

Neuroacanthocytosis presenting with late onset attention deficit hyperactivity disorder: A case report

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SUMMARY

Neuroacanthocytosis (NA) disorders are described by different neurological irregularities joined by strange red platelets called acanthocytes. Neuroacanthocytosis disorders might be isolated into 4 primary subgroups. These subgroups are named as; Center NA conditions, Degenerative problems where acanthocytosis is sporadically seen, Paroxysmal dyskinetic endlessly messes with diminished blood lipoproteins and acanthocytosis. This report centers around a patient with Chorea-acanthocytosis who gave engine signs in the third ten years of life. In the later course of illness, the patient created consideration shortfall hyperactivity jumble and different spasm problems. We needed to underline the significance of multidisciplinary care in the administration of chorea-acanthocytosis.

INTRODUCTION

Neuroacanthocytosis (NA) conditions are portrayed by different neurological irregularities joined by unusual red platelets called acanthocytes [1]. These problems were initially named as "Levine-Critchley condition" since they were initially remembered to have homogenous etiology. After the distinguishing proof of the capable qualities, it is perceived that neuroacanthocytosis conditions might be separated into 4 primary subgroups. These subgroups are named as; Center NA conditions, Degenerative problems where acanthocytosis is sometimes seen, Paroxysmal dyskinetic endlessly messes with diminished blood lipoproteins and acanthocytosis. Our case report centers around Chorea-acanthocytosis, which is one of the two center NA conditions alongside McLeod disorder.

Chorea-acanthocytosis is an autosomal passive problem. An exceptionally uncommon problem is assessed to influence 1,000 individuals overall. It is equitably appropriated among guys and females and seems, by all accounts, to be more common in Japan and French-Canadian populace. Beginning side effects typically present in 20s and advances gradually north of 15-30 years.

Starting show of Chorea-acanthocytosis might misdirect. Frequently patients foster mental or mental side effects before the neurologic indications. Indications might differ broadly. Most unusual and trademark element of this condition is orolingual dystonia that includes the lower face and tongue. This show may likewise happen in different problems however the seriousness is exceptionally huge in chorea-athetosis. Taking care of dystonia is likewise important for this disorder and it happens with eating. When the tongue contacts the food bolus, it pushes the food out of the mouth. Furthermore, self-ruining lip or tongue gnawing might be seen, which looks like Lesch-Nyhan condition. One more related problem that might inflict any kind of damage is an amount of side effects called as elastic man appearance, depicted as unexpected head and trunk flexion and expansions that might cause brutal head banging or hazardous falls. Parkinsonism may likewise create and some of the time be the underlying show. Going with bradykinesia typically grows later in the process. As stressed later in this paper, mental part of the sickness may likewise be extremely prevailing and early introducing. These incorporate spasms that look like Tourette disorder particularly assuming they were available before the neurological side effects. Likewise, even 10 years sooner than the neurological signs, patients might give detachment, misguided thinking, fanatical urgent problem and schizophrenia like psychosis [2].

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Despite the name of the syndrome suggests, acanthocytes are not always seen in peripheral blood smears of these patients. Acanthocytes levels tend to change during the course of the disease and these levels don't correlate with the severity of disease. Serum Creatinine Kinase (CK) levels are a much more useful parameter regarding its moderate or evident elevation is a frequent finding among patients. Also, elevated liver enzymes are seen in nearly 50% of the patients. As a confirmatory test, Western blot assay of the Chorein protein, product of VPS13A gene, may be performed. Additionally, electroneurography may demonstrate sensorimotor axonal neuropathy. On magnetic resonance imaging (MRI), specific involvement of the head of caudate nucleus is seen. Putamen and Globus Pallidus are also predominantly affected.

In this case report, we present a rare case of chorea-acanthocytosis with attention deficit hyperactivity disorder (ADHD).

CASE PRESENTATION

A thirty-three-year-old female presented to neurology outpatient clinic with choreiform movements in both her upper and lower extremities and difficulty with fine motor movements. She reported that these complaints have been going on for 12 years. In the last 4 years, she has also developed forgetfulness, attention deficit, orofacial dyskinesia and tic disorder that affects her eyes.

Physical examination was insignificant except for decreased deep tendon reflexes in her four extremities. Echocardiography of the patient and cranial imaging with MRI yielded no abnormalities. Upon clinical suspicion, hematology referral was made, and peripheral blood smear was obtained [3]. Smear revealed 15% acanthocyte formation.

To further investigate, EMG was performed and showed sensory axonal peripheral neuropathy which is expected in chorea-acanthocytosis. In addition to these, creatine kinase levels were highly elevated.

