Case Report

A case of choreoacanthocytosis with marked weight loss: Impact of orolingual dyskinesia

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Choreoacanthocytosis (ChAc) is a rare autosomal recessive neurodegenerative disorder characterized by progressive onset of hyperkinetic movements and red cell acanthocytosis. The most striking clinical feature is that of the orofacial and lingual movement abnormalities leading to severe feeding difficulties. Maintenance of proper nutrition in ChAc is a challenge. We report on a case of ChAc in a 32-year-old male in whom dramatic weight loss due to orolingual dyskinesia was the major consequence of the disease. This case report warrants more attention to the impact of orolingual dyskinesia on nutritional status in patients with ChAc.

Key words: Choreoacanthocytosis, orolingual dyskinesia, weight loss

Introduction

Choreoacanthocytosis (ChAc) is a rare autosomal recessive disorder: It is characterized by orofacial dyskinesia, hyporeflexia, seizures, aberrant behavior, atrophy of the caudate nucleus and putamen and acanthocytes in the blood with a normal concentration of lipoproteins. The most striking clinical feature is that of the orolingual movement disorder. Few case reports have been devoted to the impact of orolingual dyskinesia or dysphagia as the dominant clinical picture in patients with ChAc. We report on a case of ChAc in a 32-year-old Iranian male in whom significant weight loss due to orolingual dyskinesia was the major consequence of the disease.

Case Report

A 32-year-old male with drooling and involuntary movements of tongue and limbs was admitted to our service. The fourth child of non-consanguineous parents, his gestation, delivery and early developmental milestones had been unremarkable. The patient was well until early adolescence when he began to show motor tics such as jerky movements, hitching up his shoulders and frequent sniffing. Since 3 years ago, drooling, impaired deglutition and occasional tongue biting have been added. Gait problem due to excessive twisting of feet evolved later. These symptoms were progressive. At the time of admission, he was not able to perform usual daily tasks because of severe involuntary movements of limbs, tongue and mouth. He was also unable to hold up his neck and head. On admission, he had a body weight of 45 kg and a body mass index of 16.53 kg/m². (His height was 1.65 m.) He reported significant weight loss of about 30 kg within the past year. Dietary history was normal. Family history was unremarkable; the patient had five siblings (two sisters aged 34 and 40 and three brothers aged 24, 27 and 30), who were all clinically normal.

On neurologic examination, he scored 27 out of 30 in Mini-Mental Status Examination. He had abnormal speech and produced incomprehensible sounds. Cranial nerves were intact except for orolingual dyskinesia and impairment of deglutition. He exhibited choreathetotic movements in limbs and had obvious pyromotor restlessness, which led to frequent throwing of surrounding objects. Motor power was 4 out of 5 in upper limbs and right lower limb and 3 of 5 in left lower limb. Left-sided foot drop was detected as well. Superficial cutaneous necrosis of distal phalanx of left finger and scattered scratches were also seen. Deep tendon reflexes were hypoactive in upper limbs and lost in lower limbs. Both plantar reflexes were flexor; His sensory examination was normal. Neither ataxia nor dysmetria was found in finger-to-nose and heel-to-shin tests. No behavioral abnormality including apathy was detected in the patient.

Routine biochemical tests and blood cell count were within normal limits. Serum electrolytes and thyroid function tests revealed no abnormality. Serum B12, folate, copper, ceruloplasmin and lipoproteins and 24-hour urine copper were all within normal range. ANA, anti-dsDNA, ANCA and antiphospholipid antibodies revealed no abnormality. The serum level of CPK was elevated (780 IU/L, normal range was 24-195 IU/L). Light microscopic assessment of wet blood film preparation of EDTA blood sample revealed no abnormality. The patient was treated with physiotherapy and occupational therapy to control his dyskinesia. He was discharged with a body weight of 40 kg and a body mass index of 16.05 kg/m². (His height was 1.65 m.)

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showed that more than 50% of the patient’s red blood cells were acanthocytes [Figure 1]. Brain CT scan and MRI revealed mild bilateral atrophy of caudate nuclei [Figure 2]. Two of the patient’s brothers, who were available, were clinically normal and had no acanthocytes on wet blood film preparation. He was placed on sodium valproate (600 mg/day) and tetrabenazine (37 mg/day) and showed some amelioration of his involuntary movements. A feeding tube was also inserted. The patient gained about 10 kg within the first 6 months after insertion of feeding tube.

Discussion

ChAc is a rare neurodegenerative disease characterized by a progressive movement disorder, a myopathy that can be subclinical, cognitive and behavioral changes and acanthocytosis of the red blood cells.[5] It is an autosomal recessive disorder caused by mutations in VPS13A gene on chromosome 9q21 that encodes a large protein called chorein.[6] Mean age of onset in ChAc is about 35 years of age, although it can develop as early as the first decade or as late as the seventh decade.[1] A full spectrum of movement disorders may be seen, including tics, dystonia and akinetic-rigid syndrome.[1]

Involuntary movements particularly affect the orofacial region, causing tongue and lip biting, vocalizations, dysphagia and dysarthria.[6] This orolingual movement disorder, characterized by a combination of dystonia and chorea and marked pseudobulbar disturbance, is the most striking clinical feature.[1,7] Dysphagia is also common and sometimes progresses to severe feeding difficulty. Dystonic protrusion of the tongue induced by chewing and attempted swallowing, sometimes referred to as ‘feeding dystonia,’ can result in mutilating lip and tongue.[1]

Although in this case, muscle wasting and energy expenditure from choreathetoid movements might account for some weight loss, orolingual dyskinesia leading to feeding difficulty was the major cause. Behavioral disturbances may also contribute to weight loss in ChAc, but we did not detect any behavioral abnormality in this patient. Insertion of feeding tube and pharmacologic therapy led to partial amelioration of symptoms and the patient began to regain weight.

ChAc runs a chronic progressive course and may lead to major disability within a few years. There is no cure and life expectancy is reduced. Treatment for the disease is symptomatic and supportive. Maintenance of proper nutrition in ChAc is a challenge. A feeding tube may be needed for some patients, as the disorder progresses, to assist feeding and to prevent aspiration.[8] Antipsychotic drugs can provide stage-dependent relief from chorea and tics. Severe feeding difficulty may require gastrostomy. Botulinum toxin may also be helpful to decrease the orofaciobuccolingual dystonia that interferes with eating.[9]

This case report warrants more attention to the impact of orolingual dyskinesia on nutritional status in patients with ChAc.

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References