



BIOINFORMATICS SEMINAR

HO MING CHOW

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GENETIC STUDIES OF DEVELOPMENTAL STUTTERING: FROM LINKAGE ANALYSIS TO NEXT-GENERATION SEQUENCE

Developmental stuttering is a highly heritable neurodevelopmental disorder, characterized by involuntary disruptions in the flow of speech. It affects approximately 5% of children and 1% of adult populations, including the former vice-president of the U.S. Joe Biden. Using linkage and massively parallel DNA sequencing studies of consanguineous families from Pakistan and a large polygamous family from Cameroon with high prevalence of stuttering, researchers at the NIH have thus far identified rare variants associated with stuttering in four genes (GNPTAB, GNPTG, NAGPA, and AP4E1). Importantly, follow-up studies have confirmed that the prevalence of rare mutations in these genes are significantly higher in the North American population of unrelated people who stutter than people who do not have a history of stuttering. These four genes are involved in intracellular trafficking of lysosomal enzymes. In this talk, Dr. Carlos Frigerio Domingues will present the current evidence for the genetic causes of stuttering. In the second part of the talk, Dr. Ho Ming Chow will discuss the biological pathways involved the genes associated with stuttering and a factor that may help explaining why the mutations associated with stuttering appear to affect fluent speech production specifically.

BIOGRAPHIES

Dr. Carlos Frigerio Domingues obtained his bachelor's degree in Biomedical Science and postgraduate degrees (M.S. and Ph.D.) in Genetics from the University of Sao Paulo State University, Brazil, and received his postdoctoral training at the NIH. Dr. Frigerio Domingues will join the Department of Communication Sciences and Disorders, the University of Delaware as a Research Associate and continue to investigate the genetic contributions to developmental stuttering.

Dr. Ho Ming Chow is an Assistant Professor at the Department of Communication Sciences and Disorders, the University of Delaware. Dr. Chow obtained his Bachelor of Engineering and M.Phil. from the University of Hong Kong. He received his Ph.D. in Cognitive Science from the University of Osnabrueck, Germany and postdoctoral training in multimodal neuroimaging at the NIH. His research focuses on understanding the genetic factors and neurobiology underlying functional and structural anomalies associated with persistent stuttering and the neural re-organization leading to recovery from childhood stuttering.



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