

## Short Report

## Neuro-acanthocytosis — a rare cause of chorea

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Neuro-acanthocytosis is a rare neurological disorder characterised by stereotyped chorea, especially of the mouth, areflexia and acanthocytes seen in the peripheral blood. No cases have been described in the literature from South Africa. We report here a case of neuro-acanthocytosis seen in a black woman who presented to Johannesburg Hospital.

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A 32-year-old, right-handed Zulu woman presented with a 2-year history of abnormal involuntary movements of the hands, arms and face, involuntary grunting, vocal tics and dysphagia. Her symptoms began abruptly with no precipitating factors and were progressive. She had never been under any psychiatric care or received neuroleptic agents. She denied ever having, and was not currently using, any prescription or over-the-counter medication, specifically anti-emetic preparations, and had undergone no exposure to witchdoctor toxins. There was no family history of neurological disorders, but a paternal cousin had a history of mental problems. She had one child who was well. Previously the patient had also been well and had never been treated for any other medical conditions. She did not drink alcohol or smoke and had never used any illicit drugs.

General examination showed her to be of short stature; she had obvious choreiform involuntary movements of the upper and lower limbs and made frequent grunting and smacking noises. Systematic examination did not reveal any further abnormalities.

On neurological examination, she had a Mini-Mental-State Examination score of 24/30 which was slightly below the expected finding. The rest of her higher functions were intact on examination. She had normal visual acuity, with a full range of eye movements, but demonstrated difficulty with generating voluntary saccades, and instead utilised head thrusts. Kayser-Fleischer rings were absent. She demonstrated orofacial buccal dyskinesia and vocal tics. Examination of the bulbar cranial nerves revealed a marked dysarthria. There was no evidence of tongue, lip or buccal mucosa lesions from biting. Dysphagia with regard to solids, as well as marked difficulty with mastication, was present. Motor examination revealed widespread stereotyped chorea of the limbs and trunk; this was much more marked in the upper limbs. Tone and power were normal. Ankle jerk reflexes were depressed bilaterally. Sensory and cerebellar examinations were normal. Her gait was dystonic.

**Laboratory investigations.** A full blood count was normal. Two additional fresh smears were submitted and 4% and 3% acanthocytes (Fig. 1), respectively, were seen (normally < 3%). Urea and electrolytes, liver function tests, thyroid function tests, a lipogram and a full auto-immune work-up were normal. The creatine kinase level was moderately elevated at 185 IU/l. Rapid plasma reagin and treponema pallidum haemagglutination syphilis serology were negative, as were tests for HIV antibody. Biochemistry, protein and microscopy assessments of the cerebrospinal fluid were normal.

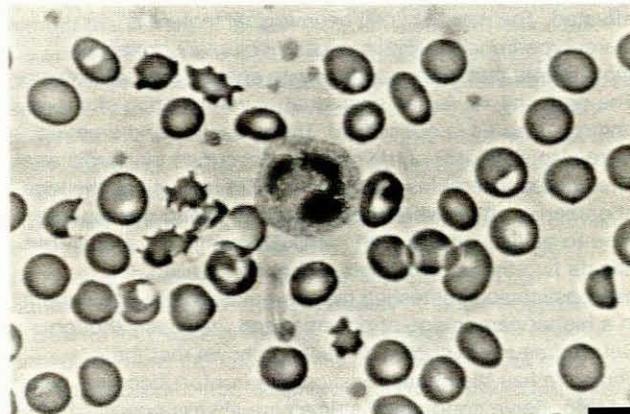


Fig. 1. Peripheral blood smear demonstrating large numbers of acanthocytes (scale: bar = 10 µm).

Tests for trinucleotide repeats, in order to consider the possibility of Huntington's disease (HD), were not performed since acanthocytes were noted early in the investigations.

**Neurophysiology.** Nerve conduction studies and electromyography failed to detect any abnormalities.

**Radiographic investigations.** Computed tomography of the patient's brain was normal. Specific attention was paid to the basal ganglia, including measurement of the intercaudate ratio; findings were within normal limits.

**Neuropsychological testing.** Owing to the patient's low level of education and language difficulties, formal neuropsychological testing could not be undertaken.

**Family studies and genetic studies.** Unfortunately the patient's parents and siblings were not available for testing since they lived in rural areas.

## Assessment and management

In view of the typical clinical and haematological features and the exclusion of other more common causes of chorea, a diagnosis of neuro-acanthocytosis was made. A lipogram was normal, excluding Bassen-Kernzweig abetalipoproteinaemia which is also characterised by morphologically abnormal red cells. The patient was started on haloperidol 8-hourly with some reduction in the chorea. She is currently being followed up at the movement disorders clinic.

## Discussion

Chorea is a common manifestation of diseases that affect the basal ganglia and its connections.<sup>1</sup> Neuro-acanthocytosis is a rare syndrome that was first described

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by Estes *et al.* in 1967.<sup>2</sup> A further 25 cases have been fully described in the literature since then<sup>3</sup> and case reports have appeared in the English and Japanese literature, although full details of the latter are not available. In 1991 Hardie *et al.* described 19 cases from the UK,<sup>3</sup> this study being the most detailed to date. Although the syndrome has been described in Europe, Asia and the USA, to the best of our knowledge there have been no cases from Africa.

The case described here displays many of the typical features described previously. Onset of symptoms is typically in the third decade, with males and females equally affected. The most striking neurological feature is stereotyped chorea, with marked involuntary orofacial movements that often interfere with speech and swallowing.<sup>3</sup> The orofacial movements are often a combination of chorea and dystonia as in our patient. The vocal tics and grunting were also seen in 9 of the patients described by Hardie *et al.* The chorea resembles that seen in HD but involves the legs at an earlier stage while the arm and forehead movements tend to be less severe than in HD. Dementia is variable, but affects two-thirds of patients, although detailed neuropsychological testing may reveal cognitive impairment in a higher percentage of patients than previously thought. Detailed neuropsychological testing shows that the dementia has the characteristics of a frontosubcortical type.<sup>4</sup>

Motor nerve conduction studies are always normal,<sup>3</sup> but sensory nerve action potentials are reduced in about 50% of reported cases. CT brain scans show varying degrees of abnormality from normal to generalised cerebral atrophy, particularly in the striatum and subcortical regions. A normal brain scan in our patient is in keeping with previous reports. In those cases which have come to necropsy, neuronal loss involving the caudate, putamen and pallidum has been described. The genetic basis of neuro-acanthocytosis remains unclear with autosomal-dominant, recessive and X-linked genetic inheritance proposed. Sporadic cases, such as the patient under discussion, have been reported, but their frequency is probably underestimated.

Awareness of this cause of chorea should prompt fresh blood slides to be examined in any patient with stereotyped or patterned chorea. While most patients present with a movement disorder, some well-documented patients with familial neuro-acanthocytosis did not.

## Conclusions

Neuro-acanthocytosis is a rare neurodegenerative disorder which is underdiagnosed worldwide. We report here a case seen at Johannesburg Hospital and suggest that the condition occurs in black Africans and should be considered in patients who present with chorea. Atypical features such as vocal tics, orofacial dyskinesia or areflexia may add to the clinical suspicion.

## REFERENCES

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