BCCGN
British Columbia Clinical Genomics Network

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IN THIS ISSUE:

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BCCGN Newsletter

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Physician Education

The need for continuing medical education on genomics was identified as a critical issue in BCCGN's annual physician survey. While BC Physicians are already being impacted by genomics in their practice, very few are confident in their knowledge of and their ability to respond to patients about genomics. More details can be found in our November 2011 newsletter.

As a result, BCCGN has expanded its role in providing genomic education through the following events:

Upcoming Hospital Rounds:

- March 30th, 2012: Pediatric Grand Rounds, BC Children's Hospital, Speaker: Dr. Jan Friedman
- April 18th, 2012: Hospital Rounds, Sunny Hill Health Centre, Speakers: Dr. Cris Dias, Shelin Adam and Ruth Thomas

Past presentations around BC for specialists and family practitioners, have been very well received.

For more information please check our website or contact us. For someone from BCCGN to speak with physicians in your area contact Ruth Thomas.

Upcoming Conferences

In 2012, BCCGN will be hosting three conferences! They will educate BC physicians on a wide array of genomic topics, carefully chosen to ensure that they have the highest clinical relevance.

- "Updates in Clinical Genetics - 2012" on March 2nd, 2012 from 12:30 -5:30pm in the Chan Auditorium, Child and Family Research Institute- Presented by BCCGN, the UBC Department of Medical Genetics and the Provincial Medical Genetics Programme.
- 4th Annual BCCGN Conference entitled "Clinical Genomics: The good, the bad and the ugly" on April 20th, 2012 from 8:30-4:45, at the Vancouver Convention Center.

Breaking News

BCCGN Researcher Discovers Gene for Weaver Syndrome

Dr. Bill Gibson recently discovered the major gene responsible for Weaver Syndrome, a rare genetic condition that causes overgrowth, characteristic facial features, intellectual disability and predisposition to cancer. The study was part of Canada's FORGE initiative to find the genes for rare disorders. It included the collaborative efforts of many researchers, including Professor Steven Jones from the Genome Sciences Centre and Dr. David Weaver, who first identified the syndrome in 1974. The results, which identified the gene EZH2 as the cause of Weaver syndrome in the three families studied, were published in Am. J. Hum. Genet.. EZH2 is a gene that was previously identified as mutated in leukemia, B-cell lymphoma and some other blood cancers. EZH2 is involved in gene regulation, as it helps control how DNA is packaged.

Weaver syndrome typically occurs only once in a family and arises from a new mutation in either the egg or the sperm. This study utilized genomic technologies to identify new mutations that were unique in the children with Weaver syndrome, and not present in their healthy parents.

Dr. Gibson is quoted as stating, "for the families among whom we identified the gene, this discovery definitely brings the diagnostic odyssey to a close – it's DNA confirmation that their children have Weaver syndrome." The researchers all agreed that this study illustrates the tremendous power of Next-Generation Sequencing in identifying rare disease-causing mutations, even when only a small number of individuals are involved.

Dr. Gibson continues to collaborate with the Network on a study which investigates how copy number variants (small deletions or duplications of DNA) contribute to severe obesity in children.

BCCGN Activities

Physician Workshops

BCCGN has now hosted three CME accredited genomics workshops for physicians. The one day event, "Introduction to Genomic Technologies" introduces participants to the available genomics technologies, provides lab tours, and examples on how genomics is or will be being used clinically. These workshops have been highly successful, and participants say they now have a better understanding of clinical genomics, and the technologies being used. Participants unanimously agreed that they would recommend this program to their colleagues. Please see our website for the exact date of the next workshop, tentatively scheduled for May 2012

Student Competitions 2012

For the fourth year in a row, BCCGN is holding its annual Summer Student Research Award Competition. We aim to support up to five BC medical students' and undergraduates' in genomics related research projects, supervised by experts in the field. The deadline for applications is March 30th, 2012, with winners being announced mid-April.

BCCGN is also sponsoring 3 genomics research projects in the CFRI annual summer student research program and up to 4 Summer Student Research Projects with the Faculty of Medicine, UBC. Each year, our students have provided positive feedback about their experience. Many indicated that the experience sparked an interest in genomic research.

Member Awards

Dias, Rupps, Friedman, Arbour, Robinson, Zong, Steinraths, Birol, Brown and Gibson were awarded microgrants by the Rare Disease Foundation to use genomic technologies in various research projects.