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Case Report

Neuroacanthocytosis: A Rare Case Report

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Abstract: Neuroacanthocytosis (NA) is a diverse group of disorders in which nervous system abnormalities co-occur with red blood cell abnormalities (acanthocytes). We report the case of a 30-year old male who presented with eight months' history of progressive involuntary movements, dysphagia, difficulty in walking, and slurred speech. On examination there were multiple bite marks over lips and cheeks (features suggestive of self-mutilation) with dysarthria and nasality of voice along with hypotonia and areflexia. There was presence of involuntary movements suggestive of chorea, facial and vocal tics. He had normal power in all the four limbs and the plantar reflex was flexor bilaterally. On investigations his peripheral blood smear showed presence of acanthocytes. Hence a diagnosis of neuroacanthocytosis was made.

Keywords: Neuroacanthocytosis, Chorea, Acanthocytes

Introduction

Neuroacanthocytosis (NA) refers to a diverse group of disorders in which nervous system abnormalities co-exist with red blood cell abnormalities namely acanthocytes (deformed erythrocytes with spike-like protrusions). NA was first described in 1960 as "Levine–Critchley syndrome"[3] It is estimated that 500–1,000 people worldwide have chorea-acanthocytosis (ChAc), an entity under NA syndrome.[1] Acanthocytes are contracted erythrocytes with unevenly distributed thorny projections.[2]

Neurologic problems usually consist of either movement disorders or ataxia, personality changes, cognitive deterioration,[4] axonal neuropathy, and seizures. The characteristics of NA are movement disorder, behavioural and cognitive changes. The movement disorders comprise of chorea, dystonia and tics. The orofacial dystonia with dystonic tongue is conspicuous movement and may interfere with eating.[5] Orofaciolingual dyskinesia may also lead to lip and tongue bites with marked dysphagia and dysarthria in many patients. Neuro-acanthocytosis is classified into 2 broad categories:

1. Core Neuroacanthocytosis syndromes.
2. Conditions with altered lipid metabolism.

Core Neuroacanthocytosis syndromes consist of Choreo-acanthocytosis (ChAc), Mcleod syndrome, Huntington disease like 2 (HDL2) and Pantothenate kinase associated neuro-degeneration (PKAN).

Case Report:

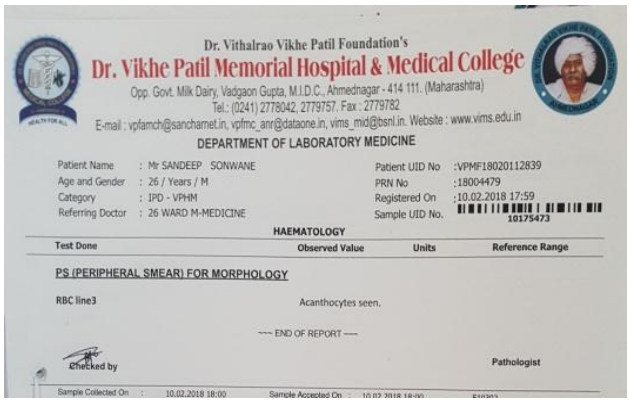
A 30 years old male, without any known comorbidities was referred to medicine outpatient with history of insidious onset progressive involuntary movements, dysphagia and difficulty in walking for eight months and slurred speech for six months. The onset of symptoms was insidious and course slowly onset progressive. Movements were jerky, repetitive, involuntary and purposeless, involving whole body including head, face and neck. The movements could not be suppressed voluntarily but used to disappear during sleep. There was associated shrugging of shoulders and spasmodic movements of the neck. There were also stereotyped repetitive movements particularly involving face and while speaking which could be suppressed at will but the patient had increasing urge which could only be relieved by those movements. He also had difficulty in eating due to abnormal twisting movement of tongue while chewing and hence he used to push the food bolus with fingers inside the mouth. He walked with a bizarre gait with truncal instability and sudden violent jerky movements of the entire body. Since last six months he had developed slurring of speech with a nasal twang.

