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Case report
Olgu sunumu

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Neuroacanthocytosis in a case presenting to emergency department with acute respiratory failure and loss of consciousness: A case report

Acil servise akut solunum yetmezliği ve bilinç kaybı ile başvuran bir olguda nöroakantositoz: Bir olgu sunumu

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Abstract

The diagnosis of neuroacanthocytosis (NA) is made by the presence of acanthocytes via peripheral smear and by the accompanying clinical picture. Laboratory tests for diagnosis include blood smears to detect acanthocytosis, creatine kinase, genetic analysis and / or chorein level examination with western blot technique. This paper describes the case of a 34 year-old male patient who presented to the emergency department with acute respiratory failure and loss of consciousness. He was intubated and received mechanical ventilatory support after admission to the intensive care unit, and was diagnosed with Chorea akanthozytose (ChAc). We aim to emphasize the need to consider NA among the differential diagnoses for patients presenting with complex clinic such as acute respiratory failure and altered consciousness.

Keywords: Acute respiratory failure, Axonal neuropathy, Chorea akanthozytose, Intensive care unit, Neuroacanthocytosis

Öz

Nöroakantositoz (NA) tanısı, periferik yayma yoluyla akantositlerin varlığı ve beraberindeki klinik tablo ile yapılır. Teşhis için laboratuvar testler; akantosit tespit etmek için kan smear yapılması, kreatin kinaz, genetik analiz ve/veya western blot tekniği ile koreine düzey muayenesidir. Bu yazıda acil servise akut solunum yetmezliği ve bilinç kaybı şikayeti ile başvuran 34 yaşında bir erkek hasta sunulmuştur. Yoğun bakım ünitesine alındıktan sonra entübe edildi ve mekanik ventilatör desteği aldı ve Chorea akanthozytose (ChAc) tanısı aldı. Akut solunum yetmezliği ve bilinçte bozulma gibi karmaşık bir klinik ile başvuran hastalarda ayırıcı tanılar arasında NA düşünülmesinin gerekliliğini vurgulamayı hedefliyoruz.

Anahtar kelimeler: Akut solunum yetmezliği, Aksonal nöropati, Kore akantositozis, Yoğun bakım ünitesi, Nöroakantositozis

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Introduction

The term "acanthocyte" describes the spiculated appearance of red blood cells [1]. Acanthocytosis is a condition in which more than 3% of all red blood cells in the peripheral smear are defined as acanthocytes [2]. Neuroacanthocytosis (NA) refers to a genetically heterogeneous group of diseases characterized by neurological signs and symptoms that are accompanied by spiculated red blood cells in the peripheral smear. Neurological signs of these diseases include choreiform movements at the end of the extremities, dysarthria, dysphagia, muscle atrophy, hypoactive deep tendon reflexes, and epilepsy. During the advanced stages of NA, patients may experience dementia, marked distal muscle atrophy and weakness, movement disorders (e.g., dystonia, orofacial dyskinesia, tics and ataxia), cognitive impairment and personality changes, axonal neuropathy, epilepsy, and possibly Parkinsonism [3,4]. NA syndromes can be divided into 2 groups as autosomal recessive Chorea akanthozytose (ChAc) and McLeod Syndrome (MLS) with X chromosomal transition. The first group includes ChAc syndromes that manifest with neurodegeneration and involve a genetic defect in their etiology. In ChAc, there is a mutation in the VPS13A gene found in 9q21 and a defect in the protein called chorein.

