Neuroacanthocytosis is a rare, hereditary, heterogeneous, neurodegenerative disorder characterized by adult-onset involuntary choreiform movement and erythrocytic acanthocytosis in the peripheral blood, first described by Citchley et al. Neuroacanthocytosis is mainly classified into autosomal recessive chorea-acanthocytosis (ChAc) and X-linked McLeod syndrome (MLS). The clinical manifestations of neuroacanthocytosis resemble those of Huntington disease (HD), including chorea, cognitive impairment, and psychiatric symptoms. Neuroimaging features of neuroacanthocytosis are atrophy of the striata, particularly the caudate nucleus, and increased signal intensity from the striata on T2-weighted imaging similar to that seen in HD.

To the best of our knowledge, cerebellar atrophy associated with neuroacanthocytosis has been described in only 2 articles. We report herein the cases of 2 siblings with ChAc showing cerebellar atrophy along with atrophy and signal intensity changes in striata on MR imaging.

Case Reports

Case 1
A 35-year-old man presented with exacerbation of involuntary movements and erythrocytic acanthocytosis in the peripheral blood, first described by Citchley et al. and Levine et al. Neuroacanthocytosis is mainly classified into autosomal recessive chorea-acanthocytosis (ChAc) and X-linked McLeod syndrome (MLS). The clinical manifestations of neuroacanthocytosis resemble those of Huntington disease (HD), including chorea, cognitive impairment, and psychiatric symptoms.

Neuroimaging features of neuroacanthocytosis are atrophy of the striata, particularly the caudate nucleus, and increased signal intensity from the striata on T2-weighted imaging similar to that seen in HD.

To the best of our knowledge, cerebellar atrophy associated with neuroacanthocytosis has been described in only 2 articles. We report herein the cases of 2 siblings with ChAc showing cerebellar atrophy along with atrophy and signal intensity changes in striata on MR imaging.

Case 2
A 32-year-old man, the younger brother of the patient in case 1, presented with mild disturbance of talking and ataxia. He displayed mild oral involuntary movements, muscular hypotonia, and occasional mild choreal involuntary movements. No abnormalities of the eyes were apparent, unlike the patient in case 1.

MR imaging of the brain showed atrophy of bilateral striata, mildly increased signal intensity of the putamen on T2-weighted imaging, and cerebral atrophy and cerebellar atrophy as in case 1.

On laboratory examination, serum creatine kinase level was elevated to 1068 IU/L, whereas other levels were normal. ChAc was also diagnosed from clinical manifestations and findings from brain MR imaging and blood examinations. Genetic screening was declined and was not performed. The patient was diagnosed with ChAc on the basis of clinical manifestations and findings from brain MR imaging and blood examinations.

Discussion
The term neuroacanthocytosis is normally used to refer to ChAc and MLS. However, other movement disorders may display erythrocytic acanthocytosis, such as Huntington disease–like 2 and pantothenate kinase–associated neurodegeneration. Abetalipoproteinemia, which is included under the umbrella term of neuroacanthocytosis, is characterized by the absence of serum apolipoprotein B; symptoms include spinocerebellar ataxia and peripheral neuropathy without involuntary movement. Abetalipoproteinemia was not considered present in the patients described herein, based on the normal β-lipoprotein profiles and clinical symptoms.

The characteristic clinical features of ChAc are orofaciolingual dyskinesias, including feeding dystonia and self-mutilation. Conversely, characteristic clinical features of MLS consist of myopathy, hepatosplenomegaly, and heart disease. On laboratory examination, elevated serum creatine kinase levels...
and acanthocytosis are observed in both diseases. Absence or weakness of Kell antigen is a characteristic feature of MLS.

Both of the patients in our case report fulfilled the criteria for ChAc. Patient 1 had microphthalmia of the right eye and decreased visual acuity, whereas patient 2, the younger brother of patient 1, showed no visual handicaps, and visual handicaps associated with ChAc have not previously been reported. Thus, we consider that associations between the visual handicaps of patient 1 and ChAc may be poor.

The clinical manifestations of neuroacanthocytosis, ChAc, and MLS resemble HD with chorea, cognitive impairment, and psychiatric symptoms. Characteristic features of neuroacanthocytosis such as orofaciolingual dyskinesia and acanthocytosis help to distinguish this condition from HD. However, overlapping clinical manifestations between neuroacanthocytosis and HD and the low sensitivity for detection of acanthocytosis on microscopic examination preclude the diagnosis of neuroacanthocytosis. Genetic examination is very useful but is not simple to perform.

On neuroimaging, the characteristic findings of ChAc and MLS resemble those of HD with atrophy of the bilateral caudate nuclei and putamina, with or without increased signal intensity on T2-weighted imaging. Frontal lobe atrophy or general cerebral atrophy has also been reported. To the best of our knowledge, cerebellar atrophy in neuroacanthocytosis, as seen in the cases of our 2 patients, has only been mentioned in 2 articles. Tsai et al reported 1 case of obvious cerebellar folia atrophy without any abnormality of basal ganglia like spinocerebellar degeneration, though whether the case involved ChAc or MLS was unclear. Nicholl et al reported 1 case of cerebellar atrophy and hyperintense periventricular white matter. Although some reports have described cerebellar preservation in patients with neuroacanthocytosis, we consider cerebellar atrophy as an additional finding of neuroacanthocytosis. Cerebellar atrophy on neuroimaging may have been overlooked in cases diagnosed by CT alone without MR imaging because of involuntary movements.

At present, neuroacanthocytosis cannot be distinguished from HD solely on the basis of neuroimaging without clinical manifestations, laboratory examinations, and genetic examinations. In HD, cerebral atrophy is mentioned as a common finding, whereas cerebellar atrophy is mentioned as a minor finding. Rodda reported only 3 of 300 cases of HD displaying...
severe cerebellar atrophy. Cerebellar atrophy may thus help to
distinguish neuroacanthocytosis from HD on neuroimaging.

In conclusion, we have reported herein the cases of 2 sib-
lings showing cerebellar atrophy along with striatal atrophy
and signal intensity changes of the putamina. In addition to
atrophy and striatal signal intensity changes mentioned as a
characteristic finding of neuroacanthocytosis on neuroimag-
ing, cerebellar atrophy may represent an additional finding for
neuroacanthocytosis. More images of patients with neuroac-
thocytosis should be accumulated to confirm the presence
of cerebellar atrophy, with the hope that this manifestation
will enable differentiation between neuroacanthocytosis and
HD on neuroimaging.

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