Article: 546
Topic: 40 - Bipolar Disorders

PHELAN-MCDERMID SYNDROME AND ATYPICAL BIPOLAR DISORDER

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Introduction: The Phelan-McDermid syndrome (PMD) is characterized by intellectual disability (ID), childhood hypotonia, severely delayed or absent speech, and autistic features without major dysmorphisms and somatic anomalies. Haploinsufficiency of the *SH3 and multiple ankyrin repeat domains 3* (*SHANK3*) gene in 22q13.3 is thought to be causative for the PMD-phenotype comprising, in addition to sleep disturbances and deficits in language and communication, several behavioural abnormalities and increased reactivity to sensory stimuli. Data about neuropsychological dysfunctions or psychiatric symptoms are not available as yet.
Objectives: Investigating the possible psychopathological phenotype of PMD.
Aims: Diagnostic evaluation of one geriatric female and two male adult patients.
Methods: Detailed neuropsychiatric and neuropsychological examination of 3 patients with genetically proven PMD.
Results: In an adult male with a history of mood and behaviour fluctuations, psychiatric examination at the age of 24 disclosed symptoms of a severe major depression with irritability, loss of initiative, and marked sleep disturbances. Severe ID and marked deficits in expressive and receptive language were established. After addition of nortriptyline to valproic acid, depressive symptoms gradually remitted. His older brother has a similar history but with less severe symptomatology. The 70-years-old institutionalized female patient, had a history of affective and behavioural imbalance over many decades. Monotherapy with carbamazepine led to acceptable stabilization. Genetic evaluation in the two brothers and the female patient demonstated an identical loss of 2.15Mb and a 610kb deletion in 22q13.3, respectively, all including *SHANK3* gene.
Conclusions: Provisonal diagnosis of atypical bipolar disorder could be made in all three patients.

https://doi.org/10.1016/S0924-9338(13)75839-5 Published online by Cambridge University Press