worsening. Methylphenidate may exacerbate such hyperthyroidism symptoms as tachycardia, headache, insomnia, and emotional lability. In our first case, symptoms could not be attributed to medication since it showed good tolerability initially and required no dose adjustment. The International Classification of Headache Disorders includes headache attributed to hypothyroidism under the heading ''Headache attributed to other metabolic or systemic disorders'' (A10.7.1).[14] In our second patient, chronic headache may be attributed to hyperthyroidism since no other causes were found and symptoms improved after starting treatment with antithyroid drugs. Behaviour and mood disorders have been described in up to 21% of the patients with hyperthyroidism,[5] as in the 2 cases presented here (behaviour disorder in the first case and marked emotional lability and generalised anxiety in the second). We should also highlight that our 2 patients had insomnia. Elevated thyroid hormone levels may cause neuropsychiatric alterations or exacerbate neuropsychiatric symptoms already present.[7] In our first patient, a previous thyroid hormone measurement had yielded normal results, which suggests that ADHD and behaviour disorders had worsened with hyperthyroidism. On the other hand, our second patient had no history of psychiatric or neurological symptoms; we therefore feel that these were caused by Graves disease. Both patients displayed typical symptoms of hyperthyroidism (tachycardia, tremor), which helped guide the diagnosis. Hyperthyroidism should be included in the differential diagnosis of patients with acute-onset or slow-progressing behaviour disorders, especially when they display tachycardia and tremor. In-depth knowledge of the neuropsychiatric manifestations of this condition is therefore essential.

Conflicts of interest

The authors have no conflicts of interest to declare.

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2173-5808/

Neuroacanthocytosis: A new mutation☆

Neuroacantocitosis, una nueva mutación

Dear Editor:

Neuroacanthocytosis encompasses a group of neurodegenerative disorders characterised by neurological symptoms and spiculated erythrocytes. One of these disorders is chorea-acanthocytosis, an autosomal recessive condition usually manifesting in the second decade of life.[1] It is characterised by obsessive behaviour, impulse control disorders, infantile behaviour, cognitive impairment, seizures, high CPK levels, dystonia, chorea, and tics.[2,3]

Neuroacanthocytosis is associated with mutations in the *VPS13A* gene, which is located at 9p21.2; this gene codes for chorein, a protein found in the brain, testicles, kidneys, splenium, and erythrocytes. Chorein deficiency leads to apoptosis of striatal neurons, but the exact role of this protein is yet to be fully understood.[4,5]

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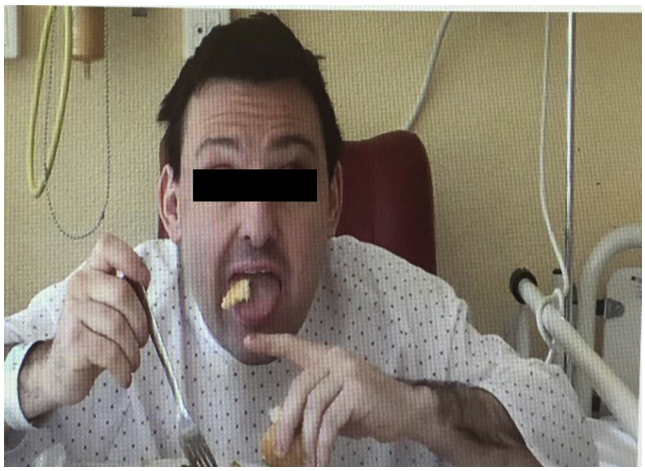

Figure 1 Dystonic movements of the tongue while eating led to expulsion of the bolus.

We present the case of a 38-year-old man whose psychomotor development had been normal until the age of 26, when he began to experience seizures. He had no personal or family history of interest and reported no long-term drug use. After the first seizure, he displayed extremely high CPK levels (> 4000 IU/L) which did not normalise between seizures (around 600 IU/L). The neurological examination revealed mild dysarthria, numerous facial tics, and dystonic movements of the tongue while eating, which led to expulsion of the bolus (Fig. 1). A neuropsychological examination found no cognitive impairment, although our patient displayed infantile behaviour with increasing disinhibition and impulsiveness. A neuroimaging study revealed caudate nucleus atrophy and slightly hyperintense putamina. He was negative for the Kell antigen. Results from a neurophysiology study (EMG) were normal. No mutations were detected in the gene *IT15*. A blood smear requested in view of our patient's seizures, high CPK levels, tics, and lingual dystonia (associated with radiological findings) revealed presence of acanthocytes, but they were not numerous enough to be considered pathological. Due to the strong clinical suspicion of acanthocytosis, the haematology department applied the technique described by Feinberg et al.[6]: erythrocyte dilution in normal saline/EDTA and in vitro ageing, which drastically increases the percentage of cells developing positive signs. Longer in vitro ageing incubation time resulted in a pathological increase in the number of acanthocytes (Fig. 2). A genetic study was conducted due to clinical suspicion of chorea-acanthocytosis. The molecular study detected 2 different heterozygous mutations in the *VPS13A* gene. In the first mutation, C was replaced by G (c.1901-3C>G); this pathological change may affect correct mRNA processing. In the other allele, we detected a deletion of 4 nucleotides (c.9446_9449del), which presumably leads to reading frame changes and appearance of a premature stop codon (p.ILe3149Thrfs*38). Although these genetic alterations had not been described previously in the databases that we had consulted, they were very likely to be pathological. Our patient's parents underwent a genetic study, which revealed that each of them carried one of these mutations. This confirmed that the molecular alterations observed in our patient were located in different alleles; genetic changes were very likely responsible for neurological symptoms.