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These syndromes present with basal ganglia involvement, movement disorders, orofacial dyskinesia and common choreiform movements in the extremities, cognitive impairment, and psychiatric symptoms. In MLS, the XK gene in the Xp21 is affected. This group manifests with lipoprotein metabolism disorders. Dorsal column degeneration and polyneuropathy are the prominent features in the clinical picture [5]. Each of the two major types of NA described above has its own etiology. Although they generally have autosomal recessive inheritance, autosomal dominant and X-linked recessive inheritance have also been described. Sporadic cases have also been reported [6,7]. Although ChAc syndromes usually start in the third to fourth decades of life, patients with MLS are older persons. These syndromes are progressive and degenerative diseases in which the age of onset, clinical and laboratory findings, family history, neurological and systemic involvement, and treatment vary according to the subtypes [8]. The time during which acanthocytes are observed in peripheral smear may vary from patient to patient. Generally, acanthocytes may be observed at the initial stages of the disease; however, in some patients, they may only show up in the late stages [9]. The most important criterion for the diagnosis of NA is the detection of acanthocytes in peripheral blood smear; however, the presence of acanthocytes is not meaningful on its own, and should certainly be supported with clinical findings. Despite the significance of acanthocytes in the peripheral smear, because they may be observed only in the late stages of the disease, the absence of acanthocytes in peripheral blood should not exclude the diagnosis, and the peripheral smear should be re-evaluated after diluting with saline. Normal controls show less than 6 % acanthocytic red blood cells of their total erythrocytes. Indeed, the observation of acanthocytes in the early stage aids in the diagnosis [1]. Genetic analysis and / or chorein level examination with western blotting can be used as further investigations. Currently, the treatment of NA syndromes is entirely symptomatic. Some drugs that are used in their treatment include anticholinergics, antipsychotics, antiepileptics, and acetylcholine release inhibitors; in addition, lithium treatment and pallidotomy are also used. Additional beneficial treatment approaches include botulinum toxin injection, dietary supplementation, and assistive walking devices [8]. Recent studies report that deep brain stimulation and deep brain bilateral pallidal stimulation can provide favorable results, especially at the end of the first month [10,11].

This paper presents the case of a 34 year-old male patient who presented to the emergency department with loss of consciousness and acute respiratory failure. After his initial evaluation, he was intubated due to inadequacy of spontaneous respiration, and he received mechanical ventilatory support after admission to the intensive care unit. He was diagnosed with ChAc, and his general condition improved after approximately 15 days of intensive care and treatment.

Case presentation

A 34-year-old male patient was brought to the emergency department due to acute respiratory failure and loss of muscle strength (especially prominent at the lower extremities), which was followed by loss of consciousness. The patient was sent to us for consult. He had stupor at his initial examination, and according to the Glasgow coma scale, he had eye opening in response to painful stimuli, his verbal response was incomprehensible sounds, and his motor response was not localized to painful stimuli. His pupils were isochoric, and he had both direct and indirect light reflexes. His spontaneous respiration was insufficient, and arterial blood gas analysis indicated respiratory acidosis and severe hypercapnia. He was hemodynamically normotensive and had normal heart rhythm. Due to the risk of respiratory arrest, the patient was immediately intubated and received mechanical ventilatory support with bilevel mode following admission to the intensive care unit. According to the statement given by his spouse, he had been followed up with in a military hospital for 2 months approximately 10 years ago due to a rapidly developing acute respiratory failure. At that time, he was given an initial diagnosis of autonomic neuropathy, and sural nerve biopsy revealed findings suggestive of Guillain-Barre syndrome. During that same period, the patient was being seen in a psychiatry clinic due to the diagnosis of bipolar affective disorder, for which he was taking olanzapine and lithium. Regarding the current episode, the patient had complained of difficulty in standing and walking due to weakness at his lower extremities, which began one week ago and choreiform movements at the end of the extremities. His loss of strength was then accompanied by respiratory difficulty, and he was brought to the emergency department after he developed a state of altered consciousness. His deep tendon reflexes were normoactive at the upper extremities, but deep tendon reflexes could not be obtained at the lower extremities. He had hammer toe deformity in both big toes, and pes cavus deformity of his feet. He also had widespread muscle atrophy, which was particularly prominent in the gluteal muscles and the quadriceps femoris muscles in the lower extremities. His laboratory tests revealed mild leukocytosis and a mild elevation of creatinine kinase. Vitamin B12 level was within normal limits, and Lipoprotein electrophoresis evaluated as normal. Due to his altered consciousness and acute respiratory failure, we first considered whether he had a cerebrovascular accident, drug intoxication, or a new attack of Guillain-Barré syndrome. Cranial magnetic resonance imaging (MRI) revealed normal results, while lumbar MRI did not show any compression to the spinal canal or the nerve roots. His clinical and laboratory findings were not suggestive of drug intoxication. A lumbar puncture was performed; biochemical analysis of the cerebrospinal fluid revealed normal results and did not yield any findings that could be associated with Guillain-Barré syndrome. Electromyography examination results were consistent with sensorimotor axonal polyneuropathy predominant in the lower extremities. His family history did not include any features. Although we primarily interpreted his initial results as a sequel sign to axonal type Guillain-Barré syndrome, which he had experienced in severe form in the past, we realized that the presence of hammer toe and pes cavus deformities could also indicate a hereditary form of neuropathy. The patient was monitored in the intensive care unit with mechanical ventilatory support, and symptomatic treatment was continued. Since the patient had a history of ataxia, orofacial dyskinesia, and parkinsonism symptoms accompanied by dysarthria and polyneuropathy, and because he had had cognitive

