BCCGN
British Columbia Clinical Genomics Network

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## IN THIS ISSUE:




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"I think this will become the medicine of the future, and I owe it to my patients to be better informed about the science: both its potential and limitations..." - *Physician, Victoria, BC*

# BCCGN Newsletter

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## BCCGN News and Updates

### BCCGN Annual Conference.

BCCGN's second annual conference, GenOMICS: Today and Tomorrow's Medicine was a huge success. A total of 217 delegates registered including family physicians, specialists, health professionals, researchers and students. Thirty-seven percent were physicians, 23% of whom travelled from outside the lower mainland as far away as Vancouver Island, Nelson, Golden, Prince George, and Dawson Creek.

Delegates enjoyed learning about sequencing, personalized medicine, metagenomics and the genomics of specific diseases. They also were impressed by the speed of change of clinical genomics and its future implications. Respondents found the direct to consumer debate to be particularly effective. Some respondents planned to change some aspect of their behavior as a result of the conference, including continuing to educate themselves about genomics. Respondents had numerous diverse suggestions for next years' conference; the most popular suggestions were sessions on common diseases (Infectious diseases, Cancer, HIV, Epilepsy, autism, prostate cancer, adult intellectual disability), case studies, current tests, technology and epigenetics. E-mail more suggestions
View Conference Proceedings

### Clinician Survey 2010

A clinician survey distributed to over 6000 physicians throughout BC. Greater than 80% of the respondents were either interested or open to the idea of participating in genomics research. The main barriers to conducting research continue to be a lack of time, opportunity and financial support. Overall knowledge of genomics by physicians has increased since last year and many conference attendees plan to seek collaborations that would permit them to participate in clinical genomics research and continue to educate themselves about genomics.

## Research in Focus

Medhi Najafzadeh, PhD Candidate – UBC Pharmaceutical Sciences, is working on a BCCGN funded study to identify barriers to the use of personalized medicine in clinical practice. Personalized medicine aims to find the right treatment for the right person based on their genetic information. First, Najafzadeh completed a qualitative assessment using 3 focus groups of BC physicians. He wanted to explore current beliefs and awareness of personalized medicine by physicians. He found that 68% favored using personalized medicine in their practice. The main barriers were 1) lack of training on genetic testing and genomics, 2) lack of appropriate guidelines, 3) ethical concerns regarding informed consent, 4) financial impact on the health care system, 5) time pressure (genetic testing interpretation would be time-consuming) and 6) mode of access (most physicians felt they should have a gate keeper role).

To determine the relative impact of each attribute on the physician's decision to use personalized medicine in their practice, a second study was conducted. The results revealed that the type of genetic test had the greatest impact on physician's decision to incorporate personalized medicine. This was followed by training opportunities, meaning that the physicians felt they required further education on genomics and interpretation of various genetic tests and the need for development of appropriate clinical guidelines for use. This research has resulted in some valuable information on the steps necessary for the translation of new genomic knowledge to clinical use by BC physicians.

# Technology in Focus

### New Genetic Test (AGH) for Clinical Practice

A karyotype is a routine microscopic test used to evaluate whole chromosomes for changes in number or structure. It is standard practice for patients with unexplained developmental delay, intellectual disability, autism spectrum disorders and multiple congenital anomalies. These disorders account for the largest proportion of cytogenetic testing due to their relatively high prevalence in the population.

A recent advance in cytogenetic testing, Array Genomic Hybridization (AGH), uses the entire genome to identify chromosomal imbalances 1/100$^{th}$ the size of those detected by karyotyping. AGH can establish diagnoses in about twice as many children as conventional testing. This improved resolution comes at a higher price, about $1,000 more than any one of a number of standard cytogenetic tests.

In BC, AGH is used clinically to detect submicroscopic duplications/deletions that are pathogenic in children with developmental delay/intellectual disability and/or birth defects. Approximately 15-20 AGH tests are performed each week at Children's & Women's Health Centre in Vancouver. The Provincial Medical Genetics Program helps identify appropriate patients for the testing, as well as providing the necessary pre- and post-test counselling and information to families and referring physicians. It takes about 2 months and additional validation tests are required in positive cases. Other labs in BC are now validating this technology, so AGH testing can be widely available in the near future providing more genetic diagnoses for children with multiple congenital abnormalities.

# BCCGN Activities

### Announcements:

►BCCGN has sponsored 7 summer students for genomic research projects. They are Rebecca Dayan, Valerie Taylor, Lu Li, Gareth Evans, Sarah Konefal, Cindy Zhang and Scott Brown.

### Events:

►Gene Screen BC Film Competition call for submissions is open until August 15$^{th}$. $7,000 in cash prizes will be awarded on Sept 8th!

### Member Awards:

► The Rare Disease Foundation awarded microgrants of $3500 to BCCGN members: Michelle Demos, Millan Patel, and Farah Zahir. ► Jan Friedman received the UBC Killam Teaching Prize for 2009/2010.

► Michael Hayden received the 2009 Jacob Biely Faculty Research Prize.

### Publications

►Co-occurrence of Joubert syndrome and Jeune asphyxiating thoracic dystrophy. A.M. Lehman et al., Am J Med Genet A. 2010 Jun; 152A(6): 1411-9.

►Human health at the ends of the earth. L. Arbour et al., Rural Remote Health. 2010 Jun; 10(2):1534.

►Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies. D.T. Miller et al. Am J Hum Genet 2010 May 14:86(5):749-64.

►Value for Money? Array Genomic Hybridization for Diagnostic Testing for Genetic Causes of Intellectual Disability: D.A. Regier et al., Am J Hum Genet 2010 May 14:86(5):765-72.

►Polyalanine expansion in the ZIC3 gene leading to X-linked heterotaxy with VACTERL association: a new polyalanine disorder? Wessels MW et al., J Med Genet. 2010 May; 47(5):351-5.

►The utility of quantitative methylation assays at imprinted genes for the diagnosis of fetal and placental disorders. Bourque DK et al., Clin Genet. 2010 Apr 8.