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Institute of Psychiatry and Neuroscience of Paris

UNIVERSITÉ PARIS DESCARTES

Psychiatry and Neuroscience Seminar

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(Host N Ramoz)

Montreal Neurological Institute
McGill University, Canada

Importance of rare diseases in psychiatry: examples of genetic discoveries in schizophrenia

Friday June 29th, 2018, 12 pm

Room R04-45, 102-108 rue de la santé - 75014 Paris

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Our studies are focused on the identification of genes or epigenetic modifications associated with specific forms of schizophrenia. Schizophrenia is a major psychiatric illness affecting one per cent of the population worldwide. Patients suffering from schizophrenia present delusions, hallucinations, abnormal behavior, impairment in daily life and cognitive deficits. Current treatments are insufficient in addressing all of these symptoms.

The heritability of schizophrenia is estimated at 80-90%, but the identification of genes associated with this illness has been challenging. To identify genetic variants of this disorder, we use current available technologies from genome-wide association studies GWAS to sequencing. Recent evolution to next generation sequencing “NGS” dedicated to exons or whole genome is helpful to identify rare variants, mutations and de novo mutations. Furthermore, our strategies are also to investigate extreme and severe forms. Thus, we can focus on childhood-onset schizophrenia, a rare disease with a prevalence of approximately 1 in 40,000 to identify novel genes of schizophrenia.

Missense variants in ATP1A3 and FXYD gene family are associated with childhood-onset schizophrenia. Chaumette B, et al. Mol Psychiatry. 2018