A case of chorea-acanthocytosis with suicidal ideation and obsessive-compulsive disorder

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SUMMARY

Chorea-acanthocytosis is one of the neuroacanthocytosis syndromes; a rare (1–5 per 1,000,000) and progressive neurodegenerative disorder characterized by abnormalities in the nervous system accompanied by erythrocyte acanthocytosis. Various neurological dysfunctions and psychiatric symptoms coexist, significantly reducing both quality of life and life expectancy. Due to the rarity of the disease, diagnosis can sometimes be delayed; the initial presentation may include vague cognitive or psychiatric symptoms, leading to prolonged misdiagnoses and incorrect management. In middle-aged adults presenting with chorea and tic-like involuntary movements alongside psychiatric disorders, neuroacanthocytosis syndromes should always be considered. A thorough neurological and psychiatric examination should be conducted, and necessary imaging and laboratory tests should be performed.In this case report, we present the detailed diagnostic evaluation process of a patient suspected of having chorea-acanthocytosis with neuropsychiatric symptoms, in light of the existing literature.

Key words: Neuroacanthocytosis, feeding dystonia, movement disorder, obsessive compulsive disorder, vocal tic

INTRODUCTION

Neuroacanthocytosis (NA) is a rare syndrome characterized by the coexistence of abnormalities in the nervous system and erythrocyte acanthocytosis. The estimated prevalence is fewer than 1 to 5 cases per 1,000,000 individuals (1). It leads to a variety of neurological dysfunctions including seizures, movement disorders, peripheral neuropathy, speech–swallowing difficulties, psychosis, and dementia, leading to reduced life expectancy (2).

NA syndromes can be divided into several groups: 1. Core NA syndromes (1a. Chorea-Acanthocytosis, 1b.McLeod syndrome) 2. Degenerative disorders occasionally associated with acanthocytosis 3. Disorders related to decreased blood lipoproteins and acanthocytosis 4. Paroxysmal dyskinetic disorders (3).

Chorea-acanthocytosis (ChAc) is a progressive neurodegenerative disease that may initially present with subtle cognitive or psychiatric symptoms, often years before neurological signs appear (1). Historically, clinicians have focused primarily on the progressive external motor features of the disease; however, increasing evidence indicates that these disorders may also present with significant psychiatric and neurocognitive comorbidities (3).

In this report, we present a case with neuropsychiatric complaints in whom a diagnosis of chorea-acanthocytosis was considered. This case draws attention to the rare coexistence of obsessive–compulsive symptoms and suicidality in a genetically confirmed patient with Chorea-Acanthocytosis, highlighting the need for awareness of psychiatric-onset presentations in neuroacanthocytosis syndromes.

Case Presentation

Clinical Course

A 32-year-old single male was admitted to our inpatient unit due to the presence of vocal tics,

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feeding dystonia, weight loss, obsessive thoughts, involuntary movements, and subsequent suicidal ideation. His complaints had begun five years earlier with intermittent syncopal episodes, which were later accompanied by compulsive reassurance seeking and hoarding behavior. Following his first psychiatric evaluation that same year, he was treated with topiramate and lamotrigine; however, impulse control difficulties, excessive spending, gait disturbance, problems with anger regulation, and depressive symptoms gradually emerged. Within the past year, he had been hospitalized twice at different centers with diagnoses of Generalized Anxiety Disorder and Conversion Disorder but no significant clinical improvement was observed. Six months before admission, he developed orofaciolingual dyskinesia with involuntary lip biting and gasping vocal tics. He required a straw to consume liquids and experienced weight loss; 7 kg in one month, 30 kg overall. Four months earlier, he had been admitted to the intensive care unit following a generalized tonic-clonic seizure, complicated by right arm weakness. Initially confined to the facial region, tics progressed to involve shoulders and hips.

Over time, his obsessive–compulsive symptoms, which had initially appeared as reassurance seeking and repetitive behaviors, progressively worsened; he spent most of his day engaged in ritualistic behaviors, such as stringing beads or painting in a fixed sequence and became anxious and irritable when interrupted. These repetitive acts occupied several hours daily and interfered with his ability to engage in social activities. He repeatedly made identical bracelets using beads of the same color and felt compelled to complete a specific number of them in a particular order or symmetry. He frequently sought reassurance about his illness and treatment, repeatedly asked the same questions, and rechecked his drawings or bead arrangements until they felt "just right." He described feeling a persistent sense of tension and discomfort until these tasks were completed exactly as he imagined. Although aware that these behaviors were excessive, he was unable to resist them, resulting in marked distress and functional impairment. The escalation of vocal tics led to social withdrawal due to embarrassment, while his depressive symptoms intensified, culminating in suicidal ideation. For further diagnostic clarification and treatment planning, he was admitted to our clinic.

