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# Wes of a Consanguineous Family with Schizophrenia and Mental Retardation in North Algeria.

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## Introduction:

Several studies have asserted the existence of a strong and complex genetic component in the determination of psychotic disorders. However, the genetic architecture of these disorders remains poorly understood. GWAS studies conducted over the past decade lead to the identification of only a few low effect associations, and the major part of this heritability remains unexplained , thus calling questioning the hypothesis of «common disease - common variants » for a model involving a large number of rare variants.

**Aims:** Here, we studied a multigenerational multiplex family in which co-exist psychotic and mental retardation disorders and a high rate of consanguinity, located in the northwest of Algeria. This study aims to identify inherited rare variants of schizophrenia and mental retardation.

**Methods:** This family have received complete clinical (DIGS, DSM-IV creteria) and genealogical investigations and whole exomique sequencing WES (by GAIIx Illumina / HiSeq 2000) that were performed in the departement of genetics in the university hospital of Geneva.

**Results:** 07 individus (02 parents and 05 siblings) has been analyzed. The parents were healthy. In siblings, 02 individus had schizophrenia diagnosis, 01 individu had mental retrdation, 01 had both schizophrenia and mental retardation diagnosis and 01 was healthy. Recessive model was retained in this family. For schizophrenia, WES have identified new rare mutations in known genes: rs17057255 (EPHX2), MAF=3%, rs35337118 (SPZ1), MAF=10%, rs2259633 (CCDC67), MAF=17%.

**Conclusion:** In total, these highly informative family have identified new genetic variants of schizophrenia. The search for these mutations in contol population in north west of algeria and a wider population of affected individuals with schizophrenia would be useful to validate these data.

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