Molecular analysis

After informed consent had been obtained from the brothers, genomic DNA was extracted from peripheral blood by standard procedures. Exons of the *XK* gene were subsequently amplified by polymerase chain reaction as described by Ho *et al*.[1] The analysis showed a five base deletion in exon 3 at nt positions 938 to 942 from the 5′ end of the cDNA. This mutation results in a frame shift at codon 286 and the premature stopping of translation at codon 301, as reported previously.[2] This mutation was found in both cases 1 and 2, whose clinical phenotypes were extremely different.

After mutation analysis of the *XK* gene, we confirmed the presence of acanthocytes in a peripheral blood smear of case 1.

Comment

To date, the clinical features of McLeod syndrome have been reported to be heterogeneous.[2 5] The clinical features and conventional pathological findings in this condition are sometimes difficult to distinguish from other neuromuscular disorders because the expression of symptoms and signs seems to be variable, even among siblings.[2 5] In many cases, chorea, seizures, or muscular atrophy are the most frequently presented symptoms. Danek *et al* recently reported clinical features of 22 affected patients with mutation analysis of the *XK* gene.[2] In their investigations, limb chorea—which reflects CNS involvement in McLeod syndrome—was described in all patients. It is extremely difficult to make a diagnosis of this disease where the symptoms and signs are restricted to the peripheral nervous system.

In the present investigation, case 2 was characterised clinically by choreic movement and mild muscular atrophy, frequently seen in the reported cases of McLeod syndrome. In contrast, the symptoms in case 1 were extremely rare. Case 1 showed late onset of symptoms, slowly progressive weakness and amyotrophy of the lower extremities, areflexia, glove and stocking type sensory impairment, an increased level of serum CK, and pathological features with axonal degeneration of the nerve biopsy specimen. He showed no apparent central nervous system involvement 14 years from onset.

Our case 1 was clinically and pathologically indistinguishable from an axonal form of Charcot-Marie-Tooth disease without McLeod serology.

McLeod syndrome should be considered in patients with axonal sensorimotor neuropathy and high CK activity. Abnormal red cell morphology may be a clue to the diagnosis.

M Wada, M Kimura, M Daimon, K Kurita, T Kato
Third Department of Internal Medicine, Yamagata University School of Medicine, Yamagata, Japan

Y Johmura, K Johkura, Y Kuroiwa
Department of Neurology, Yokohama City University School of Medicine, Yokohama, Japan

G Sobue
Department of Neurology, Nagoya University School of Medicine, Nagoya, Japan

Correspondence to: Dr Manabu Wada, Third Department of Internal Medicine, Yamagata University School of Medicine, 2-2-2 Iida-Nishi, Yamagata 990-9585, Japan; mwada@yacht.ocn.ne.jp

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NHS Direct for headache

NHS Direct is a government sponsored, nurse led, telephone helpline available throughout the United Kingdom, offering confidential medical advice without recourse to a doctor by using computerised assessment systems based on clinical algorithms.[1] As algorithms for the management of headache have been formulated, this might be construed as a condition for which NHS Direct would be well suited to offer an appropriate service. Following a protocol used in previous studies of the use of NHS Direct by patients attending neurology outpatient clinics,[2–4] patients with headache were specifically asked about their use of this service.

Of 1000 consecutive unselected patients seen in 118 general neurology outpatient clinics over a period of approximately 10 months by one consultant neurologist, headache was the principal reason for referral or patient complaint during consultation in 208 (21%), a frequency similar to that previously reported by others.[5] The neurologist's diagnoses, using standard diagnostic criteria,[6] were: chronic daily headache of tension type (157), drug overuse headache (12), episodic tension type headache (3), and migraine (34); one patient had a cerebral neoplasm, with typical postural features and visual obscurations, and one had coital cephalalgia. Of these 208 patients, 120 (58%) had heard of the NHS Direct telephone helpline. Of these 120 patients, 36 (30%; or 17% of all headache patients) had used the service; only three patients volunteered this information spontaneously. Of the 36 users, in 14 the call to NHS Direct related to headache (39% of NHS Direct users, or 6.7% of all headache patients); two volunteered this information spontaneously. The percentages for awareness and use of NHS Direct in this cohort are similar to those previously reported for an unselected general neurology outpatient clinic surveyed in 2002.[3]

Of those calling NHS Direct for advice about their headache, five of the 14 reported that they were told to go to hospital or call an ambulance immediately. The neurologist's diagnoses in these five patients were chronic daily headache of tension type in three, episodic tension type headache in one, and migraine without aura in one (in whom the reported NHS Direct diagnosis was cerebral haemorrhage). One patient was told to go to a local NHS walk-in centre (final diagnosis: chronic tension type headache), and another two patients were told to attend their general practitioner (both with chronic tension type headache). NHS Direct diagnosed transient ischaemic attack in a man thought by the neurologist to have migraine without headache (migraine equivalent). One patient with chronic tension type headache was told to lie in a dark room. One patient phoned for information about side effects of analgesic medication. Three could not recall the outcome of their call to NHS Direct.

Proposals for changes in the primary care of headache in the UK, issued by the British Association for the Study of Headache (BASH), described the role of NHS Direct in headache management as ''uncertain,'' as ''algorithms in use cannot provide for the taking of an adequate history to inform advice given.''[7] The current study, although hospital based and reliant on patient report, with all their inherent biases, has provided no evidence to contradict that view. The suggestions emanating from NHS Direct were neither dangerous nor useful. Hence the study does not suggest that NHS Direct can currently replace clinical assessment by a practitioner trained in the diagnosis and management of headache disorders.

A J Larner
Walton Centre for Neurology and Neurosurgery, Lower Lane, Liverpool L9 7LJ, UK

Correspondence to: Dr A J Larner; a.larner@thewaltoncentre.nhs.uk

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Isolated total tongue paralysis as a manifestation of bilateral medullary infarction

Isolated acute bilateral hypoglossal nerve (CXII) paralysis is a very rare clinical condition which has been described in the context of traumatic mechanical injuries to the nerves.[1] The two nuclei of CXII, located at the tegmentum of the medulla oblongata, are in close proximity and may be damaged at the same time.[2] However, isolated bilateral CXII paralysis has not been described in cases of medullary infarction. We report a patient presenting with isolated complete tongue paralysis and a small ischaemic area in the medulla affecting both CXII nuclei exclusively.