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Phelan-mcdermid Syndrome in an Adult Female with Mild Intellectual Disability

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Introduction: With microarray analysis, several novel microdeletion syndromes have been ascertained that are often accompanied by a specific behavioural phenotype requiring specific treatment modalities.

Objectives: Investigating the neuropsychiatric phenotype of Phelan-McDermid syndrome.

Aims: Diagnostic evaluation of an adult female with mild intellectual disability.

Methods: Detailed genetic, neuropsychiatric and neuropsychological examination.

Results: The patient is a 22-years-old female. Aged 4, conventional karyotyping demonstrated a translocation 11;22. Her history is characterized by neonatal hypotonia accompanied by feeding problems, global intellectual disability, sleep disturbances, delayed development of speech, and ritualistic/ compulsive behaviours. Since about two years recurrent mood changes paralleled by an increase of pre-existent autistic behaviours became prominent. Psychotropics induced severe extrapyramidal symptoms without, however, any reduction of problematic behaviours.
At examination, no dysmorphic features were observed. Her behaviour showed autistic-like elements and mood alterations. Neuropsychological assessment revealed mild intellectual disability (SON-R IQ: 61) and a developmental age of 6;3 years with marked attentional deficits. Visuomotor, speech, and memory functioning were in line with her developmental age. Fluctuation in mood and affect was substantiated. MRI-brain disclosed no abnormalities. Microarray analysis confirmed the *de novo* t(11;22) that had resulted in a 11qter duplication of 8,77Mb and a 515kb 22qter deletion, encompassing the *SHANK3* gene, in agreement with a diagnosis of Phelan-McDermid syndrome.

Conclusion: Here a never reported unbalanced translocation leading to Phelan-McDermid syndrome with its characteristic neuropsychiatric phenotype is demonstrated. Apart from the implementation of systematic environmental measures, a plasma-concentration controlled treatment with valproic acid for mood stabilisation was advised.

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