BCCGN
British Columbia Clinical Genomics Network

Co-Leaders
Prof Jan Friedman
Prof Michael Hayden

IN THIS ISSUE:

Contact Information
Shelin Adam M.Sc.
Network Coordinator
ashelin@interchange.ubc.ca

Lesley Phillips Ph.D.
Business Manager
lesleyp@interchange.ubc.ca
www.bccgn.ca

BCCGN Newsletter

June 2012

BCCGN Diary Dates:

- Wednesday, June 6, 2012: Neuropsychiatry Rounds, UBC Hospital, Speaker: Dr. Neal Boerkoel
- Thursday, June 7, 2012: Solving Clinical Problems with New Technologies – How Genomics can help, Vernon Jubilee Hospital, Speaker: Ruth Thomas, Genetic Counsellor, BCCGN
- Friday, June 8, 2012: Clinical Genomics, Penticton Regional Hospital, Speaker: Ruth Thomas, Genetic Counsellor, BCCGN
- Monday, June 11, 2012: Solving Clinical Problems with New Technologies – How Genomics can help, Ridge Meadow Hospital, Maple Ridge, Speaker: Ruth Thomas, Genetic Counsellor, BCCGN
- For more information go to www.bccgn.ca or contact Ruth Thomas (rthomas@cfri.ca) if you are interested in having someone from BCCGN speak with physicians in your area.

Breaking news

Improving Treatment Options for Childhood Cancer

The Canadian Government, as part of a larger commitment, has just announced $4.3 million dollar award to a BC Children's Hospital research team dedicated to improving childhood cancer treatment. With the increasing curability of childhood cancers, and an estimated 30,000 childhood cancer survivors in Canada, there remain a growing number who suffer lifelong and often debilitating side effects as a result of their cancer treatment. The research team, being funded at BC Children's Hospital and led by Dr. Kirk Schultz, will assess biomarkers involved with hearing loss, kidney failure, blood clotting problems, and a specific form of tissue rejection, in order to identify those children at risk of adverse effects of their successful cancer treatments. They will also attempt to predict the course of development of their long-term complications. Being able to identify these high risk children will lead to pre-emptive and timely therapies to reduce or eliminate these unwanted effects. **BCCGN member, Dr. Rod Rassekh** is one of the team's co-principal investigators along with Dr. Mary McBride.

Detection & Treatment of Ovarian Cancer

Dr. David Huntsman, a professor in the Dept. of Obstetrics and Gynaecology and the Dept. of Pathology and Laboratory Medicine, and a consulting pathologist at Vancouver General Hospital, will become the first person to hold the Dr. Chew Wei Memorial Professorship. This new professorship, created by the family of late Dr. Chew Wei, will be devoted to finding new ways of detecting and treating women with ovarian and other gynecological cancers. Dr. Huntsman's pathology background has enabled him to pursue methods of earlier diagnosis and enabled the identification of the major genetic mutations in two types of ovarian cancer.

Research in Focus

Detection & Treatment of Ovarian Cancer

Dr. David Huntsman, Prof. Dept. Obstetrics & Gynaecology and of Pathology and Laboratory Medicine, and a consulting pathologist at Vancouver General Hospital, will be the first to hold the Dr. Chew Wei Memorial Professorship. It was created by the family of late Dr. Chew Wei and is devoted to finding new ways of detecting and treating women with ovarian and other gynecological cancers. Dr. Huntsman's pathology background has enabled him to pursue methods of earlier diagnosis and enabled the identification of the major genetic mutations in two types of ovarian cancer.

Diagnosing Children with Bone Marrow Failure and Immune Deficiencies

Pediatric hematologists and oncologists routinely treat children with bone marrow failure and immune deficiencies, which can have detrimental effects on multiple organ systems. Despite extensive work-ups, many patients don't receive a diagnosis for the underlying cause of their disease. Management of these cases relies on treating symptoms, without being able to offer curative treatment. There is often an underlying genetic cause for the disease in these children, but due to the heterogeneous nature of the illnesses, identifying the specific mutation for each patient can be a lengthy process, necessitating testing multiple genes serially, when MSP funding can be obtained. When diagnosis is not made early in the disease course, the patient is subject to repeated and severe infections and organ damage, leading to significant morbidity and mortality

To shed light on these diagnostic dilemmas, Dr. Jacob Rozmus, a Pediatric Hematologist/Oncologist at BC Children's Hospital, has begun a project with BCCGN using the latest Next-Generation sequencing technology. The study will use exome sequencing to target the protein coding regions of the genome, which contains the vast majority of known disease causing mutations. If the underlying genetic mutation causing the disease is identified, it will illuminate which specific biological pathway is affected. With this better understanding of the cause of the disease, physicians will be able to offer curative treatments like bone marrow transplants. As bone marrow transplants are dangerous procedures, an accurate diagnosis is imperative to determine the safest, most effective conditioning regiment, donor selection and prophylaxis for graft versus host disease.

Through this proof of principle project, Dr. Rozmus would like to demonstrate that using exome sequencing for these patients can result in a timely diagnosis, prevent unnecessary morbidity and also the possibility of an effective and safe curative treatment.

BCCGN Activities

BCCGN has Joined (PMI)

In BC, a new Personalized Medicine Initiative (PMI) has sprung forth led by Drs Cullis, Huntsman (BCCGN member), Fraser, Hayden (BCCGN Co-Leader) and others to bring together expertise from diverse technological and healthcare communities to bring personalized healthcare to BC patients. Projects addressing the issues surrounding adverse drug reactions as well as other unmet healthcare needs will be followed through technology development, clinical testing and population delivery. The PMI will use advances in genomics, proteomics, metabolomics and informatics to improve clinical decision-making and therapeutic options across BC's health care system. It is building a framework inclusive of any disease and encompassing all facets of medical treatment, diagnosis and decision-making, including ethics, economics, and society.

Announcements:

► 5 BCCGN Summer Student Research Awards were received by James Cairns, Maria Kovalik, Jesica Ho, Cory Weissman, and Jieqing Xu.
► BCCGN funded 3 CFRI Summer Student Awards for Sean Addison, Edward Choi, and Charles Yang.
► BCCGN also funded 4 Faculty of Medicine Summer Student Research Project Awards for Angela Burleigh, Michael Horkoff, Mathew Michaeleski, and Minju Park.

Events:

► Entries for Gene Screen BC Film Competition are due on August 17th 2012 and the winners will be announced at a Gala Screening on September 25th
► BCCGN's fourth annual conference, *"Clinical Genomics - the Good, the Bad and the Ugly"*, was held on Friday 20th April 2012. The keynote address was from award winning author, Tania Katan who spoke about her experience with breast cancer. There were personal accounts of genomic testing over the internet, an explanation about direct-to consumer testing, genomic technology and its applications, such as pharmacogenomics and cancer. Footage is on www.bccgn.ca
► BCCGN hosted its fourth CME accredited workshop for physicians entitled "Introduction to Genomic Technologies" on May 24th. The next session will be in the fall.

Member Awards:

► Rare Disease Foundation Microgrants: Dr. Liam Brunham, Dr. Marion Coulter-Mackie, Dr. Patrice Eydoux, Dr. Isabel Filges, Dr. William Gibson, Dr. Gabriella Horvath, Dr. Anna Lehman, Dr. Shu Sanatani, Dr. Clara van Karnebeek received awards to use genomic technologies in their research.