ABSTRACT

The following is a case of Niemann-Pick disease (NPD) type B associated with a psychiatric disorder. A 27-year-old male was admitted to the hospital with Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition—Text Revision criteria for paranoid schizophrenia. He was ameliorated after initiation of treatment by amisulpiride 400 mg/day. Auditory hallucinations, persecutory delusions, depersonalization, and derealization regressed and was able to work. He was followed up for >10 years. The patient had typical history of NPD type B (hepatosplenomegaly and pulmonary infiltrates) diagnosed at 3 years of age.

INTRODUCTION

Niemann-Pick disease (NPD) is an inborn error of lipids metabolism. In types A and B, there is a clear deficiency of sphingomyelinase, resulting in widespread lysosomal deposition of sphingomyelin liquid crystals. The most common disorder (type A) begins shortly after birth with hepatosplenomegaly, failure to thrive, neurologic impairment, and early death. The adult form (type B) is a relatively benign disorder with hepatosplenomegaly and pulmonary infiltrates, and is characterized by the sparing of brain involvement. Some authors report adult cases of NPD type B with neurologic and/or psychiatric symptoms. Patients with neurologic impairment (mental retardation, cerebellar ataxia, extrapyramidal signs, or cherry red-spots) have intermediate forms between type A and type B. Sometimes, brain storage is inapparent (found at autopsy). Patients with psychiatric disorder and with NPD, without any neurologic symptoms, are excessively rare—only two cases could be found in the literature. These two cases most likely belong to type B and there could be a chance association of schizophrenia and NPD.

NPD type C is a secondary cholesterol storage disorder without sphingomyelinase deficiency. Neurologic signs are often pointed out (juvenile form) and psychosis may be the only clinical manifestation of this disease (adult form). Hepatosplenomegaly is often mild.

FOCUS POINTS

• Niemann-Pick disease (NPD) could be associated with a psychiatric disorder.
• Psychiatric symptoms without neurologic impairment may be a manifestation of NPD.
• Psychiatric disorder found in NPD type B could be schizophrenia.
• The mechanisms underlying psychiatric disturbance are unknown.

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NPD type C is a secondary cholesterol storage disorder without sphingomyelinase deficiency. Neurologic signs are often pointed out (juvenile form) and psychosis may be the only clinical manifestation of this disease (adult form). Hepatosplenomegaly is often mild.
CASE REPORT

The following is a case report of NPD type B associated with a psychiatric disorder. A 27-year-old male was admitted to the hospital. He met Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition–Text Revision criteria for paranoid schizophrenia. He had auditory hallucinations and persecutory delusions. He also had feelings of strong depersonalization and derealization. Since 3 years of age, he had magical thinking and ideas of reference and had reduced his social activity. There were no neurologic or intellectual impairments. He had no history of developmental disorder. There was no drug abuse. The patient had a typical history of NPD type B (hepatosplenomegaly and pulmonary infiltrates) diagnosed at 3 years of age. The sphingomyelinase activity in leucocyte extracts was at 0.10 µKat/Kg proteins (normal at 0.67±0.25). Computed tomography scan and magnetic resonance imaging revealed a slight ventricular enlargement. An electroencephalograph indicated no abnormalities. There was no ocular abnormality. Motor nerves conduction velocities were normal.

He was ameliorated after initiation of treatment by amisulpiride 400 mg/day. Auditory hallucinations, persecutory delusions, depersonalization, and derealisation regressed and he was able to work. The patient has been followed up for >10 years. He worsened every time he discontinued amisulpiride or reduced the posology of 400 mg/day; the relapse was on the same mode, including delusions, hallucinations, and behavioural disturbance.

This is the third case of NPD type B associated with a psychiatric disorder, the first which presented NPD before schizophrenia. Hepatosplenomegaly had developed before the onset of psychiatric symptoms. Complete exploration of the central nervous system revealed no abnormality. The mechanisms underlying psychiatric disturbance are unknown. The authors of this case report could make no correlation between brain damage and psychiatric symptoms.

CONCLUSION

Psychiatric symptoms without neurologic impairment may be a manifestation of NPD (intermediate type AB) or a chance association (type B). In this case presented, the presence of enzyme protein in fibroblasts revealed an intermediate type AB.

REFERENCES