Chorea-acanthocytosis: A Case Report with Review of Oral Manifestations

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Abstract

Chorea-acanthocytosis (ChAc) is an autosomal recessive, progressive neurological disorder due to mutation in VPS13A gene causing defects in sorting of protein making the cell membrane unstable, leading to star-shaped erythrocytes. This neurological disorder includes features such as elevated creatinine kinase, atrophy of basal ganglia, and oral manifestations such as frequent cheek and tongue biting. It is a rare neurological condition with an estimate of <1000 cases worldwide. A case of 47-year-old male patient with a history of seizures and neurological problems presenting with oral ulceration has been discussed. The diagnosis of ChAc was confirmed by molecular investigations showing VPS13A gene mutation. The physical appearance includes chorea and dystonia with impaired gait. We attempt to highlight the oral features of ChAc. The oral manifestations include frequent tongue and cheek biting occurring due to dystonia affecting the muscles of head and neck region.

Keywords: *Chorea-acanthocytosis, orofacial dyskinesia, oromandibular dystonia*

Introduction

Chorea-acanthocytosis (ChAc) belongs to one of the types of a group of disorder known as neuroacanthocytosis. ChAc is a rare, progressive neurological disease. The lesion is very rare with reported literature limited to <200 cases. ChAc was first described by Critchley *et al*. in 1967 in a single family whose members showed features of progressive neurologic disease including self-mutilating lip and tongue-biting, dystonia, chorea, cognitive impairment, and acanthocytosis.[1] Although acanthocytosis is pathognomic to ChAc, only one case of ChAc without acanthocytosis had been reported.

Orofacial involvement is a prime feature of this lesion and sometimes the earliest symptom occurring along the progression of disease. The range of orofacial features is wide, but there is evident lack of literature that clubs together these findings. Furthermore, the orofacial features go unnoticed or are not assessed by the clinician because of the rarity and restricted knowledge of the disease.[2] We present a case report along with a thorough review of literature with emphasis on orofacial features.

Case Report

Here, we report a case of a 47-year-old male patient with neurological impairment. Physical examination showed ectomorph body type with thin and fine-boned appearance, impaired and sluggish gait with shrugged shoulder. The patient gave a history of weight reduction of 45 kg in the past 2 years. At the time of examination, the patient was not able to perform usual daily tasks due to muscle wasting [Figure 1] and involuntary movements of muscles of the limbs and orofacial region. He was under medication for epilepsy for the past 3 years. Memory impairment and insomnia were other neuropsychological findings.

The orofacial examination of the patient showed drooling of saliva, because of which the patient needed to wear a bib throughout the day [Figure 1]. Dysarthria and slurred speech were evidently noted. Due to involuntary tongue movements, tongue protrusion, and dystonia, the patient complained of difficulty in mastication and dysphagia. It was difficult for the patient to retract the tongue into his mouth. The patient also complained of frequent tongue and cheek bite. At the time of examination, a traumatic ulcer was also noted on the right buccal mucosa [Figure 2 shows sutured traumatic ulcer].

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Laboratory investigations demonstrated normal serum ceruloplasmin and elevated creatine phosphokinase levels. The molecular test for spinocerebellar ataxia and Huntington's disease allele mutation were negative. There was marked reduction in level of chorein protein and the confirmed diagnosis of ChAc was made by the detection of VPS13A gene mutation by Western blot method. Although acanthocytosis is pathognomonic to ChAc, peripheral blood examination was done on two occasions, which revealed that acanthocytosis was <2% [Figure 3].

Discussion

A total of 49 case series and reports were evaluated which revealed 121 cases. Out of these, 39 articles have assessed the orofacial features representing 97 patients. A high percentage (20%) of cases were noted for nonassessment of orofacial features.

Features such as dysphagia, orofacial dyskinesia, dysarthria, involuntary tongue movements, and vocalization represent the prime manifestations of this disease.[2,3] Tongue protrusion, feeding dystonia, drooling of saliva, facial grimacing, self-mutilating bites, and bruxism were other findings.[4,5] A case of facial cellulitis was seen because of repetitive trauma ultimately invading the connective tissue.

ChAc is an adult-onset, autosomal recessive neurodegenerative disorder manifesting with movement disorders comprising of chorea, tics, and dystonia. The involvement of head and neck region is mainly attributed to muscle dystonia. Orolingual dystonia and tongue protrusion can be early clinical symptom.[6] ChAc should be considered in patients with elevated levels of acanthocytes in a peripheral blood film on multiple occasions. The serum creatine kinase is also elevated. Protein function tests demonstrate reduced chorein levels and the diagnosis should be confirmed by analysis of genetic mutation of VPS13A gene.[7]

The disease is rare and due importance to the orofacial features is a must. It is not unusual to find case reports not assessing the orofacial features. We attempt to describe the full spectrum of orofacial features of ChAc comprehensively as it has not been done in literature in the past.