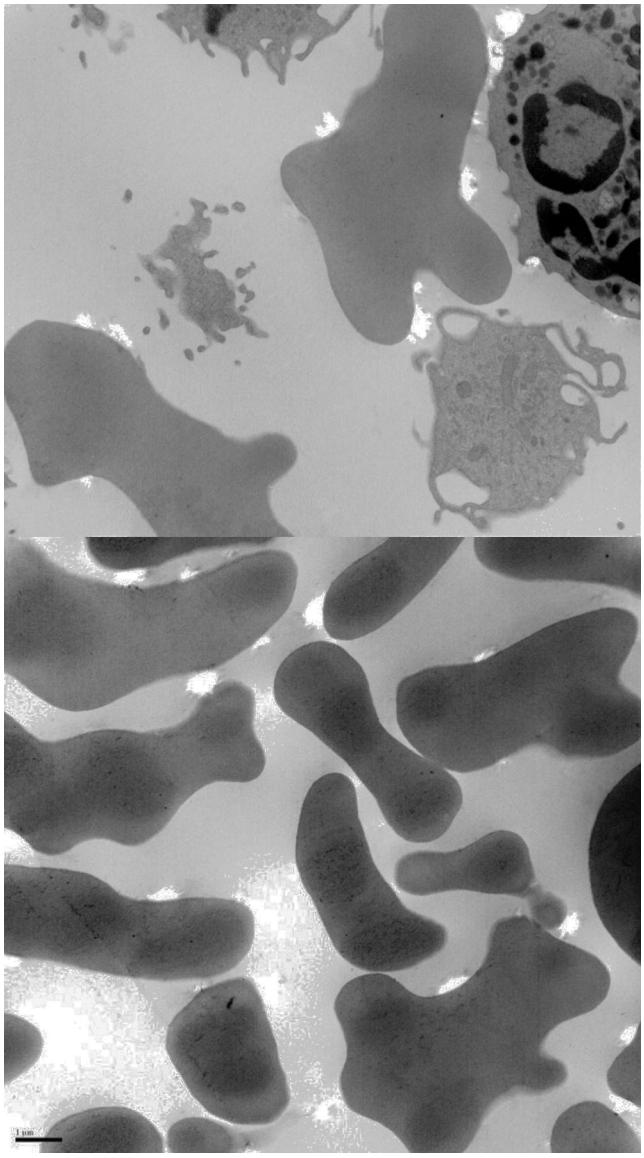

Figure 2 Electron microscopy images showing erythrocytes with thick cytoplasmic projections.

Chorea-acanthocytosis has a high genetic variability. In 2011, Tomiyasu et al.[7] found 36 pathological mutations, 20 of which had not been described previously. Sixteen of these patients displayed complete absence of chorein in erythrocytes.

Various antiepileptic agents had been employed to manage seizures, with poor results. Following the patient's diagnosis, we administered phenytoin dosed at 150 mg/8 h (necessary to maintain therapeutic plasma levels); our patient has remained seizure-free ever since. The patient was treated with SSRIs, clonazepam, tetrabenazine, trihexyphenidyl, pimozide, and risperidone for behaviour disorders and dyskinesia, with no clinical improvement.

We wish to highlight that blood smears to determine the presence of acanthocytes may yield false negative results; when there is a strong clinical suspicion of acanthocyto-

sis, the haematology department should use the Feinberg technique. In our patient, we achieved good seizure control with phenytoin, but we were unable to control movement and behaviour disorders. This patient displayed 2 previously undescribed mutations, one on each allele, which resulted in a wide array of symptoms.

Conflicts of interest

The authors have no conflicts of interest to declare.

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2173-5808/

Unilateral retrobulbar optic neuropathy as the initial manifestation of human immunodeficiency virus infection☆,☆☆

Neuropatía óptica retrobulbar unilateral como primera manifestación de la infección por el virus de la inmunodeficiencia humana

Dear Editor:

Infection with human immunodeficiency virus (HIV) is associated with a wide range of neuro-ophthalmological manifestations, from ocular motility disorders to impairment of the afferent visual pathway.[1] Most of these symptoms present in patients already diagnosed with HIV infection. However, some neuro-ophthalmological disorders, such as acute retinitis or optic neuropathy, may appear as initial manifestations of the disease. Vision loss in HIV-positive patients may indicate presence of a pathological process affecting structures ranging from the cornea to the visual cortex.[2] Most cases of optic nerve involvement are due to opportunistic infections (herpes zoster, cytomegalovirus, syphilis, toxoplasmosis, cryptococcosis, histoplasmosis), CNS tumours, or drug toxicity (ethambutol and didanosine).[3—6] HIV has also been described as one of the causes of optic neuropathy.[1] In these cases, the typical pattern of affectation consists of anterior ischaemic optic neuropathy or neuroretinitis secondary to severe autoimmune microangiopathy. Retrobulbar optic nerve involvement secondary to HIV infection usually manifests with long-term binocular asymmetrical vision loss.[7] We present the exceptional case of a patient with unilateral retrobulbar optic neuropathy which we attributed to the action of the HIV virus, and appearing as the initial manifestation of HIV infection.

Clinical case

Our patient was a 59-year-old patient who visited our hospital due to a 3-month history of vision loss affecting the

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