and psychiatric diagnoses in the past, NA was suspected in the differential diagnosis; therefore, a peripheral blood smear was performed. The peripheral smear revealed acanthocytes at a ratio greater than 20%, and the patient was diagnosed with NA. Since Genetic testing for VPS13A mutations was unavailable, protein analysis was performed Western blotting technique from tissue samples. The diagnosis which ChAc was confirmed by detection of chorein deficiency with the help of the free chorein Western blot technique. The polyneuropathy present in the patient was thought to be associated with ChAc (Figure 1).

In addition to his symptomatic treatment, the patient was given 1 g/day methylprednisolone pulse therapy for 10 days while he was being monitored with mechanical ventilatory support. During this time, his level of hypercapnia regressed, his consciousness returned to normal, and he was extubated as his spontaneous respiration became sufficient. The patient then underwent respiratory physiotherapy, muscle strengthening, and walking and balance exercises. At the end of the treatment period, his muscle strength increased, his overall condition returned to normal, and he was able to walk without any support. He had a normal state of consciousness and showed complete orientation and cooperation. His speech was dysarthric and his apprehension was normal. His eyes were at midline, and his eye movements were free in all directions. The patient's Rankin scale score increased to 2. The patient was referred to the neurology-psychiatry department in his present clinical condition.

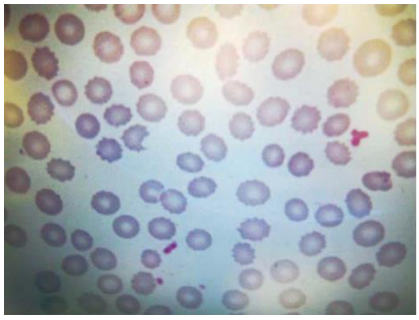

Figure 1: Acanthocytes observed in the peripheral blood smear of the patient

Discussion

NA Syndromes are composed of different diseases where nervous system abnormalities are seen together with red blood cell acanthocytosis [12]. NA is a neurodegenerative disease and the patients show some psychiatric signs, chorea, dementia as well as acanthocytes. In cranial MRI caudate nucleus atrophy may be present [13]. And patients mainly suffer because of the basal ganglia degeneration.

Presence of acanthocytosis on peripheral blood smear can vary, and they are not necessary to confirm the diagnosis. ChAc is an autosomal recessive, and MLS is an X-linked inherited NA syndrome. They share some similarities such as psychiatric symptoms, chorea, dystonia, myopathy, seizures, and peripheral neuropathy [5,14]. There are some additional sporadic conditions associated as well. For example, there has been a report of a NA case developing cerebellar atrophy [15].

Genes mutated in patients with MLS, ChAc, and Huntington's disease-like 2 have been identified. The mutations, the proteins and associated pathophysiological process have been identified [14,16], but the pathophysiology of the related red blood-cell anomalies is not yet clarified. There are studies focusing on the association between the integral membrane protein and the cytoskeleton [17]. Described herein is the case of a 34-year-old male patient. According to his history, symptoms of his disease were present at least 10 years prior to his current episode. This leads one to question whether the symptoms of NA begin when the patient is much younger, even though the diagnosis is typically made at later ages. Although there are reports of NA cases diagnosed in their first and seventh decades, the average age of onset for NA is 35 years [18]. A recent study from China including 66 cases reported that the age of onset of disease symptoms varied between 5 and 74 years [19].

NA disorders are extremely rare, and this rarity may be due to underdiagnosis. Thousands of ChAc and a few hundred MLS cases are thought to be present throughout the world. MLS are reported from Americas, Europe, and Japan and there seems to be no geographical selection [5]. ChAc seems to be common in Japan, and this may be related to some genetic predisposition [20], also some cases are present in geographically isolated communities of the French-Canadian population [21].

