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Identification and Characterization of Genes Involved in Common Developmental Brain Diseases

Status	Current
Competition	III
Sector	Health
Genome Centre	Genome Quebec
Project Leaders	Guy Rouleau & Pierre Drapeau

Project Description

Schizophrenia and autism are severe brain diseases that result in enormous human suffering and high healthcare costs. Despite decades of research, the causes of these diseases are still largely unknown. However, both diseases are believed to be associated with genetic (inherited) factors, and can therefore be investigated using genomics.

Dr. Guy A. Rouleau, an authority on the genetic basis of human brain tumours, human neurodegenerative diseases and psychiatric illnesses at the University of Montreal, and Dr. Pierre Drapeau, Director of McGill's Centre for Research in Neuroscience, are project leaders of Identification and Characteristics of Genes Involved in Common Developmental Brain Diseases.

Drs. Rouleau and Drapeau believe that genes causing schizophrenia and autism carry a high rate of mutations, and tend to be located at synapses, which are junctions between nerve cells (and other cells) in the brain where information is communicated and processed. In other words, mutations in specific synapse genes may cause these diseases.

Drawing on a collection of 5000 blood samples collected from individuals affected by schizophrenia and autism (and their family members), the research team will analyse 1000 synpatic genes in 276 patients. This analysis will make it possible to perform the first direct chemical examination of the genes in humans coding for particular synapses, and subsequently to validate the biological effects of disease-related mutations of these synapse genes in different animal model systems.

The project team expects to identify 10 to 20 genes that directly cause or increase susceptibility to schizophrenia or autism. This in turn will open the way to development of new diagnostic tests, new treatments and improved clinical management for patients – which will be of benefit to health policymakers as well as the genetics and neurosciences research communities around the world.