award for basic science teaching. The award was presented to Prof. Friedman on November 6th at the UBC Medical Genetics Research Day.

Announcements - Rare Disease Foundation (www.rarediseasefoundation.org) is offering microgrants to expedite exploration of new ideas for understanding rare disease and improving patient care.

Events Calendar - Canadian College of Medical Geneticists 33rd Annual Scientific Meeting Nov 12-14th 2009; Canadian Association of Genetic Counsellors Annual Education Conference Nov 11–14th 2009 – Fairmont Banff Springs, Banff, AB; Poster by BCCGN

For more information please visit our website www.bccgn.ca

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