Psychiatric symptoms such as anxiety, depression, paranoid delusions, apathy, compulsive disorder (such as impulsivity), emotional lability, and cognitive disturbances are commonly observed in NA [18]. The case described herein was diagnosed with bipolar affective disorder, for which he was receiving treatment. NA is a rare, heterogeneous group of neurodegenerative diseases that can manifest diverse neurological signs. It has been reported that the possibility of developing dystonia and Parkinsonism are more likely than the possibility of developing chorea if the disease begins at an early age. Orofacial dyskinesia, dysarthria, and dysphagia can be observed in NA patients. These patients may also bite their own tongue and lip during eating due to orolingual dystonia (known as "eating dystonia") [13]. Our case had eating dystonia and tongue protrusion during the initial period after extubation; therefore, during that time, he was fed formula through a nasogastric tube. Oral intake was initiated later. Dementia is observed in approximately 50% of cases with NA. It is common for patients with the diagnosis of ChAc to have generalized type epileptic seizures [13]. Our patient had history of seizure. In a recent study including 66 cases [24]. accompanying findings are reported as hyperkinetic movements (88%), dyskinesia in orofacial region (80%), dystonia (67%), dysarthria (68%), caudate atrophy or enlarged lateral ventricles on neuroimaging (64%), and elevation in creatine kinase level (52%). Specific genetic tests or western blotting can use in confirmed the diagnosis as ChAc or MLS.

Studies show that myopathy and axonal type polyneuropathy are often found during electrophysiological examinations of patients with neuropathy. Histological examinations of these patients often reveal the involvement of generally large-diameter myelinated fibers and the presence of chronic axonal neuropathy [22]. In the electrophysiological examination of our current case, his sensory potential could not be obtained, and his motor potentials had significantly reduced amplitudes at the lower extremities. These findings are consistent with widespread, symmetrical, and sensorimotor axonal type polyneuropathy. NA syndromes are categorized as hereditary neuropathies that are associated with central nervous system

involvement, and nearly half of these patients have neuropathy [22]. Our current patient did not have deep tendon reflexes, and he had a loss of muscle strength at the lower extremities. NA patients may have elevated levels of creatinine kinase (CK) and may also suffer from amyotrophy. Elevated serum CK levels can also be observed without myopathy, especially in the ChAc and MLS subtypes, in which CK levels are usually between 300-3000 (U/L) [5]. The serum CK level of our current case was 920 (U/L). In NA, radiological imaging findings are not specific to the disease. For example, computed brain tomography, brain MRI, positron emission tomography (PET), and single-photon emission computed tomography can all be used as radiological examination methods. In patients with NA, brain MRI may reveal atrophy in the caudate and lentiform nuclei, as well as hyperintensity at the lateral putamina area in T2 sequence. Some cases may also show diffuse changes in white matter. In Figure 2 cranial MRI of several patients with NA are seen. In patients with NA, computed brain tomography can demonstrate caudate atrophy and ventricular enlargement, which is especially prominent in the anterior horns of the lateral ventricles. PET may show hypometabolic areas in the neostriatum and in the frontal cerebral cortical regions. Additionally, patients with NA often have reduced norepinephrine levels in the putamen and globus pallidus. Similar to findings in patients with Parkinson's disease, PET scans of patients with NA show a 42% reduction in DOPA re-uptake in the posterior putamen. Using single photon emission computed tomography (SPECT), the hypometabolic areas may be observed as areas of hypoperfusion [18,23]. Further, there have also been reports of cases with cerebellar atrophy [15]. Our current case only underwent brain MRI, the results of which were reported as normal. Autopsy examinations of NA cases demonstrate neuronal loss and gliosis in the caudate, putamen, globus pallidus, and substantia nigra, while the cerebral cortex is preserved. Apart from the basal ganglia, neuronal loss may also be profound in the thalamus and in the anterior horn of the spinal cord. As in Parkinson’s disease, in NA, neuronal loss in the substantia nigra is most profound in the ventrolateral region; however, nigral neuronal loss is more diffuse in neurodegenerative NA syndromes [24].