There was no history suggestive of Rheumatic fever, use of antipsychotic drugs, psychiatric illness, cognitive dysfunction, jaundice or haematemesis or any addictions in the past. He was born out of non-consanguineous marriage and had two siblings. There was no family history of jaundice or history suggestive chronic hepatocellular failure to suggest possibility of Wilson's disease. His elder sister had similar illness and she had committed suicide, probably due to depression secondary

to the illness.

On examination, the patient was conscious, oriented, thin built and poorly nourished. His vital parameters were within normal range. There were multiple bite marks over lips and cheeks. The neurological examination revealed normal higher mental functions. The cranial nerves examination was unremarkable except for nasal quality of voice and occasional nasal regurgitation. Examination of lower four cranial nerves revealed normal palatal movements and normal gag reflex. Indirect laryngoscopy was also unremarkable. Examination of ocular fundi and slit lamp was normal. Motor system examination revealed wasting of all groups of muscles of upper and lower limb with hypotonia and areflexia. However, power in all four limbs was normal and plantar reflex bilaterally flexor. Sensory and cerebellar examination was normal. The gait showed truncal instability and sudden, jerky, extension movements of trunk while walking. There was sudden, intermittent, momentary flexion of hip and knee joints while walking giving it resemblance of rubber man's Gait. Extrapyramidal system examination revealed choreiform movements of all four limbs involving large joints. There was intermittent shrugging of shoulders and dystonic neck movements along with facial and vocal tics. The vocal tics were in form of whistling sound that interfered with talking. (Figures 1, 2). The examination of other systems was unremarkable.

On investigations peripheral smear showed presence of acanthocytes (Figure 3, 4).



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DEPARTMENT OF LABORATORY MEDICINE

Patient Name	: Mr SANDEEP SONWANE	Patient UID No	:VPMF18020112839
Age and Gender	: 26 / Years / M	PRN No	:18004479
Category	: IPD - VPHM	Registered On	:10.02.2018 17:59
Referring Doctor	: 26 WARD M-MEDICINE	Sample UID No.	101754 73

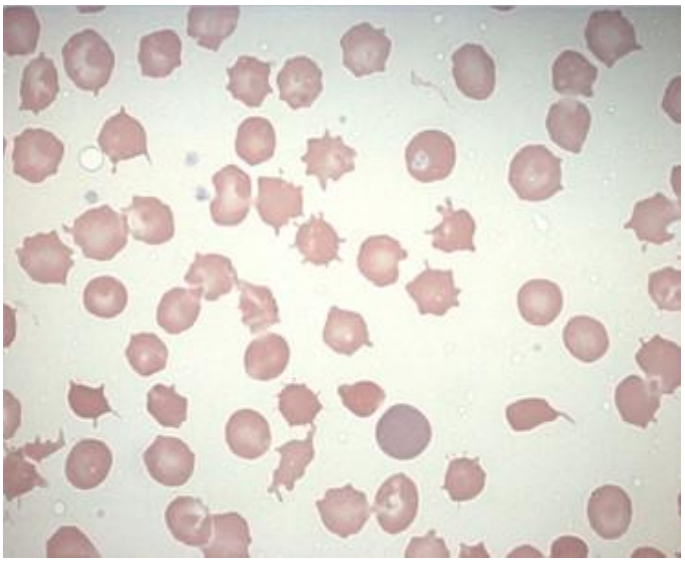
HAEMATOLOGY

Test Done	Observed Value	Units	Reference Range
PS (PERIPHERAL SMEAR) FOR MORPHOLOGY			
RBC line3	Acanthocytes seen.		

---- END OF REPORT ----

Checked by — Pathologist

Sample Collected On : 10.02.2018 18:00 Sample Accepted On : 10.02.2018 18:00



Muscle CPK-MB was raised. All other investigations including, ECG, echocardiography, chest X ray, liver function tests, renal function tests, thyroid function tests, serum lipids, lipid electrophoresis, serum ceruloplasmin, urinary copper, serum vitamin B 12 levels and electroencephalogram were normal. Genetic testing of PCR for CAG repeats of Huntington's disease was normal. Nerve conduction study showed sensorimotor axonal neuropathy. MRI Brain Plain revealed no abnormality.

