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, 5/1/14, 4 PM, coffee at 3:45 PM
Weill Auditorium

Genomic Segmental Duplications in Human Brain Evolution and Disease

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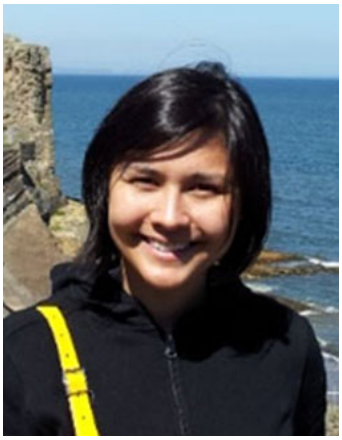

Abstract:

Though numerous studies have sought to identify the underlying mechanisms contributing to human-specific neurological traits and diseases, a large proportion of causal genes remain undiscovered. My research seeks to identify the role that human-specific "segmental duplications" (or genomic regions >1 kbp in size with a >98% sequence identity) play in neurodevelopment and disease. Not only do these regions contain genes with significant neurological functions, but they also predispose humans to large-scale genomic deletions and duplications associated with neurodevelopmental disorders such as autism, schizophrenia, and epilepsy. Due to their high sequence similarity, segmental duplications are often missing or misrepresented in the human reference build. Leveraging novel sequencing strategies, we show that these duplicated regions and genes likely play an integral role in human brain evolution and disease.

Recent relevant publications:

Dennis MY*, Nuttle X*, Sudmant PH, Antonacci F, Graves TA, Nefedov M, Rosenfeld JA, Sajjadian S, Malig M, Kotkiewicz H, Curry CJ, Shafer S, Shaffer LG, de Jong PJ, Wilson RK, and Eichler EE (2012). Evolution of human- specific neural *SRGAP2* genes by incomplete segmental duplication. Cell 149(4):912-22.

Girirajan S*, Dennis MY*, Baker C, Malig M, Coe BP, Campbell CD, Mark K, Vu TH, Alkan C, Cheng Z, Biesecker LG, Bernier R, and Eichler EE (2013). Refinement and discovery of new hotspots of copy number variation associated with autism spectrum disorder. AJHG 92(2):221-37.

Dennis MY*, Antonacci F*, Huddleston J, Sudmant PH, Meltz Steinberg K, Graves TA, Vives L, Malig M, Denman L, Baker C, Amemiya CT, Stuart A, Tang J, Muson B, Rosenfeld JA, Shaffer LG, Wilson RK, and Eichler EE. Palindromic *GOLGA* core duplicon promotes 15q13.3 microdeletion, inversion polymorphisms, and large-scale primate structural variation. American Society of Human Genetics Annual Meeting, Boston, MA (October 2013).

Weill Cornell Medical College