Olgu sunumu / Case report

Neuroacanthocytosis in a psychiatry clinic: a case report

Rahime Hülya BİNGÖL ÇAĞLAYAN,[1] Gökçen GÖZÜBATIK ÇELİK,[2] Neşe KOCABAŞOĞLU,[3] Güneş KIZILTAN[2]

ABSTRACT

Neuroacanthocytosis (NA) syndromes are a group of genetically defined diseases characterized by peripheral blood acanthocytes, central nervous system as well as neuromuscular symptoms. The clinical presentation of neuroacanthocytosis typically includes chorea and dystonia. Psychiatric and cognitive symptoms may be significant including obsessive-compulsive disorder, depression and schizophrenia-like psychosis. Here, we aim to discuss a case presented with psychiatric symptoms and orofacial dyskinesia with the diagnosis of neuroacanthocytosis. A 25 year-old man was admitted to our outpatient clinic with depression, tongue biting and a speech disorder alleviated. His symptoms began after the death of his mother one year ago. Psychiatric examination showed that he had depression childish speaking as well as behavioral abnormalities. His history revealed dependent personality disorder. Neurological examination showed chorea, tics in his face and arms and hypotonia in all extremities. His cranial magnetic resonance imaging was normal, muscle enzymes were elevated, peripheral blood smear showed increased number of acanthocytes. The differential diagnosis was made between chorea-acanthocytosis, McLeod's syndrome and Huntington's disease. The gene analysis for Huntington's disease was negative, his lipid profile was normal. Symptomatic treatment was commenced. ***(Anatolian Journal of Psychiatry 2015; 16(4):304-306)***

Key words: *neuroacanthocytosis, psychiatric symptoms, orofacial dyskinesia*

Psikiyatri kliniğinde nöroakantositoz: Olgu sunumu

ÖZET

Nöroakantositoz sendromlar periferik kan akantositlerin, merkezi sinir sisteminin yanı sıra nöromüsküler belirtiler ile karakterize, genetik olarak tanımlanan bir hastalık grubudur. Nöroakantositozun klinik görünümü tipik kore ve distoni içerir. Psikiyatrik ve bilişsel belirtiler obsesif kompulsif bozukluk, depresyon ve şizofreni benzeri psikoz da dahil olmak üzere önemli olabilir. Burada, psikiyatrik belirtiler ve nöroakantositoz tanısı ile orofasiyal diskinezi ile başvuran bir olgu tartışılmıştır. Yirmi beş yaşındaki bir erkek, depresyon, dilini ısırma ve hafifletilebilir bir konuşma bozukluğu yakınması ile polikliniğimize başvurdu. Belirtileri bir yıl önce annesinin ölümünden sonra başlamış. Psikiyatrik muayene, depresyonu ve çocuksu konuşmasının yanı sıra davranışsal anormallikler olduğunu gösterdi. Onun geçmişi bağımlı kişilik bozukluğu saptandı. Nörolojik muayenede tüm ekstremitelerde yüzünde ve kollarında ve hipotoni de kore, tikler gösterdi. Onun kranial manyetik rezonans görüntüleme normal, kas enzimleri yükselmiş idi, periferik kan yayması akantositlerin artan sayıda gösterdi. Ayırıcı tanı kore-akantositoz, McLeod'ın sendromu ve Huntington hastalığı arasında yapıldı. Huntington hastalığı için gen analiz onun lipid profili normal olduğunu, negatif oldu. Semptomatik tedavi başlandı. ***(Anadolu Psikiyatri Derg 2015; 16(4):304-306)***

Anahtar sözcükler: *Nöroakantositoz, psikiyatrik belirtiler, orofasiyal diskinezi*

INTRODUCTION

Psychiatric symptoms may be early symptoms of some organic disease if they contain neurological examination findings like chorea. Chorea can be also caused by different etiology like degenerative, sporadic, metabolic, structural, autoimmune, infection, neoplastic, toxic and psychiatric disease. However, neuroacanthocytosis (NA) are a group of genetically defined diseases characterized by peripheral blood acanthocytes, central nervous system as well as neuromuscular symptoms. The clinical presentation of NA typically includes chorea and dystonia. Psychiat-

[1] Department of Child and Adolescent Psychiatry; [2] Department of Neurology; [3] Department of Psychiatry, Cerrahpasa Faculty of Medicine İstanbul University, İstanbul, Turkey
Yazışma adresi / ***Correspondence address:***
Uzm.Dr. R.Hülya BİNGÖL ÇAĞLAYAN, İstanbul Üniversitesi Cerrahpaşa Tıp Fakültesi Çocuk ve Ergen Psikiyatrisi ABD, Kocamustafapaşa/İstanbul, Türkiye
E-mail: rhulyabingol@gmail.com
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ric and cognitive symptoms may be significant including obsessive-compulsive disorder, depression and schizophrenia-like psychosis.[1,2] Here, we aim to discuss a case presented with psychiatric symptoms and orofacial dyskinesia with the diagnosis of NA.

