due to either of these mechanisms will lead to a fall in the CMAP. It seems that on the scale of wasting fibres here, the relation to CMAP amplitude is linear. Thus wasting progressing from mild to moderate is associated with the same relative change in CMAP amplitude and wasting progressing from moderate to severe.

Two clinically relevant conclusions can be drawn from these findings. Firstly, an LMN lesion, causing the CMAP to fall by roughly half, may be present and yet the MRC score may not register weakness. This holds true whether the reduction in CMAP amplitude is due to axonal degeneration and hence reduction of either the number or diameter of excitabile fibres, or due to conduction block, in which the number of muscle fibres may remain normal but they are inaccessible from nerve stimulation—that is, conduction block may be present even though there is no overt weakness. Secondly, wasting on a simple four point classification better reflects the amount of excitable tissue than does the MRC score, at least in the first dorsal interosseous muscle.

Management of parkinsonism and psychotcic depression in a case of acute intermittent porphyria

Although parkinsonian syndromes are common, and the acute porphyrrias affect one in 10 000 of the population in the United Kingdom, little is known of the effects of dopamine agonist drugs on porphyrin metabolism. Many people with intermittent porphyria remain asymptomatic but all are at risk of developing an acute attack if exposed to precipitating factors, of which drugs are the most common. Although levodopa has been widely used and anticonvulsant drugs have been used safely in patients with acute porphyria, both lysuride and bromocriptine may precipitate an acute porphyric attack.5 Similarly, tricyclic antidepressants and monoamine oxidase inhibitors are contraindicated in porphyria, and there is conflicting evidence concerning the safety of selective serotonin reuptake inhibitors (SSRIs) in patients with acute intermittent porphyria (AIP).6

A 56 year old right handed woman was found to have biochemical AIP on screening prompted by the acute presentation of her brother with painful axial and anticholinergic syndromes. She had never experienced symptoms which could have been due to an acute attack of porphyria, her blood level of porphobilinogen deaminase activity was 12.7 μmol/h (normal female range 30–54 μmol/h), but her urinary porphyrins had never been significantly raised.

Eight years previously she was diagnosed as having a parkinsonian syndrome but treatment was difficult from the onset. Although she displayed levodopa responsive-ness, there was pronounced end of dose dyskinesia, and prominent peak dose dyskinesia which became remarkably sensitive to the size and frequency of the levodopa unit dose. Conventional history of exposure to dopamine receptor blocking agents and no autonomic symptoms. As her parkinsonism advanced she became increasingly depressed with hypochondriacal and paranoid delusions. She fulfilled the criteria for a severe depressive episode with psychotic symptoms (International Classification of Diseases (ICD) 10), and showed marked visual hallucinations and was noted to have a fluctuating level of consciousness.

Neurological examination disclosed increased axial tone, impaired postural reflexes, and a short, hesitant gait. She had hypomotile saccadic eye movements, orofacial dyskinesia, and slow, random alternating movements of her tongue. There was a resting tremor in all four limbs, associated with a symmetric akinetic rigid syndrome. On three hourly co-beneldopa (62.5 mg) she altered between profound bradykinesia prelevodopa (motor subsaction of the unified parkinsonism rating scale6 (UPDRS) = 44; Hoehn and Yahr stage 47), and a florid dyskinesia 30 minutes after her medication. The lowest periods of her mood coincided with the periods of choreodystonic movements but there was no evidence of pyramidal, cerebellar, or autonomic dys-function.

The patient is one of 13 siblings. Two brothers and five other sisters have biochemical AIP. One brother and biochemical AIP also fulfils UK Brain Bank criteria for Parkinson's disease (three hours after levodopa of 24 motor subsaction UPDRS = 24). A maternal family member was also diagnosed as having Parkinson's disease.

Acanthocytes were absent on a blood film. Urinary organic acid, amino acid, and copper studies were normal. No basal ganglia abnormalities were found on CT and MRI and autonomic function studies were within normal limits.

Benzhexol led to a modest improvement in her tremor. After this, periglode was slowly increased to 800 μg increments to 250 μg thrice daily. Her urinary porphyrin concentrations were monitored every 48 hours and remained unaltered. Her mobility and independence improved. Three weeks after benzhexol, she had no benefit and a modest improvement in her effect but the psychotic symptoms persisted. She responded well to six applications of electroconvulsive treat-ment (ECT) using propofol as the anaes-thetic agent. Over the subsequent eight months she did not develop any symptoms suggestive of acute porphyria and her urinary porphyrins remained within the normal range. Her psychotic depression relapsed within three months despite lepofenamine maintenance but she again responded to further ECT.

This patient had idiopathic Parkinson's disease in addition to biochemical AIP. The erratic and "brittle" response to levodopa treatment experienced by this patient may have been a result of porphyria modulating the expression of the disease or the influence of the pharmacological "window" for levodopa. Similarly, although porphyria may have been responsible for or have influenced the expression of her affective disorder, the visual hallucinosis and closing of consciousness raises the possibility of diffuse Lewy body disease, or an adverse effect of her antiparkinsonian medication.

In some case reports, ECT has improved both bradykinesia and depression in patients with idiopathic Parkinson's disease, and it was the only effective intervention in this patient's depression. Her parkinsonian syndrome improved while the periglode dose was being increased without adverse consequences, providing some evidence to support the safety of pergi- lode in the treatment of porphyria.