Neurological and Psychiatric Findings

On psychiatric examination, the patient was conscious, cooperative, and oriented, with mildly impaired self-care. Speech was dysarthric, hypophonic, and frequently interrupted by vocal tics. The patient showed compulsive reassurance-seeking, depressed mood, mild distractibility, and coherent thought processes. Sleep was reduced, while appetite was preserved but limited by swallowing difficulties. Neurological examination revealed right hand strength of 4+/5, impaired vibration sense in the lower extremities, absent lower and hypoactive upper reflexes, postural instability, and orofacial self-injury with gasping movements. Past medical history included febrile convulsions, obstructive sleep apnea, and epilepsy. Family history revealed no psychiatric or neurological disorders.

The Structured Clinical Interview for DSM-5 Disorders (SCID) was administered as a screening tool. It indicated symptoms consistent with Major Depressive Disorder, Generalized Anxiety Disorder, Obsessive-Compulsive Disorder, and Adult Attention-Deficit/Hyperactivity Disorder. However, based on clinical evaluation, only depression and obsessive-compulsive symptoms were considered diagnostically relevant. Wechsler Adult Intelligence Scale (WAIS) indicated borderline intellectual functioning (Full-Scale Intelligence Quotient [FS-IQ]=77). The Addenbrooke's Cognitive Examination–Revised (ACE-R) yielded a total score of 82/100, with impairment most pronounced in memory and fluency. On the Yale-Brown Obsessive-Compulsive Scale (Y-BOCS), the patient scored 26/40, falling within the severe symptom range.

Laboratory and Genetic Results

Laboratory findings revealed elevated serum creatine kinase (CK: 2360 U/L), myoglobin (450.9 ng/mL), and aspartate aminotransferase (AST: 63 U/L). Other basic biochemical parameters and complete blood count were within normal limits.

Ceruloplasmin levels and 24-hour urinary copper excretion were normal, thereby excluding Wilson's disease. Brain MRI demonstrated caudate nucleus and putaminal atrophy, while EEG findings were unremarkable. EMG revealed reduced sensory nerve action potential amplitude in the right ulnar nerve, with no additional abnormalities. Peripheral blood smear revealed 3–4% acanthocytes. Genetic testing identified two heterozygous, likely pathogenic variants in the VPS13A gene: c.1795_1796del (p.Asn599Ter) in exon 19 and c.4313_4317del (p.Thr1438LysfsTer21) in exon 37.

Treatment and Follow-up

During hospitalization, clomipramine was titrated to 150 mg/day to target depressive and obsessive-compulsive symptoms, and quetiapine 75 mg/day was initiated to address sleep disturbances. The patient had previously failed to respond adequately to multiple SSRI trials; therefore, clomipramine was selected for its anti-obsessional efficacy and prior partial benefit.

Quetiapine was preferred due to its favorable tolerability in the presence of marked movement disorder and was gradually titrated to 400 mg/day to manage agitation and sleep disturbance. Lamotrigine, which had previously shown benefit, was reintroduced to help with persistent depressive symptoms and titrated to 100 mg/day. Over the course of admission, vocal tics, involuntary movements decreased, accompanied by moderate improvement in obsessive thoughts (Y-BOCS score reduced from 26 to 19). Suicidal ideation resolved, and no seizures were observed.

Outpatient follow-up over a three-year period included regular psychiatric and neurological monitoring. His final treatment regimen consisted of clomipramine 150 mg/day, lamotrigine 200 mg/day, diazepam 15 mg/day, quetiapine 400 mg/day, and tetrabenazine 25 mg/day. At follow-up, feeding difficulties improved, vocal tics and involuntary movements decreased, and depressive symptoms showed remission. Suicidal ideation did not recur. Implementation of a dental guard effectively prevented lip biting, and the patient and his family were referred to social services for future planning, given the progressive nature of the disease. He continues to be followed jointly in psychiatry and neurology clinics.

DISCUSSION

Neuroacanthocytosis refers to a group of rare neurodegenerative disorders characterized by orofaciolingual dyskinesia, seizures, psychiatric manifestations, additional movement abnormalities, and the presence of deformed erythrocytes known as acanthocytes in peripheral blood. Although symptom onset most commonly occurs in the third decade of life, cases have been reported across a wide age range, and the disorder is associated with reduced life expectancy (4). In our patient, symptoms began at the age of 26.