On the basis of positive family history and presence of characteristic orolingualfacial dyskinesia, feeding dystonia, self lip and tongue mutilation, choreiform movements and peripheral neuropathy along with presence of acanthocytes in peripheral smear, raised creatinine kinase and normal lipids and lipoproteins, led to the diagnosis of neuroacanthocytosis (ChAc variant).

He was treated symptomatically with tetrabenazine, haloperidol, clonazepam and supportive treatment. The patient responded well to treatment with marked reduction in abnormal movements and he could eat properly with considerable ease.

Discussion:

Neuroacanthocytosis (NA), inherited as autosomal recessive trait, is a rare neurodegenerative disorder in which neurological abnormalities consisting of movement disorders

and behavioural and cognitive changes are found along with acanthocytes in the peripheral blood smear. Movement disorders consists of chorea, dystonia and tics. Clinical neuromuscular findings include areflexia, sensory-motor type neuropathy, weakness and muscular atrophy.

Our patient presented with chorea, tics, peripheral neuropathy and a positive family history. Since our patient had presented with adult onset chorea, the Huntington's disease (HD) was considered as strong possibility. But in HD, the presentation of chorea is more severe, usually accompanied with optokinetic nystagmus, neuropsychiatric symptoms, cognitive impairment and dementia[5]. HD does not present with peripheral neuropathy. Genetic testing with PCR for CAG repeats of Huntington disease was also negative, ruling HD out. Other causes of adult onset chorea include drug-induced, pregnancy, stroke, thyroid disease, and lupus. The idiopathic NA is usually seen in older patients. The young age and bilateral manifestations made stroke an unlikely differential. Patient's sex, age, and drug history and normal thyroid function test essentially ruled out other causes. Tardive dyskinesia was unlikely as there was no history suggestive of psychotropic drug use. Wilsons disease was another differential but was ruled out by absence of any signs of liver cell failure, no K-F ring on slit lamp examination and normal serum ceruloplasmin and urine 24-hour copper level. Tourette syndrome has an early age of onset of symptoms and is mainly dominated by tics, making this diagnosis unlikely.[6]

Finally, the core NA syndromes namely, autosomal recessive ChAc, HDL2, X-linked McLeod syndrome and PKAN were the only possibility left. HDL2 is an autosomal dominant disorder associated with CAG/CTG trinucleotide repeats on chromosome 16. The age of onset of HDL2 is usually in the fourth decade. Though the presentation is same as that of ChAc i.e. choreoathetosis, dystonia, and parkinsonism, HDL2, however, does not involve orofacial dyskinesias, self-mutilation and peripheral neuropathy. Moreover, the typical features in HDL2 are hyperreflexia and cognitive impairment; both were absent in this patient. McLeod syndrome also presents with skeletal muscle wasting with chorea and acanthocytosis and may have an elevated CPK level as well. Cardiomyopathy is also commonly described in McLeod syndrome.[8,9] But McLeod Syndrome is associated with the Kell null blood group phenotype, not present in our patient. PKAN is an autosomal recessive condition with onset in childhood. The condition is included in the group of syndromes that typically involve neurodegeneration with brain iron accumulation. The typical MRI findings of the “eye of the tiger” sign suggests the diagnosis. Finally, on the basis of clinical approach of elimination in the background of positive family history and clinical features of chorea, tics and dystonia with peripheral smear showing presence of acanthocytes a diagnosis of ChAc was made.

Conclusion:

Neuroacanthocytosis is a rare movement disorder. It should be suspected in patients with characteristic features of chorea with orofaciolingual dystonia in the back ground of positive family history. A simple test like demonstration of acanthocytes in peripheral blood smear can lead to the diagnosis of this rare disorder.

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