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Neuropsychiatric signs and symptoms in treatable inborn errors of metabolism

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Possible underlying organic causes of psychiatric symptoms can be overlooked in the clinical setting. It is important to increase awareness amongst psychiatric and neurological professionals with regard to certain inborn errors of metabolism as, in some cases, disease-specific therapies are available that can, for instance, treat underlying metabolic causes. The following talk describes the basic pathophysiology, clinical and neurological features, and available diagnostic procedures of six treatable metabolic diseases that are associated with neuropsychiatric symptoms: Wilson's disease, cerebrotendinous xanthomatosis, porphyrias, homocysteinemia, urea cycle disorders, and Niemann-Pick disease type C (NP-C). NP-C is taken as a particularly relevant example because, while it is traditionally considered to be a condition that presents with severe neurological and systemic manifestations in children, an increasing number of patients are being detected who have the adolescent- or adult-onset form, which is frequently associated with neuropsychiatric condition usually based on patients' initial presentation with psychotic or schizophrenia-like symptoms. Underlying organic causes of psychiatric disorders such as psychosis should be considered among patients with atypical symptoms and/or resistance to standard therapy. Alongside improved frameworks for additional multidisciplinary diagnostic work in patients with suspected organic disease, the development of convenient and affordable biochemical screening and/or diagnostic methods has enabled new ways to narrow down differential diagnoses

Biography

Saba Nia received her Medical Degree in 2005 from the Medical University in Vienna, Austria. She began her Clinical career in the field of Psychiatry in Austria and Germany. After receiving her board certification in Psychiatry in 2011, she continued her training in the field of Neurology in Vienna, which she concluded in 2014. She is specialized in Rare Metabolic Diseases in the Neuropsychiatric field and has organized several congresses featuring neuropsychiatric symptoms in treatable inborn errors of metabolism. Her interests include movement disorders, organic psychosis and dementia in young adults.

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