In 21 cases, orofacial symptoms were recorded among the early features indicating a clue for diagnosis to the clinician. These symptoms were orofacial dyskinesia, tongue protrusion, and involuntary tongue movements.[8] Although feeding dystonia is present in only 12.8% of the cases [Table 1], this feature is pathognomic to ChAc. It refers to the forceful expulsion of bolus by tongue from the mouth on touching it. Feeding dystonia along with involuntary tongue movements altogether creates difficulty in mastication.[9]

The mainstay of the treatment is supportive only. Botulinum toxin has shown promise in causing improvement in involuntary movements. Management of ChAc is symptomatic. Anticholinergics and dopamine-depleting drugs may be useful in controlling motor symptoms. There are case reports of deep brain stimulation of the internal

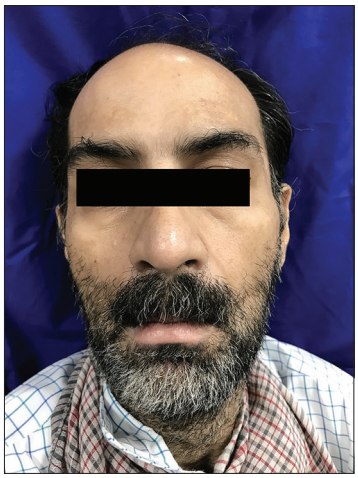

Figure 1: Clinical picture depicting muscle wasting associated with drooling of saliva

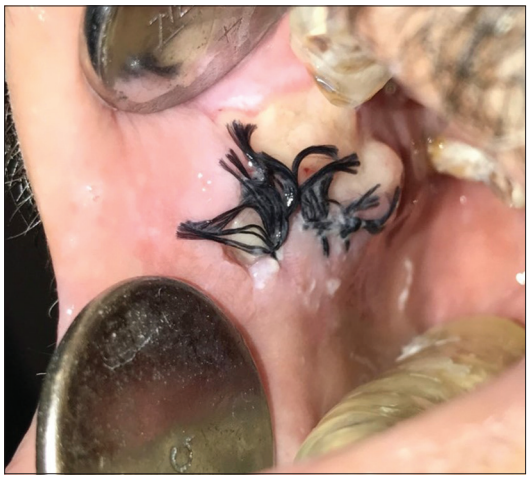

Figure 2: Sutured traumatic ulcer

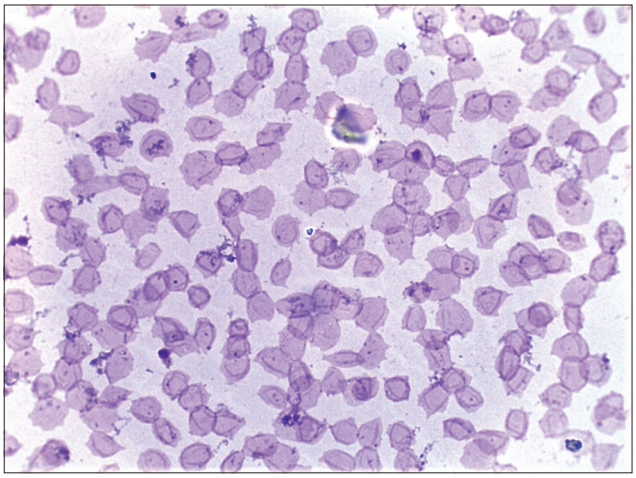

Figure 3: On peripheral blood examination, acanthocytosis was <2%

Table 1: Orofacial features seen in chorea-acanthocytosis patients

Prominent orofacial features (%)	Minor orofacial features (%)
Dysphagia: 62.7	Drooling of saliva: 9.9
Orofacial dyskinesia: 51.4	Facial grimacing: 7.9
Traumatic bite and ulcers: 47.5	Self-mutilation: 5.9
Dysarthria: 40.5	Bruxism: 5
Involuntary tongue movements: 27.7	Slow tongue movements: 2.9
Involuntary vocalizations and tics: 22.8	Lip smacking and licking: 2
Tongue protrusion: 13	Tongue tremor: 1
Feeding dystonia: 12.8	Facial cellulitis: 1

part of the globus pallidus showing significant results. Gastrostomy, physiotherapy, and occupational therapy may help patients to maintain nutritional status and functions of daily living.[10] Currently, there is an evident lack of specific treatment modalities with consistent results in improving the dexterity of muscle tone, especially the muscles of mastication and tongue.

Conclusion

We attempt to elucidate the oral implications of ChAc in order to better understand the spectrum of oral features seen in this rare neurological disease. Due to nonassessment of oral features in many case reports and rarity of the disease, the data are very limited, but the presence of oral features as an early manifestation can be useful to aid the dental surgeon to early diagnosis and emphasize the imperative role of dental surgeon. Diagnosis of ChAc should be considered for patients presenting orofacial dystonia with marked tongue involvement, feeding dystonia, and neurological features.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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