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, 3/1/18, 4 PM, coffee at 3:45 PM

Weill Auditorium (C-200)

"Paths to Understanding the Genetic Basis of Neural Tube Defects"

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My research program focuses on identification and characterization of genes predisposing to neural tube defects (NTDs). NTDs, including anencephaly and spina bifida, represent a group of common congenital malformations affecting 1-2 infants/1000 births. They are caused by failure of neural tube closure during embryogenesis. NTDs have a complex etiology involving both environmental and genetic factors. My group has adopted two main approaches to identify and characterize NTD genes: a candidate gene approach focused on molecular analyses of key genes of noncanonical Wnt/planar cell polarity pathway and a whole exome sequencing approach in NTD families.

Recent Relevant Publications:

1. Z. Kibar, et al. 2007. Mutations in *VANGL1* associated with neural tube defects. *New Engl J Med*. 356: 1432-1437. PMID: 17409324
2. Lemay et al. 2015. Investigation of *de novo* variants by whole exome sequencing identifies novel genes associated with neural tube defects. J.of Med. Genet. . 52(7):493-7. PMID: 25805808
3. P. Lemay P. 2017. Rare deleterious variants in *GRHL3* are associated with human spina bifida. Hum. Mutat. 38:716-724. PMID: 28276201

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