

2p16.3 (NRXN1) Deletions

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CNVs spanning the 2p16.3 (NRXN1) and the 15q11.2 gene rich region have been associated with severe neuropsychiatric disorders including schizophrenia. Recently, studies have also revealed that CNVs in non-coding regions play an essential role in genomic variability in addition to disease susceptibility. In this study, we describe a family affected by a wide range of psychiatric disorders including early onset schizophrenia, schizophreniform disorder, and affective disorders. Microarray analysis identified two rare deletions immediately upstream of the NRXN1 gene affecting the non-coding mRNA AK127244 in addition to the pathogenic 15q11.2 deletion in distinct family members. The two deletions upstream of the NRXN1 gene were found to segregate with psychiatric disorders in the family and further similar deletions have been observed in patients diagnosed with autism spectrum disorder. Thus, we suggest that non-coding regions upstream of the NRXN1 gene affecting AK127244 might (as NRXN1) contain susceptibility regions for a wide spectrum of neuropsychiatric disorders.