



The **Sunnybrook Hurvitz Brain Sciences Program (Sunnybrook Health Sciences Centre)** is a proud partner of **CIHR STAGE** and jointly presents:



## Dr. Joseph H. Lee

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### Examination of human knock-in models to identify protective genetic factors for Alzheimer disease: Exploring PSEN1 founder mutation carriers and adults with Down syndrome

**Friday  
December 1, 2017  
12:15 – 1:15 pm**

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

#### Abstract:

Natural experiments take advantage of extreme conditions, created by biological, geographical or political events, and allow us to investigate how genetic and/or environmental factors influence phenotypes of interest. In so doing, such experiments can simplify the complexity inherent in human biology and may reveal genetic factors that have modest to weak effect size in the general population. For this talk on Alzheimer disease, I will discuss some of the findings from our two studies on high-risk cohorts: Puerto Rican families that have multiple PSEN1-G206A mutation carriers and adults with Down syndrome. These high-risk individuals who carry the PSEN1 mutation or those who have three copies of the APP gene should develop Alzheimer disease at a relatively young age. Yet, some resist developing Alzheimer disease despite their old age. By examining these individuals, we aim to identify genetic factors that may delay the age at onset of Alzheimer disease, thereby providing novel therapeutic targets.

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