A psychiatry referral was made to evaluate the relatively newer symptoms of the patient. Psychiatry department performed DIVA scoring system and diagnosed the patient with ADHD [15]. The patient was enrolled with routine psychiatric follow up for adult onset ADHD.

Upon diagnosing the patient with chorea-acanthocytosis, treatment with clonazepam was initiated as recommended in the literature.

RESULTS AND DISCUSSION

Neuroacanthocytosis is a general term that is utilized for a gathering of illnesses that present with strange erythrocyte morphology and multi-framework neurologic signs. This gathering of illnesses incorporate pantothenate kinase related neurodegeneration (PKAN), Huntington's sickness like 2 (HDL-2), McLeod condition (MLS) and Chorea-acanthocytosis. The last two are additionally called as center disorders of the gathering [4].

Chorea-acanthocytosis is an uncommon autosomal latent illness brought about by changes in the 73-exon long VPS13A quality that encodes the protein Chorein. The transformations in this quality might bring about complete shortfall of the protein or practical weakness. It's job in mitochondrial support, actin polymerization, intracellular dealing of the vesicles and lipid amalgamation have been recently exhibited. Likewise, for ADHD side effects, inclusion of frontostriatal network is proposed to assume a significant part. This idea was additionally upheld by different information from neuroimaging, neuropsychology, hereditary qualities and neurochemical studies. Nonetheless, the specific pathogenesis of chorea-acanthocytosis stays subtle.

At first, patients might give mental or mental side effects a very long time before the neurological indications create. These side effects incorporate melancholy, schizophrenia-like ways of behaving and impulsive problems. This generally prompts misdiagnosis and puzzles the neurological indications. Hindered memory or leader capabilities might be available. Character changes, Touretteism and trichotillomania may likewise be seen.

For 33% of the patients, the underlying side effect is a seizure, regularly summed up type 14. Generally, the predominant development problem is chorea. Taking care of dystonia is an unusual and trademark side effect of the illness. This side effect happens while eating. Contact of food and patient's tongue triggers fit of the tongue and pushes the food out, subsequently weight reduction might be found in patients. Ofacial dystonia is likewise a typical indication and may present as scowling, compulsory vocalization and dysarthria as well as self-ravaging tongue and lip gnawing. Another significant show is 'Elastic man appearance'. This alludes to truncal flimsiness and unexpected trunk fits which might bring about head banging and self-hurt. Once in a blue moon, Parkinsonism might be experienced in these patients [5].

Analysis of chorea-acanthocytosis is extremely difficult. Its initial mental side effects by and large leads doctors to wrong analysis and bewilder the neurological signs that foster later in the sickness course. Lab workups and imaging procedures might be useful in the finding. Serum Creatine Kinase levels are generally raised in these patients. Presence of acanthocytosis in fringe blood smear might be a useful symptomatic sign; nonetheless, presence or number of acanthocytes don't correspond with sickness movement. In this manner, fringe smear has extremely low awareness for the conclusion. It is critical to take note of that, non-presence of acanthocytes can't preclude the infection. In neuroimaging, caudate head decay and expansion of front horns of the horizontal ventricles are seen with MRI13. Fluorodeoxyglucose - positron discharge tomography (FDG-PET/CT) filter shows hindered glucose digestion inside the basal ganglia. To affirm the finding, western smudge might be performed.

Our patient originally given engine side effects (choreiform developments) in her mid 20s. Later throughout sickness, she created neglect, consideration

lack, orofacial dyskinesia and a spasm problem that is influencing her eyes. In mental assessment, she was determined to have ADHD.

Evaluation of this patient was complex and very demanding. Despite being a neurological disease, the initial symptoms may vary widely, and these patients may be evaluated in different departments even for years. Unrevealing the underlying disease may not be possible if the physician is not aware of the neuroacanthocytosis or includes it in the differential. To evaluate our patient properly, we needed a team including psychiatrist, hematologist and a neurologist. Patients, depending on their symptoms and severity, may present to any of these three specialties for initial evaluation. For this reason, multidisciplinary approach is very important to rule out other pathologies and include the neuroacanthocytosis in the differential diagnosis.

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CONCLUSION

In this case report, we presented a rare case of acanthocytosis that presents with ADHD in the later stages of disease. We wanted to emphasize the importance of multidisciplinary care in the management of choreoacanthocytosis.

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CONFLICT OF INTEREST

The authors certify no conflict of interest with any financial organization about the material described in the manuscript.