CIHR **STAGE**

Strategic Training for Advanced Genetic Epidemiology

Campbell Family Mental Health Research Institute | camh Centre for Addiction and Mental Health

The **Centre for Addiction and Mental Health** is a proud partner of **CIHR STAGE** and jointly presents:

International Speaker SEMINAR SERIES

Dr. Julia Bailey

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Successful localization of genes that predispose to epilepsy using large pedigrees

Friday
February 2, 2018
12:00 – 1:00 pm

The Hospital for Sick Children
Daniels Hollywood Theatre
Room 1246, 1st Floor, Black Wing
555 University Avenue, Toronto, ON

Abstract:

The ability to localize genes that cause diseases has been influenced by methodologies and technologies. After the first RFLP markers were discovered we started doing linkage analyses on large, complex pedigrees that had multiple affected members across several generations. Then SNPs were discovered which were also useful for linkage. When large pedigrees were deemed too difficult to collect, many study designs moved towards affected sibling pairs, which required more families, but they were easier to locate and recruit. Technology has advanced where we now can generate genetic data on a lot more people, and case-control GWAS has become popular. Next generation sequencing has allowed for the detection of hundreds of thousands of variants in each individual that differs from the 'norm' and the new search is focused not on detecting the variation but on determining which of the variation is causal. Using individuals that are affected and related allows for the filtration of variants and makes the search for causality easier. Epilepsy is a complex disorder with both environmental and genetic risk factors. Juvenile Myoclonic Epilepsy (JME) is one of the most common genetic epilepsies. Our consortium has collected hundreds of JME pedigrees, with the number of affected per family varying from 1 to 12. These pedigrees are large enough to produce significant linkage signals, which decrease the search space for causal variants. Our methods have been successful in detecting several major genes including EFHC1, and most recently ICK.

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