“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:

3. P. Lemay P. 2017. Rare deleterious variants in GRHL3 are associated with human spina bifida. Hum. Mutat. 38:716-724. PMID: 28276201