Chorea and Orofaciolingual Dystonia in a 40 Year Old Male

Lulup Kumar Sahoo¹, Kali Prasanna Swain², Ashok Kumar Mallick³, Geeta Mohanty³, Maheswar Samanta², Srikanta Kumar Sahoo¹

Abstract
Neuroacanthocytosis is a heterogeneous group of disorders which result in progressive neurodegeneration, predominantly of the basal ganglia, and erythrocyte acanthocytosis. We report a case of neuroacanthocytosis with typical phenotype of choreoacanthocytosis. A 40 year male presented with features of chorea and orofaciolingual dystonia producing eating and speech difficulties. There were features of self mutilation in form of lip and tongue biting. Peripheral blood smear examination revealed acanthocytes in our patient. Neuroimaging showed bilateral caudate atrophy and nerve conduction study showed motor axonal neuropathy. This case report describes the typical features and investigations to diagnose this rare disorder which is usually underdiagnosed.

Introduction
Neuroacanthocytosis is a rare genetic movement disorder. This clinical entity was described by Edmund Critchley from two families, one from USA, the other from UK¹ and was initially named as Levine- Critchley syndrome. It is characterised by movement disorder, behavioural and cognitive changes. The movement disorder consists of chorea, dystonia and tics. There is prominent orofacial dystonia with dystonic tongue movement interfering with eating. Many patients develop lip and tongue biting with prominent dysphagia and dysarthria. The term “acanthocyte” is derived from the Greek word for “thorn”. Acanthocytes are contracted erythrocytes with unevenly distributed thorny projections. These acanthocytes are distinct and unique for neuroacanthocytosis. We report a case with typical features of neuroacanthocytosis and how by simple clinical examination and peripheral blood smear study we can diagnose this rare disorder.

Case Report
A 40 year old male presented with abnormal, repetitive, involuntary, non-purposeful movements of the whole body including head, face and neck for one year. These movements disappeared during sleep and were not suppressed with voluntary action. These movements were associated with shrugging of shoulder and spasmodic movements of the neck. He also had difficulty in eating due to abnormal twisting movement of the tongue while eating. He used to eat by pushing the food bolus with fingers inside mouth. Occasionally, he had nasal regurgitation of liquids with resultant choking and coughing. He had frequent lip and cheek biting. Since last six months, he had slurring of speech with a twang. There was history of significant weight loss. There was no history of any psychiatric manifestation or behavioural changes or change in cognition. There was no history of blurring of vision, limb weakness or sensory symptoms. Bowel and bladder habit was normal. There was history of similar illness in younger sister.

General examination showed cachexia with lip biting and cheek biting marks (Figure 1) suggestive of self-mutilation and orofacial dyskinesia.

He was conscious, oriented with normal memory and behaviour. There was dysarthria with nasality of voice. There was generalised hypotonia with power of 5/5 in all limbs. Generalised areflexia with flexor plantar response was present. There was chorea in form of abnormal, repetitive, involuntary, non-purposeful movements of limbs, trunk and neck. He walked with bizarre gait with abnormal movements of the trunk. There was orofacial and lingual dyskinesia. Sensory, cerebellar and autonomic nervous system examination revealed no abnormalities. Other systemic examination was normal.

The routine investigations, complete blood count, renal function tests, liver function tests, serum electrolytes, serum ceruloplasmin were within normal limit. Creatinine phosphokinase was raised to 553 U/L (normal- 39 to 238 U/L). Peripheral blood smear showed multiple acanthocytes (Figure 2). Nerve conduction study showed motor axonal neuropathy. Magnetic resonance imaging (MRI) of brain showed significant bilateral caudate atrophy (Figure 3).

With the classical clinical features of chorea, orofaciolingual dystonia and feeding dystonia in the background of positive family history, the clinical diagnosis of autosomal recessive choreo-acanthocytosis was made. It was supported by presence of acanthocytes in peripheral blood smear with typical bilateral caudate atrophy in neuroimaging. Genetic confirmation of diagnosis could not be made due to unavailability of facility for analysis of VSP13A gene. He was treated symptomatically with dopamine depleter tetrabenzine. The patient responded well to treatment with gross reduction in abnormal movements and he could eat properly.
without much difficulty.

**Discussion**

Neuroacanthocytosis (NA) is a syndrome consisting of movement disorder, behavioural and cognitive changes. The core neuroacanthocytosis syndrome includes autosomal recessive choreo-acanthocytosis and X-linked Mcleod syndrome. Autosomal recessive choreo-acanthocytosis (ChAc) is characterised by adult onset at around 35 yr of age. The movement disorder consists of chorea, dystonia and tics. Parkinsonism may occur in more advanced stages. There is also prominent orofaciolingual dystonia producing frequent lip and tongue biting. It also interferes with eating due to feeding dystonia. Neuropsychiatric symptoms are prominent in neuroacanthocytosis and may appear several years before the onset of neurological manifestations. The gait of choreo-acanthocytosis patients may have a “rubber man” appearance with truncal instability and sudden, violent truncal spasms. Psychiatric manifestations are common. Most choreo- acanthocytosis patients have elevated levels of creatinine phosphokinase. In contrast to Mcleod syndrome, myopathy and axonal neuropathy are usually mild. Clinical neuromuscular manifestations include areflexia, sensory-motor neuropathy, variable weakness and atrophy. In at least one third patients, seizure, typically generalised, are the first manifestation of the disease. It is due to mutation of VPS13A gene which encodes for the protein chorein.

Mcleod syndrome, an X-linked disorder, begins at around 50 yrs of age and slowly progressive. Neuromuscular and cardiac involvement is more common in Mcleod syndrome. In contrast to Choreo-acanthocytosis, only exceptional Mcleod syndrome patients have lip or tongue biting, dysphagia, dystonia or parkinsonism. Huntington’s disease closely resembles neuroacanthocytosis and has similar phenotype of chorea, behaviour and cognitive deficits.

**Conclusion**

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

**References**