CASE

A 25 year-old man was admitted to our outpatient clinic with depression, tongue biting and a speech disorder alleviated by putting a napkin in his mouth. These complaints had begun after his mother died one year ago. He was born to uncertain related parents. His history revealed that he has been dependent personality disorder since childhood. His family history showed that his two brothers, one sister and newborns of his relatives died at young age with unknown etiology and his father's family has infertile and mental retardation patients. However; he had taken different antidepressant and antipsychotic drugs given by other clinic in different time intervals due to the diagnosed with depression, his complaints progressed after his father's second marriage. He had taken still carbamazepine 400 mg, quetiapine 400 mg, biperiden 2 mg, and sertraline 100 mg per a day for one year. Psychiatric examination showed that he had depression, childish speaking as well as behavioral abnormalities and impaired the ability of abstraction. Although, neurological examination showed hypotonia in all extremities, his strength and gait were normal. Deep tendon reflexes were diminished. Choreic movements and tics affected his face, arms and in all extremities. He had also intermittent swallowing during speaking intercalary. However, laboratory investigations revealed increased number of acanthocytes in peripheral blood smear, elevated CK (1193 U/L) and CKMB (45 U/L) levels, his cranial magnetic resonance imaging was normal (Figure 1). Urea, creatinine, sedimentation, iron parameters, lipid profile and fundus examination were normal. The copper level was also normal in blood and 24-hour urine. The gene analysis for Huntington's disease was negative. His CATTEL test was normal and mini-mental score was 24/30 (calculation and countdown defect). His neuropsychiatric test was shown that his ability of attention, categorized change, abstraction, judgment, skills were in the mid-level disruption.

DISCUSSION

NA is a rare heterogeneous neurodegenerative disorders which is known as peripheral blood acanthocytes deformed by the presence of erythrocytes with unknown etiology. NA was recognized first in 1950 by Bassen and Kornzweig.[3,4] The other form of neurodegenerative disease with acanthocytes was identified as normal lipoprotein levels by Levine and Critchley discretely.[5,6] The clinical features of NA include orofacial dyskinesia, feeding dystonia, cognitive impairment and psychiatric symptoms such as personality changes, attention deficit-hyperactivity disorder and obsessive-compulsive disorder, epilepsy, peripheral neuropathy, cardiomyopathy.

Psychiatric symptoms as behavioral abnormallities, cognitive impairment, movement disorders, acanthocytosis were co-existing in Wilson disease, Huntington disease, chorea-acanthocytosis, McLeod's syndrome, A-beta-lipoproteinemia.[7-12]

The combination with acantosistosis and neurological disorders may occur more rarely in Hallervarden-Spatz, HARP (hipo-prebeta-lipoproteinemia, acantosistosis, retinitis pigmentosa, pallidal degeneration) also.[13]

Wilson's disease (WD) is an autosomal recessive disorder, which is related to copper metabolism disturbance, caused by mutations in ATP7B. Although patients diagnosed with WD have chorea about 15% frequencies, it's presented with dysarthria, dystonia, ataxia or tremor commonly. WD-patients have also Kayser-Fleischer rings in the cornea, brain MRI abnormalities and low serum ceruloplasmin, high urinary copper levels.[14] However clinical manifestation of our patient preoccupied WD, the normal levels of laboratory testing as copper and ceruloplasmine levels especially, normal fundus examination suspended us from diagnosis of WD.

Hallervarden-Spatz is an autosomal dominant disorder diagnosed with mutations in PANK2 (encoding pantothenate-kinase 2). Due to the iron metabolism deficit, abnormal iron accumulation causing chorea, cognitive impairment has been detected in the basal ganglia in MR imaging.[15,16] A-betalipoproteinemia and HARP diseases are determined chorea and psychiatric symptoms. In addition, these patients have also lipid profile deficit and fundus abnormality as retinitis pigmentosa.[1,3,4] Because of normal lipid. profile, normal MRI and normal fundus examination of our patient, these differential diagnoses were eliminated.

Hungtingon's Disaese (HD) is characterized as cognitive impairment, psychiatric symptoms, and

progressive chorea with an autosomal dominant family history. Diagnostic genetic testing is Huntington gene mutation an indication of the 9q21 gene defects on chromosome 4.[17-19] Although the diagnosis for our patients had supported HD his genetic testing was negative and his MRI was normal.

Mc-Load and choreoacanthocytosis are a subgroup of NA spectrum disorders, which presented with chorea, acantocytosis, psychiatric symptoms and normal lipid profile. However their clinical findings are partially similar, their etiology and age at disease onset are different. Mc-Load syndrome is an X-linked recessive disorder, the gene encoding the Kell erythrocyte antigen. It was presented with limb chorea and facial tics at later ages than other NA spectrum disorders commonly. In additionally, subcortical dementia, cardiomyopathy, hemolytic anemia are other clinical features.[20]

Choreoacanthocytosis is an autosomal recessive disorder associated with mutations of chorein protein, is characterized by chorea, orofacial dyskinesia, tics, parkinsonism, cognitive impairment, hypotonia dominant in the lower extremities, seizures, personality disorders. The mean age of disease onset is 2 or 3rd decades. Self motilation, tongue and lip biting appear most common than Mc-Load syndrome and HD.[13,21]

If we need to discuss on the basis of our case, age of disease onset clinical features and laboratory findings demonstrated the NA spectrum syndrome as Mc-Load or choreoacanthocytosis subgroup. Our patient has not electromyography and blood testing for Kell antigen mutation due to the unwillingness; so we could not established subgroup diagnosis.

CONCLUSION

The clinical features of NA include epilepsy, peripheral neuropathy, cardiomyopathy, orofacial dyskinesia, feeding dystonia, cognitive impairment and psychiatric symptoms such as personality changes, attention deficit-hyperactivity disorder and obsessive-compulsive disorder. Findings such as acanthocytosis and elevated CK levels, although nonspecific, are essential clues to the diagnosis. Management of NA syndromes is symptomatic.[2]

Different kinds of organic disease may be presented with only psychiatric symptoms as our patient. It should be noted that there are many diseases that lead acanthocytosis in blood and for these reason, correlation of clinical psychiatry and neurology is important for diagnosis and treatment.

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