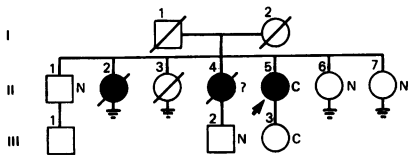

Figure The family pedigree. N = pericentric inversion of chromosome 4 not present. C = carrier of pericentric inversion. ? = ? inversion 4 carrier.

cytogenetic laboratories has not as yet resulted in any further information. Detailed neuropathological examination was not performed.

Case II5 had bilateral cataracts removed aged 55 years. Shortly after she was first observed to have an unsteady gait and slurred speech. On examination aged 66 years optic atrophy and horizontal nystagmus were documented and CT scan was reported as showing marked cerebellar atrophy. Now aged 74 years she cannot walk without support but otherwise manages well with the routine tasks of daily life. Chromosome studies undertaken on cultured lymphocytes showed a pericentric inversion of one number 4 chromosome with break points at p14 and q21 that is, 46,XX,inv(4)(p14q21).

The father (I1) and mother (I2) of these individuals were unrelated and allegedly without signs of cerebellar dysfunction until their deaths aged 63 and 87 years respectively. No signs of cerebellar ataxia have been noted in the surviving siblings who are aged 83 years (II1), 67 years (II6) and 64 years (II7), or in individuals III2 and III3 who are both aged 51 years; only one (III3) of these individuals carries the inversion 4 chromosome.

Precise classification of hereditary ataxia is notoriously difficult and this family is no exception in that the disease in Case II4 might reasonably be labelled as pure cerebellar ataxia whereas the description of optic atrophy in Case II5 suggests a more widespread disease process. Variation of this nature within families is well documented.[1]

Whatever its precise classification, familial late onset cerebellar ataxia usually, if not invariably, shows autosomal dominant inheritance,[2] whereas the apparent involvement in only one generation of this family is more consistent with autosomal recessive inheritance. However, the father of these affected women died at the relatively young age of 63 years and might therefore have developed the disorder had he survived longer so that autosomal dominant inheritance cannot be excluded.

Thus it may be that the pericentric inversion detected in one and possibly two affected individuals is a marker for the disorder with one of the breakpoints indicative of its genetic locus. Inevitably we can only speculate at present since the information as it stands is far from conclusive. Thus we are simply presenting our findings as an observation which may be of interest at a time when attention has focused on molecular biology as a means of mapping the disease loci of inherited neurological disorders.[3]

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MATTERS ARISING

Magnetic resonance imaging in Behçet's disease

We have read with interest the recent study by Besana *et al*[1] analysing the neurological involvement of eight patients with Behçet's disease (BD) by means of electrophysiological and MRI evaluation. They found two patients with MRI abnormalities. In one case there was a small lesion in the periventricular white matter and in the second there were two small lesions in the left periventricular white matter. They pointed out that characteristics of the observed lesions on MRI could be typical of ischaemic pattern, possibly due to vasculitis. Nevertheless, the authors did not exclude a demyelinating injury. We think that in some cases this last mechanism is the most likely one.

Recently, we have seen a 28 year old male with a history of recurrent oral and genital ulcers, arthritis and uveitis who was admitted because of occipital cephalalgia, mild lethargy, oral aphtous stomatitis and gait disturbance. Physical examination showed a slight right hemiparesis with right patella and ankle clonus. The right plantar response was extensor. There was evidence of dysmetria and ataxia. His temperature was 37·6°C. There was no clinical evidence of meningeal irritation. The optic fundi appeared normal. The cerebrospinal fluid (CSF) was clear and contained 70 cells/mm^3 of which 75% were lymphocytes; the glucose was 3·7 mmol/L (normal = 2·2–3·9 mmol/L) and the protein 60 mg per 100 ml. The basic myelin protein was normal. There was no evidence of infection caused by bacteria, fungi or neurotropic viruses in the CSF. There was no increase in the serum antibody titres against respiratory, cytomegalovirus, measles, herpes simplex, or varicella-zoster viruses. CT brain scan was normal. MRI showed a pathological area in the pons extending upwards to the cerebral peduncles and downwards to the medulla oblongata, which was clearly showed by the higher T2 intensity (fig a). Clinical symptoms promptly disappeared after treatment with steroids, and MRI two months later showed no abnormalities (fig b).

The full recovery in some patients with neuro-Behçet's disease after steroid therapy suggests a lesional mechanism different from ischaemic necrosis due to vasculitis. An immunologically mediated demyelinating process seems to be probable. In our case, the wide extension of pathological T2-weighted images and their resolution after treatment support this mechanism. Taking into account the higher value of MRI over radiograph CT examination in the diagnosis of demyelinating lesions, MRI could be the method of choice in the diagnosis of neurological involvement in BD.

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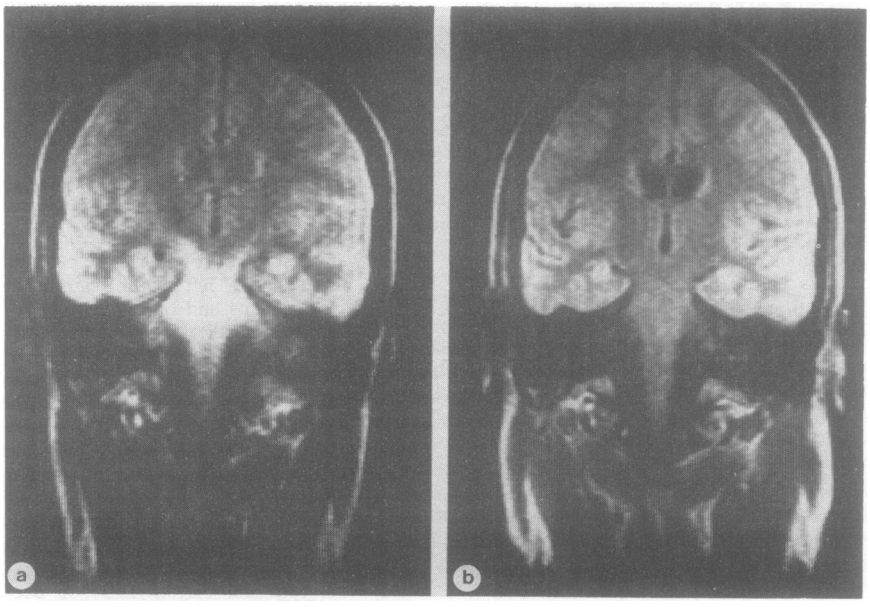

Figure (a) MRI of T2-weighted images showing a pathological area in the pons extending upwards to the cerebral peduncles and downwards to the medula oblongata. (b) MRI performed two months later showing no abnormalities.