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Adult Psychiatric Diagnosis of Niemann-pick Disease Type C (Case Study)

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Niemann-Pick disease is a rare, inborn metabolic illness with autosomal recessive inheritance. Its essential characteristic is the disorder of the lysosomal storage of cholesterol, which results in an intracellular accumulation of cholesterol and glycosphingolipoids (the latter are formulated from the former) with consequent cellular lesion. Niemann-Pick disease has three types (A-C), out of which patients suffering from the clinically heterogeneous type C can reach adulthood, and have neuropsychiatric symptoms besides hepatosplenomegaly, so the diagnosis can be identified in the adult psychiatric practice. Among rare diseases, it belongs to those few for which a disease-specific therapy exists.
A case with Niemann-Pick disorder Type C identified by us will be referred to. The patient concerned has another rare inborn disease; she has been suffering from Epidermolysis bullosa since infancy. There is no known genetic relationship between the two diseases. In this patient a juvenile form of the NP-C storage disorder has developed, and besides the innate visceral symptoms, neuropsychiatric alterations appeared in the form of learning disabilities. In the beginning, she was treated for attention disorder and specific learning difficulties, then from adolescence for the clinical syndrome of cyclothymia. The first neurological signs also manifested during puberty. The clinical diagnosis of NP-C was made in early adulthood after the recognition of atypical psychotic symptoms and dementia. Besides the symptomatic treatment, we have already been able to start the illness-specific therapy, and now we can show the results of the treatment's first year.

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