SHORT REPORT

Adult onset Niemann-Pick disease type C presenting with psychosis

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Niemann-Pick disease type C (NPC) is an autosomal recessive neurometabolic disorder that rarely presents in adulthood, and is associated with cognitive decline, various movement disorders (ataxia, chorea, dystonia, and myoclonus), a vertical supranuclear gaze palsy (VSGP), and seizures. A recent case report demonstrated a delay in diagnosis of eight years when a patient with NPC presented with psychosis. This article reviewed all cases seen at the Mayo Clinic with a possible diagnosis of NPC between 1976 and 2000. Of the 52 possible cases, five had an established diagnosis of adult onset NPC. Of these, two presented with psychosis and were not diagnosed with NPC for 5 and 15 years, respectively. NPC may initially present in adulthood with psychosis, and when psychosis is associated with VSGP, various dyskinesias, and seizures, NPC should be suspected.

RESULTS

Of these five adult onset NPC cases two presented with psychosis.

Case 1

A 61 year old woman was transferred to the Mayo Clinic with a 15 year history of an undiagnosed, progressive neuropsychiatric disorder, rendering her bed bound and unable to eat. The patient initially developed depression and hypersomnia at age 46. Her symptoms improved with an antidepressant. At 49, she was hospitalised for mood lability, loquacity, delusions, and hypervigilance. She was treated with thiothixine, imipramine, and lithium then discharged. At age 50, she developed a gait disorder with postural instability, was noted to “lean backwards” and have retropulsion. This was attributed to tardive parkinsonism. She was treated with levodopa and bromocriptine, and her gait improved. However, she again became hypersomnolent, as well as dysphagic, which required gastrostomy tube placement. One year later, auditory hallucinations began, and she became paranoid, hyperreligious, and obsessive, for example, making an excessive number of long distance telephone calls. Her differential diagnoses included: schizoaffective, bipolar, and organic affective disorders. She was then treated with thioridazine and amitryptiline. At age 55, she was noted to have saccadic pursuits, a hypokinetic and ataxic dysarthria, bradykinesia, and her first documented seizure. Between ages 55 and 61, she became demented, rigid, and virtually mute. She was transferred to the Mayo Clinic for evaluation at age 61.

On neurological examination, the patient was essentially mute. She had cranio-cervical dystonia, axial rigidity, and stimulus sensitive myoclonus. Eye movement examination was hampered by severe blepharospasm. MRI of the head showed subtle small foci of increased T2 signal changes within pons and cerebral white matter (see fig 1A). Bone marrow biopsy revealed a normocellular marrow with increased foamy macrophages. Skin fibroblast testing showed 5.5% cholesterol esterification and a positive filipin staining consistent with a diagnosis of NPC. Genetic testing was not performed. She died at age 62.

Case 2

A 32 year old woman presented to Mayo Clinic with a five year history of an undiagnosed neuropsychiatric disorder. Her birth and development had been unremarkable. At the age of 27, she developed a “nervous breakdown,” associated with paranoid delusions and was treated with haloperidol. Later that year, her psychosis resolved, but she developed facial dystonia, a shuffling gait and dysarthria, attributed to a tardive syndrome. Subsequently she experienced an annual recurrence of her psychiatric symptoms, particularly the paranoid delusions. At 32, she became dysarthric, ataxic and...
In summary, adult onset NPC may present in adulthood with psychosis. When a patient less than 60 years of age presents with the tetrad of psychosis, a VSGP movement disorders, and seizures, NPC is a possible aetiology.

REFERENCES
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