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BCCGN Newsletter

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BCCGN News and Updates

Personal Genomic Tests: What's a Doc to do?

Traditionally, genetic testing was only available through healthcare providers. Today, all kinds of genetic tests are available and marketed directly to consumers via the internet, television, and print advertising. The types of testing available range from detecting breast cancer alleles, mutations linked to cystic fibrosis to more recent, highly controversial tests to assess traits such as a child's athletic ability.

Buyer beware, these, at-home genetic tests have significant risks and limitations. Many of the marketing strategies involve exaggerated and inaccurate messages about the connection between genetic information and disease risk and are often emotional in nature. The problem is that consumers can potentially misinterpret this genetic information and therefore be misguided about their own or their children's abilities or health.

Last year, the US FDA began demanding these companies provide validation of their methodologies as their current position is against personal genomic testing. As a result of their efforts, several companies (Counsyl, Ambry Genetics) now sell their tests to physicians rather than consumers while other companies (23andMe) continue to promote personal genomic testing directly to the consumer.

What factors should physicians consider? In a recent US survey, almost half of the healthcare providers surveyed knew about personal genomic tests and all of these had one or more patients ask them about getting a personal genomic test done with 15% bringing their results in for provider discussion. How many BC physicians are prepared to discuss testing results with their patients? When faced with such a situation there are 3 key factors that should be considered; 1) Analytic validity - which is the test's ability to accurately and reliably measure the thousands of genetic variants in these tests; 2) Clinical validity - which is the test's ability to detect or predict particular diseases or health conditions. This area of risk assessment is fraught with controversy right now. Most common diseases like diabetes, cancers and heart disease are caused by multiple genes, interactions with environment and human behavior. Much more research is needed to identify all these variants and how they influence disease susceptibility. 3) Clinical utility - if a test result reports an increased or decreased risk for a disease, what can be done about it? The same common interventions such as to stop smoking, weight reduction, increased physical activity, blood pressure control are beneficial for preventing and controlling many diseases.

What advice should be offered to patients? First, discuss with your patients the limitations of personal genomic tests. These tests are not ready for routine clinical use because of their limited clinical validity and utility. Second, encourage your patients to collect and keep an updated family health history. A family health history is a better way to determine relative risk for specific diseases. Lastly, use the discussion of personal genomic tests and family history as a teachable moment to encourage patients to improve their own health.

Genomic Medicine in 2015

Tamara Taggart, a well-known CTV anchor woman gave the keynote address at BCCGN's third annual conference. She spoke passionately about her journey through the health care system and her emotional experiences of having a child with Down's syndrome. Her funny stories revealed some deep rooted issues that need to be addressed by physicians and specialists. Tamara urged the audience not to lose track that family members may be in a fragile emotional state and so what you say and how you say it is important.

The conference this year focused on how genomic medicine might change clinical practice in 2015. For example next-generation sequencing of individual whole genomes is now very cost effective and therefore accessible. The challenges today lie in how to interpret the variants we find, are they pathogenic or benign? Challenges in the years to come will be in the costs associated with the bioinformatic interpretation and the ability to provide adequate counselling.

A further issue, debated at the conference, relates to incidental findings. While the development of new genomic technologies offers the promise of identifying mutations that underlie many diseases, this same technology is also likely to uncover many unexpected genetic changes that have important medical or social implications. It is not yet clear how such findings should be handled and who should be responsible for them.

Putting genetic information into context is also going to be a big challenge. Many physicians do not feel they are prepared for these ethical situations. Will genomic medicine take away from other services? Who is taking responsibility? In many cases now and increasing in the future, research and the clinic are no longer separate activities.

Physician Workshop

Our next "Introduction to Genomic Technologies" workshop for physicians will be held in Fall 2011. The full day CME accredited workshop uses case studies to show how genomics is applied in clinical situations and provides a tour through labs involved in microarray technologies and demonstrations of DNA preparation and analysis.

Gene Screen BC

Enthusiastic film makers and health professionals attended the 2nd annual Gene Screen BC movie competition launch event on May 24th, where inspirational speaker, Penelope Buitenhaus gave a practical, educational and humorous talk about all the key elements that go into the making of a good film. Then participants exchanged ideas and began to hatch plans for entering the competition. [Click here](#) to attend the screening gala or to make an entry.

BCCGN Activities

Education:

Articles on [Genetics and Genomics for Clinicians](#)

Member Awards

BCCGN Student Competition:

- ▶ Chelsea Fauth (supervisor: S Tebbutt, i-Capture); Genomic signatures that discriminate early from dual responses in allergic asthma and rhinitis subjects after allergen challenge.
- ▶ Katherine Santos (supervisor: I Wilson, BCCA) - Pathogenicity determination of unclassified DNA variants in familial cancer syndromes.
- ▶ Eric Zhao (sup.: C Dias, UBC/Medical Genetics); Identifying genomic causes of mental retardation.
- ▶ Peter Wang (sup.r: ME Lewis, UBC/Medical Genetics); Clinical and gene signatures of autism spectrum disorders.

CFRI Summer Studentship:

- ▶ Maia Kaplan (supervisor: S Turvey, UBC/CFRI) - The functional impact of NFKBIA gene promoter polymorphisms on the innate immune system.
- ▶ Emma Broderick (sup.: D Goldowitz, UBC/CMMT) - Nicotine driven DNA methylation modifications.
- ▶ Jeffrey Choi (sup.: C Wellington, UBC/CMMT) - Characterization of apolipoprotein E modulators.

Faculty of Medicine- Summer Student Research Program (SSRP):

- ▶ Jay Ching-Chieh Wang (sup. J Matsubara, UBC) Single nucleotide polymorphisms (SNP) in the complement factor H (CFH) gene in eye tissues.
- ▶ Namrata Jhamb (sup. WT Gibson, UBC); Genetic obesity disorders: visceral fat and biomarkers of cardiometabolic risk.

CFRI Mini-Med School High School Studentship:

- ▶ Zoe Johnson (sup: S.Adam/L.Phillips, BCCGN); BCCGN physician survey.

Member Publications:

- ▶ Epigenetic impacts on neurodevelopment: pathophysiological mechanisms and genetic modes of action. (Zahir FR) *Pediatr Res* 2011 May;69(5 pt 2):92R-100R
- ▶ Comparison of genome-wide array genomic hybridization platforms for the detection of copy number variants in idiopathic mental retardation. (S Langlois, JM Friedman, M Marra, P Eydoux) *BMC Med Genomics*. 2011 Mar 25;4:25.
- ▶ The sensitivity of massively parallel sequencing for detecting candidate infectious agents associated with human tissue. (JM Friedman, RA Holt RA. *PLoS One*. 2011;6(5):e19838. Epub 2011 May 13.
- ▶ Pharmacogenomics of serious adverse drug reactions in pediatric oncology. (CJ Ross, SR Rassekh, MR Hayden. *J Popul Ther Clin Pharmacol*. 2011;18:e134-51. Epub 2011 Mar 21.