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A case of Machado-Joseph disease pre- senting with spastic paraparesis

Machado-Joseph disease is the most com- mon autosomal dominant spinocerebellar degeneration. The pathogenic gene responsible for the disease, which was recently identified, contains an abnormal CAG expansion in the coding sequence of a novel protein, MJJD1. The CAG expansion is inversely correlated with the age at onset, as in other triplet diseases.4,5 Four clinical sub- types of Machado-Joseph disease are well known.1 Type I patients show pronounced pyramidal signs and extrapyramidal signs such as dystonia, with a typically juvenile onset. Type II patients have cerebellar and pyramidal signs. Type III patients present with cerebellar signs and peripheral neu- ropathy, with a later onset. Type IV patients develop predominantly parkinsonism with distal atrophy. In this report, we describe a patient with Machado-Joseph disease who presented with spastic paraparesis without dystonia or ataxia.

A 38 year old woman was admitted to our hospital complaining of gait disturbance. She began to walk at the age of 1 year and 6 months. At the age of 7 years, she attended a school for mentally handicapped children. She walked on her toes, and sometimes she felt stiffness in her legs. At the age of 27 years, she became unable to go downstairs and walk without assistance.
were genetically expanded CAG repeat was used for the identification of the MJD1 gene. The inserted segment was sequenced using Sequenase (version 2.0; TOYOBO). CAG repeat lengths were counted according to original methods: the expanded CAG repeat included interrupted triplets of CAA. The present patient (II-5) and her sister (II-1) had expanded CAG repeats, whereas her asymptomatic brother (II-6) had no expansion. The repeat lengths of the patient (II-5), her symptomatic sister (II-1), and asymptomatic brother (II-6) were 74/14, 72/14, and 25/14 respectively.

The present patient, who was diagnosed genetically as having Machado-Joseph disease, manifested prominent spastic paraparesis. Because of poor information on their neurological status, the clinical subtype of those affected members (II-2 and II-2) is unclear, but another one (II-1) showed ataxia and spasticity, which may correspond to type I Machado-Joseph disease. By contrast, the proband could not be classified into any clinical subtypes. To our knowledge, few patients with Machado-Joseph disease present with isolated spastic paraparesis. The unique clinical features of this patient could not be ascribed to racial differences because phenotypic variations among cases of Japanese Machado-Joseph disease are similar to those in other countries.1-4 The proband (II-5) had mental retardation, whereas other affected members with Machado-Joseph disease (II-2, II-1, and II-2) did not show any history of mental retardation. Also, other mentally retarded siblings (II-3 and II-6) had no signs of Machado-Joseph disease. Furthermore, the onset of mental disorder (II-3 and II-5) was earlier than that of signs of Machado-Joseph disease in affected members and the course of mental disorder was not progressive, being different from that of Machado-Joseph disease. It is evident that mental handicap seen in this family is not linked with signs of Machado-Joseph disease.

If the size of the mutation is small in a patient, the age at clinical onset will be delayed and the neurological deficits less severe.4-6 Our proband (II-5) had 74 repeats, which is not a small expansion for Machado-Joseph disease. The CAG repeat length for her elder sister (II-1), who presented with ataxia and spasticity, was not longer than this. Sakai and Kawakami1 described two patients with Machado-Joseph disease with isolated spastic paraplegia, and proposed a new subtype of this condition. Although the marriage of their parents was consanguineous, the MJD1 gene locus was heterozygous on analysis. Some inherited factor might have modified the clinical features of their pedigree, but the pedigree we examined had no blood relationship. Our case and the case of Sakai and Kawakami1 might essentially fall into the same phenotype of Machado-Joseph disease.

In cases of inherited spastic tetraparesis, Machado-Joseph disease should also be suspected.

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**Playing piano in visuospatial neglect: a case study**

Patients with unilateral visuospatial neglect syndrome are typically characterised by the inability to recognise or respond to stimuli presented in contralateral space. There is much unconfirmed information on the visuospatial neglect, one of the most striking phenomena after a non-dominant hemispheric lesion.1,2 Hemineglect syndrome has been found after injury to the right parietal lobe, and less often after damage to the dominant frontal lobe.3 We describe a patient with left unilateral neglect who was able to play the piano.

A 71 year old, right handed man sustained a right hemisphere stroke and was admitted to the acute care hospital with left hemiplegia and evident left visuospatial neglect. His medical history showed hypertensive coronary artery disease and chronic gastritis, but not previous cerebrovascular events. Before the stroke, he was totally independent, working as a musician and composer. Brain CT showed an infarction of the right lateral frontal lobe (fig. 1). For the first 3 months after his stay in hospital the weakness improved in the left arm and leg, and the patient rapidly made a full recovery from the hemiplegia.

The left neglect persisted for three months after the acute event, and was confirmed by three diagnostic tasks. Firstly, when asked to identify the midpoint of 20 horizontal lines of three different lengths (10, 140, and 180 mm) drawn on a single sheet of white paper, the patient presented 33% deviation of the bisection to the right side of the centre. Secondly, when the patient was requested to copy a series of geometric items (star, cube, and house), he made an accurate depiction of the right side of the figures but neglected many details on the left side, without any awareness that the copy was incomplete. Thirdly, when a line crossing cancellation test was administered the patient crossed the targets on the right side of the paper, forgetting 72% of those on the left side. Thus, according to established clinical criteria, our patient showed characteristics of left visuospatial neglect.

The patient also underwent an extensive neuropsychological assessment to explore different cognitive domains—namely, verbal...
A case of Machado-Joseph disease presenting with spastic paraparesis.

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