



A Case of Neuroacanthocytosis with Cerebellar Syndrome

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Abstract

Neuroacanthocytosis is a rare slowly progressive neurodegenerative disease accompanied with movement disorders, epilepsy, cognitive impairment, multisystem psychiatric symptoms. It is usually investigated in patients presenting with movement disorders, and diagnosis is made based on clinical presentation and the presence of acanthocytes in the peripheral blood. We aimed to present a case of neuroacanthocytosis with prominent cerebellar findings.

Keywords: Neuroacanthocytosis, acanthocyte, cerebellar syndrome

Introduction

Neuroacanthocytosis is a multisystemic, slowly progressive, neurodegenerative disease associated by chorea, dystonia, orofacial dyskinesia, tics, epilepsy, cognitive involvement, and behavioural changes (1, 2). In its differential diagnosis, there can be many diseases such as Huntington disease (HD), Parkinson's disease, Tourette's syndrome, and Wilson's disease. In this study, we aimed to present the case of a patient with neuroacanthocytosis with prominent cerebellar findings.

Case Report

A 52-year-old female patient applied with 5 years history of progressive gait disturbance and dysphagia, difficulty in speech and imbalance. She had a history of epilepsy for 20 years. She also had a history of smoking a pack of cigarettes a day for 30 years. She appeared cachectic, and her cooperation was limited due to cognitive deterioration. Her speech could hardly be understood in the neurologic examination; speech fluency was decreased with impaired articulation, dysphonic, and dysarthric. The tongue movements were limited. She had orolingual dyskinesia and dysphagia. Her muscle strength was 4/5 in the extremity proximal muscles and 3/5 in the distal muscles. The right interosseous muscles were atrophic. Deep tendon reflexes were decreased and the plantar reflex were bilaterally extensor. Mild rigidity was detected in the left upper extremity. She had bilateral dysmetria, dysdiadochokinesia, and ataxia. Hemogram values; function tests of the liver, kidney, and thyroid; folic acid levels; vasculitis parameters; serum copper, ceruloplasmin, and urinary copper levels; tumor markers; protein levels; and lipid electrophoresis were within reference intervals. B12 level was 120 mg/dL and replacement therapy was administered. On the cranial magnetic resonance (MR) examination, a 4-mm cavernoma and diffuse moderate cerebral atrophy were detected (Figure 1, 2). Chronic axonal sensorimotor polyneuropathy was detected in her electromyography examination. Electroencephalography findings were normal. The CAG repeat was normal. Acanthocytes rate over 50% was found in each peripheral smear repeated three times (Figure 3). The diagnosis of neuroacanthocytosis was established by clinical and peripheral smear findings. Symptomatic treatment with olanzapine (5 mg/day) was started. Patients' written informed consent was obtained.

Discussion

Neuroacanthocytosis, consisting of deformed erythrocytes known as acanthocytes in the peripheral blood is a rare disease which presents with various neurological and psychiatric findings. Its etiology is unknown. Acanthocytes develop secondary to ultrastructural abnormalities in the erythrocyte membrane (1, 2). The percentage of acanthocytes in the peripheral blood varies between 5% and 50% in neuroacanthocytosis patients. The percentage of acanthocytes is not related to the severity of the disease (3). Acanthocytes can aggregate in wet smears diluted with saline in a 1:1 ratio, which was first implemented by Feinberg et al. (4). Spiny and

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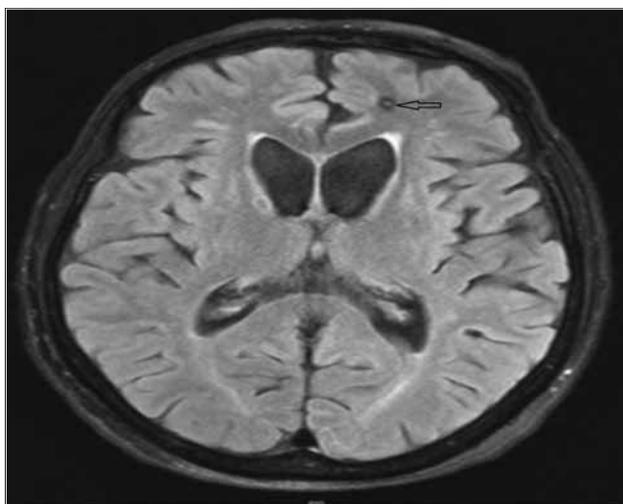


Figure 1. eft frontotemporal cavernoma in the cranial MR T2-weighted axial section
MR: magnetic resonance