The two major NA syndromes are autosomal recessive chorea-acanthocytosis and the X-linked McLeod syndrome (5). Although they share many clinical features, the hallmark of ChAc is the distinctive feeding dystonia, considered pathognomonic, in which the tongue expels food from the mouth upon contact, often accompanied by profound weight loss. Additional features include orofacial dystonia and chorea, manifesting as grimacing, involuntary vocalizations, dysarthria, and tongue–lip biting (6). Consistent with these reports, our patient presented with feeding difficulties, food expulsion, significant weight loss, dependence on a liquid diet, involuntary vocalizations, oral self-mutilation, and dysarthric speech.

The percentage of acanthocytes in peripheral blood varies from 5–50% and does not correlate with disease severity (2). Since acanthocytes may only be detectable in later stages, their absence does not exclude the diagnosis. Repeated smears or saline-diluted wet preparations, as proposed by Feinberg et al. in 1991, may increase diagnostic yield (7,8). Serum CK levels, often moderately or markedly elevated, appear more reliable for diagnostic support. Approximately half of the patients also present with elevated liver enzymes (1). In our case, 3–4% acanthocytes were observed, but saline-diluted smears could not be performed due to laboratory limitations. Laboratory findings demonstrated markedly elevated CK with mildly elevated

AST, consistent with the diagnosis.

Seizures occur in about one-third of patients and may be the initial manifestation. Although systematic studies of seizure semiology are ongoing, most patients appear to meet the criteria for familial temporal lobe epilepsy (6). Peripheral sensorimotor neuropathy, often accompanied by the absence of deep tendon reflexes, is a frequent finding and can resemble motor neuron disease in its presentation (9). Our patient had a history of seizures, absent deep tendon reflexes, vibration sense loss, and reduced sensory nerve action potential amplitude in the right ulnar nerve on EMG.

Neuroimaging studies in ChAc commonly demonstrate caudate atrophy, similar to Huntington's disease. Putaminal atrophy, globus pallidus and striatal iron accumulation, and cerebellar atrophy may also occur (10,11). Metabolic studies have shown reduced metabolism in the caudate nucleus and putamen (12). Consistent with this, our patient's MRI revealed bilateral caudate and putaminal atrophy.

Chorea-acanthocytosis is inherited in an autosomal recessive manner. The VPS13A gene on chromosome 9q21 encodes the protein "chorein" (13). Recent studies have demonstrated that chorein expression is reduced in erythrocyte membranes and other tissues in patients with ChAc, representing an important diagnostic marker (12). In our patient, heterozygous pathogenic variants in exons 19 and 37 of the VPS13A gene were identified.

NA syndromes frequently present with executive dysfunction, obsessive-compulsive disorder, depression, and, less commonly, psychosis. These psychiatric manifestations may precede the onset of overt motor and cognitive decline (13). In ChAc, compulsive behaviors involving control, cleaning, symmetry, binge eating, and hoarding have been reported (3,13,14). Furthermore, characteristic self-injurious behaviors, such as tongue and lip biting, have also been interpreted as potentially related to OCD symptomatology (15). Given the central role of the caudate nucleus in the neurobiology of OCD, it is plausible that patients with ChAc, who typically develop caudate atrophy, manifest comorbid OCD (16,17). Our patient exhibited prominent OCD features, including hoarding, control-related obsessions, behavioral disturbances, anger dysregulation, and suicidal ideation. On SCID evaluation, he met criteria for MDD, GAD, OCD, and Adult ADHD, while WAIS testing indicated borderline intellectual functioning.

NA syndromes and their psychiatric comorbidities severely impair the quality of life of both patients and their families. Given their slowly progressive course, early recognition of psychiatric symptoms and appropriate management are essential for improving outcomes and quality of life. In our case, longstanding distress and diagnostic uncertainty contributed to the exacerbation of depressive symptoms and suicidality. Following diagnostic clarification and adjustment of treatment, notable improvements were observed in sleep, feeding difficulties, involuntary vocalizations, and obsessive symptoms, with complete resolution of suicidal ideation. In addition, consistent with previous recommendations (18), sustained multidisciplinary follow-up including physiotherapy, speech and occupational rehabilitation, and psychological support is strongly advised to optimize long-term outcomes and quality of life.

In conclusion, when evaluating psychiatric conditions with adult-onset movement disorders, clinicians should carefully investigate organic etiologies before assigning a primary psychiatric diagnosis. Educating patients and families regarding potential psychiatric symptoms, and encouraging treating physicians to consider psychiatric consultation when necessary, represent important steps toward comprehensive care.

Informed Consent: Written informed consent was obtained from the patient and his family for publication of this case report.

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