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NEUROPSYCHOLOGICAL PHENOTYPE OF 16P11.2 MICRODELETION SYNDROME

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Introduction: Rapid progress in genetic techniques has revealed several novel microdeletion syndromes that, besides their relationship with intellectual disabilities, are frequently associated with an etiologically specific neuropsychiatric, neuropsychological, neurological or somatic phenotype.
Objectives: Investigating the neuropsychiatric phenotype of 16p11.2 microdeletion.
Aims: Extensive diagnostic evaluation of a 32-years-old male patient with autistic-like symptoms and a history of periodic regressive and catatonic behaviours.
Methods: Detailed genetic, neuropsychiatric and neuropsychologivcal examination.
Results: The patient failed to complete his university studies due to social and motivational mismatch (problems with inititative and structuring of daily life). Aged 25 a diagnosis of Asperger's disorder was made and the patient was admitted to a specialized department for diagnosis and treatment of autism spectrum disorders. EEG, MRI, and DAT-SPECT were all normal.The diagnosis was confirmed and he subsequently entered a sheltered home facility. Several years later he was referred for detailed re-evaluation of his persistent amotivational behaviour for which no explanation could be given.
At examination, no dysmorphic features were found. His behaviour showed autistic-like elements. No major psychiatric symptoms could be detected. Neuropsychological assessment revealed a KAIT Total IQ of 112 and undisturbed cognitive functions. As to personality and social interaction, an anxious disposition and feelings of distrust were noticed that established a marked vulnerability to psychotic experiences.
Microarray analysis showed a *de novo* interstitial deletion of 970kb in 16p11.2.
Conclusion: Here a never reported 16p11.2. microdeletion syndrome was demonstrated that appeared to be associated with a specific neuropsychological phenotype characterized by psychosis proneness.

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