



# PROGRESS IN NEUROSCIENCE PINS



Seminar Series of the  
Brain & Mind Research Institute  
Weill Cornell Medical College (WCMC)  
&

The Graduate Program in Neuroscience of  
WCMC and Sloan Kettering Institute

Thursday, 9/24/15, 4 PM, coffee at 3:45 PM  
**Uris Auditorium**

## “One Brain, Many Genomes: Somatic Mutation and Genomic Variability in the Human Brain”

Christopher A. Walsh, M.D./Ph.D.

Bullard Professor of Neurology and Pediatrics, Harvard Medical School  
HHMI Investigator and Chief, Genetics and Genomics, Boston Children’s Hospital



### Abstract:

Genetic mutations causing human disease are conventionally thought to be present in all cells, except for most cancer mutations, which arise during cell divisions in tissues of the body. Increasingly, somatic mutations are being identified in diseases other than cancer, including neurodevelopmental diseases. Somatic mutations can arise during the course of prenatal brain development and cause neurological disease even when present in a small fraction of cells, for example, resulting in brain malformations associated with epilepsy and intellectual disability. New, highly sensitive technologies allow amplification and sequencing of the genomes of single neurons from the human brain, and reveal the variety and extent of mutations distinguishing one neuron from another, in the form of copy number variation (CNV), retrotransposon insertion, repeat mutation, and point mutation, even in normal brain. We are only beginning to understand the extent to which mutations occur during normal brain development, and the extent to which they determine or modify normal and abnormal cognition.

### Recent relevant publications:

1. Poduri, A., Evrony, G.D., Cai, X. & Walsh, C.A. Somatic mutation, genomic variation, and neurological disease. *Science* 341, 1237758 (2013).
2. Jamuar, S.S. et al. Somatic mutations in cerebral cortical malformations. *New Engl J Med* 371, 733-43 (2014).
3. Lodato M, Wollworth M, et al. Somatic mutation in single human brain neurons tracks developmental and transcriptional history. *Science* (in press, 2015).



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