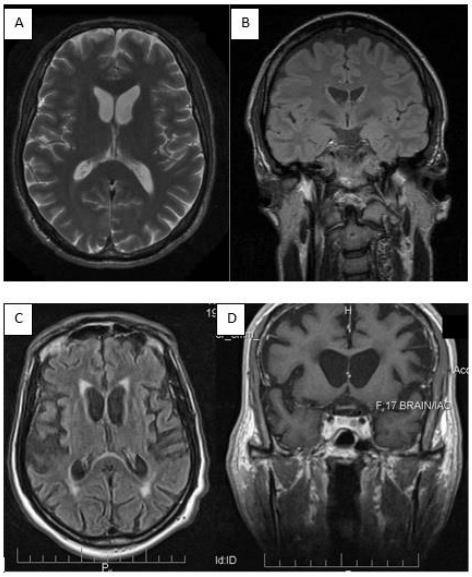

Figure 2: Cranial MRI images of Neuroacanthocytosis. Axial T2-weighted images demonstrate moderate atrophy of caudate nucleus and putamen (A).Coronal FLAIR-T1-weighted images demonstrate moderate atrophy of the caudate nucleus (B). Axial FLAIR- (C) and coronal T1-weighted images (D) demonstrate atrophy of the caudate nucleus and the fronto-temporal cortex. In addition, FLAIR images show periventricular white matter hyperintensities (courtesy of Nora Chan, MD, UCLA, Los Angeles, USA).

Diagnosis of NA is made using a combination of clinical properties, appropriate family history, and detection of acanthocytes in the peripheral blood smear and protein analysis the help of western blotting technique and/ or with genetic testes [24]. For neurodegenerative NA syndromes, demonstration of acanthocytes in the peripheral blood smear is the most important diagnostic criterion; nevertheless, clinical findings should not be underrated [25]. The percentage of acanthocytes in the blood smear of patients with NA varies between 5-50%; however, the ratio of acanthocytes does not reflect disease intensity. In our current case, we performed a peripheral blood smear upon the suspicion of NA due to the presence of suggestive clinical signs and symptoms, which revealed the presence of acanthocytes at a ratio of 20%, confirming the diagnosis of NA. Since acanthocytes may be present at various stages of the disease, it is not always possible to detect them, even though there may be clinical findings pointing towards the diagnosis of NA. In such conditions, it is recommended that the peripheral smear be repeated or re-evaluated after diluting with saline in a 1:1 ratio. Repeating the peripheral smear with a thinner layer of blood may increase the possibility of diagnosis [26]. The diagnosis was confirmed with western blotting from tissue samples as chorea ChAc.

The symptoms of NA syndromes may vary among patients, which causes misdiagnoses. In some, the disease starts with dominant psychiatric features like schizophrenia, obsessive-compulsive disorder, depression, cognitive impairment, tics, and Tourette's syndrome. In others neurological and muscular symptoms are dominant such as parkinsonism, chorea, epileptic seizures, dystonia, peripheral neuropathy, myopathy, or cardiomyopathy.

Although there have been several reports of NA cases in the literature in which neurological and psychiatric symptoms predominate the clinical condition, to our knowledge, ours is the first reported case of NA presenting to the emergency department with such severe clinical conditions as acute respiratory failure and altered conscious state. Therefore, we believe our case is significant for the literature. Moreover, it is also interesting that our case demonstrated almost all the clinical manifestations described within the context of ChAc. Our patient's presentation with acute respiratory failure suggested that his respiratory muscles were affected by his neuropathy. Our patient most likely received benefit from steroid treatment because steroids have known effectiveness in the treatment of neuropathy. It is clear that a successful emergent approach, early diagnosis, and appropriate treatment of the disease in a case presenting with such a severe condition are of vital significance. Failure to meet these requirements may lead to unwanted outcomes such as a requirement for long-term mechanical ventilation and hypoxic encephalopathy. Further, it is even possible that the clinical condition may become as dramatic as to cause the death of the patient.

Conclusion

Various presentations are known for these syndromes and, it should be noted that NA may manifest with a very severe unexpected presentation, as in our case. Therefore, we believe it is necessary to consider NA among the differential diagnoses in patients presenting with a complex clinical picture, including

acute respiratory failure, and altered consciousness. The diagnosis of NA syndromes can be made quite rapidly with the peripheral blood smear, which is an amazingly simple and cost-effective method; a rapid diagnosis leads to a prompt start to treatment. Western blotting for protein analysis and/ or genetic tests for mutations can be performed to confirm the diagnosis and to identify subgroups of NA syndromes.

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