BCCGN
British Columbia Clinical Genomics Network

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BCCGN Newsletter

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BCCGN News and Updates

BCCGN Annual Conference, April 2009

BCCGN's first annual conference, "Genomics in the Clinic," was a great success. Over 170 delegates attended including family physicians, researchers, students, industry, scientists, geneticists and others. There were 69 physicians, 17 from outside the lower mainland, showing genomics is of interest to doctors throughout BC. Delegates most enjoyed learning about epigenetics and how genomic technologies can be applied in the clinic. 95% had a better understanding of genomics after the conference and will attend next year. Throughout the day distinguished speakers presented information on various genomic technologies that are now available for clinical research in B.C. and described how genomic studies have been used to improve patient care. One of the many highlights was a keynote presentation by Ian Brown, host of CBC Radio's "Talking Books" and a feature writer for the *Globe and Mail*, who spoke about his experiences as a father of a child with a rare genetic disorder and his interactions with genetic clinicians and medical researchers. A major discussion point that emerged from Ian Brown's talk was that a communication gap exists between researchers, clinicians, patients and families. Delegates were very concerned about this and saw the efforts of the BC Clinical Genomics Network as one way to help change it. Many attendees planned to seek collaborations that would permit them to participate in clinical genomics research and continue to educate themselves about genomics.

Technology in Focus - Epigenetics

Epigenetics refers to changes in a phenotype due to non-genetic factors that do not involve changes in the DNA sequence. The human genome project has identified about 25,000 genes that are regulated for when and where they are to be expressed. All cells in an organism contain the same genome but it is the epigenome that decides how the various genes are expressed at different times of life and in different parts of the body. The best example of epigenetic change is cell differentiation during embryonic development. Stem cells within a single fertilized egg divide and change over time, resulting in more than 200 different cell types such as liver, muscle, blood or hair cells. All of this happens through activating certain genes while inhibiting others without changing the DNA sequence. While there are several ways to modify the epigenome, DNA methylation has been most studied. It

involves adding a methyl group (Ch_2) to one of the base pairs on the DNA molecule. This reversible binding acts as a chemical switch turning off gene expression. The amount of DNA methylation is not only inherited but also modified during life through exposure to dietary and environmental factors. The clinical impact of epigenetic research has been to identify factors that contribute to the large epigenetic variation that exists within the population. Life experiences, environmental hazards, disease history, and nutritional status all contribute to tissue-specific epigenetic variation. Research has shown that the nutritional status of a mother can affect the epigenome of her child, and identical twins acquire discordant epigenomes throughout life. Clearly, the occurrence of disease, its age of onset, severity and prognosis are all impacted by not only our genes, but also our epigenomes. Network member Micheal Kobor has collaborated extensively to develop a unique Epigenome Profile Database that assists researchers in identifying epigenome profiles that can be used to identify biomarkers for disease or to help in identifying potential targets for therapeutic intervention.

Research in Focus – Intellectual Disability

With the help of BCCGN, Dr Anna Lehman, an enthusiastic resident, has pursued her interest in finding the molecular causes of rare genetic disorders. Together with BCCGN researchers Dr. Neal Boerkoel and Ms Christele du Souich, she evaluated a 5 year old boy with no speech, macrocephaly, and significant intellectual disability; he was found to have a duplication of part of chromosome 19. A literature search revealed two interesting facts: this duplication had never been described in healthy or disabled persons, so it was difficult to be sure if it was the cause of the child's disorder; and the duplication includes two genes important for regulating the expression of other genes. Many people in the extended family also had learning disabilities, so, in collaboration with Dr. Patrice Eydoux, they set out to see if the duplication also ran in the family. Results from a linkage study using the Affymetrix 500K Whole Genome Array and FISH showed evidence for involvement of genetic changes through 3 generations of the family. Anna then applied other new genomic technologies to find out if gene regulation was altered in the child's cells. One of the genes that showed increased expression had previously been found to impair brain development if over expressed in pig embryos. This BCCGN collaboration shows the important contribution that a clinician researcher using genomic analysis can make in not only identifying the cause of a rare disorder but also in increasing our knowledge of gene over-expression.

BCCGN Activities

Physician Survey

BCCGN recently conducted a survey of BC physicians to better understand their educational needs in genomics. Of 125 respondents, most (80%) said they have a very low knowledge of genomics, yet 39% are interested in and 46% are open to participate in clinical research. Important barriers were lack of time (37%), funding (26%), not knowing how to go about it (20%) and not having the opportunity (31%). BCCGN is changing this by educating clinicians about genomics and overcoming barriers to research.

Publications

"What Can Genomics Do For Healthcare?", an article written by BCCGN staff will be published in the September 2009 issue of *BC Medical Journal*. This article highlights the use of **SNP Genotyping** in clinical research. Similar articles highlighting other BCCGN platforms are planned for the near future.

Member Awards

Dr. Judith Hall Professor Emerita in Pediatrics and Medical Genetics, BCCGN Oversight Committee member, recently won the Faculty of Medicine Lifetime Achievement Award and **Michael Hayden**, BCCGN Co-Leader received an honorary doctorate of science degree from the University of Alberta.

Events Calendar

BCCGN is taking the pharmacogenomics and epigentics sessions from its 2009 conference "Genomics in the Clinic," to the BCMA General Meeting. Saturday June 13, 2009; 2:00 - 3:30 pm; Sheraton Wall Centre Hotel, Vancouver, BC. Speakers are Michael Hayden, Colin Ross, rod Rassekh and Michael Kobor