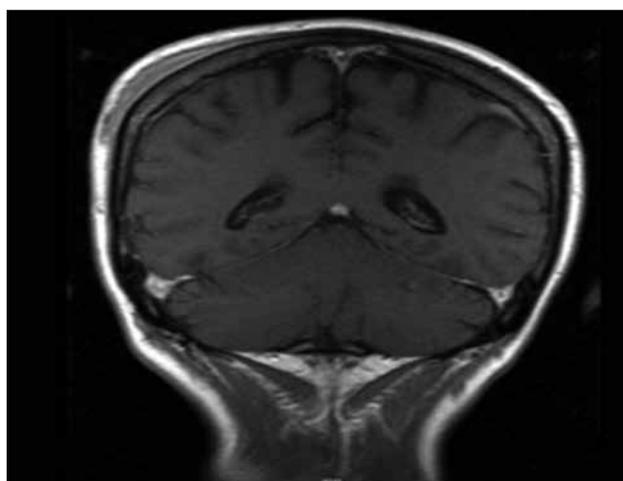


Figure 2. Figure 2. Moderate cerebral atrophy in the cranial MR T1-weighted coronal section
MR: magnetic resonance

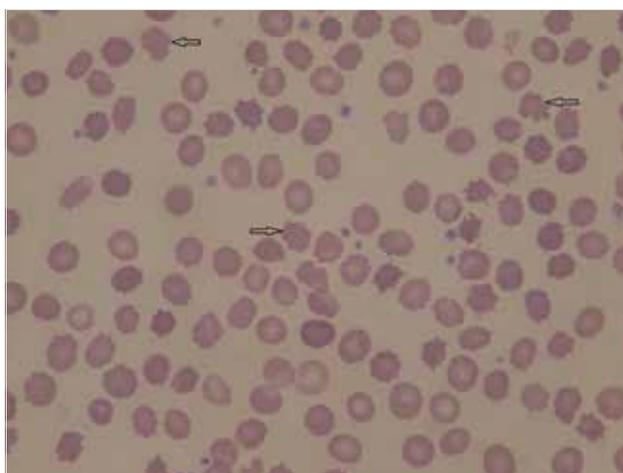


Figure 3. Acanthocytes observed in the peripheral smear

round protrusions are observed in erythrocytes during dilution with saline. This findings are not found in normal individuals and patients with HD (4, 5). In our patient, acanthocytes were observed at a ratio over 50% in all three smears. Despite cases

presenting in the first and seventh decades being reported, the onset age of the disease is in the third decade generally. The mean lifetime is shortened. It is generally autosomal recessive inherited, but autosomal dominant, X-linked recessive and sporadic forms have also been reported (6, 7).

The most common movement disorder in neuroacanthocytosis is chorea dominant to the lower extremities. Parkinsonism and dystonia can also be observed. Therefore, it can mimic HD mostly. Differently from HD, cerebellar findings, oropharyngeal dyskinesia that can cause dysphagia and dysarthria can be observed (8). In our patient, choreiform movement disorders were not present. Parkinsonism findings were of mild nature. Oropharyngeal dyskinesia that could cause severe dysphagia and dysarthria in addition to cerebellar findings were dominant. In 50% of the patients, generalized epileptic seizures usually appear as the initial finding (2, 9). Our patient had a 20 years history of epilepsy under control by antiepileptic therapy. Epileptic seizures were considered as the initial finding of our patient.

Neuroacanthocytosis is a member of the group of hereditary neuropathies progressing into MSS involvement. The presence of neuropathy, mostly axonal, has been reported in approximately 50% of the patients (2). Chronic axonal sensorimotor polyneuropathy was detected in our patient. In neuroacanthocytosis, psychiatric symptoms presenting with personality and behavior changes, anxiety, depression, obsessive-compulsive disorder, and emotional lability and cognitive deterioration are frequent (2). Since our patient was illiterate and had severe dysarthria, mental state evaluation tests could not be performed. Though cognitive deterioration was suspected. Radiological findings are not specific to the disease. On cranial MR examination, diffuse atrophy in the cerebral and caudate nucleus and ventricular expansion, which is more apparent in the lateral ventricular frontal horn are prominent (8, 10). Our patient's MR examination revealed mild cerebral atrophy, and a coincidental 4-mm cavernoma was found in the left frontotemporal region (Figure 1).

Conclusion

The diagnosis of neuroacanthocytosis is made through clinical manifestation and presence of acanthocytes in the peripheral blood. Even if this disorder is usually seen in patients presenting with movement disorders it also should be kept in mind while making the final diagnosis of patients with prominent cerebellar and multiple neurologic findings, as in our case.

Informed Consent: Written informed consent was obtained from patient who participated in this study.

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