Letters

Homocystinuria and dystonia

Sir: Homocystinuria is an abnormality of sulphur amino-acid metabolism characterised by a defect in cystathionine synthetase inherited as an autosomal recessive trait.¹ Major clinical manifestations are mental retardation, seizures, ectopia lentis, skeletal deformities and occlusive vascular disease.² Dystonia is not a common feature of the disease. We describe a patient with homocystinuria who developed dystonia.

This boy, the second of three siblings, was born in 1958 to healthy consanguineous Portuguese parents. There was no family history of neurological disorder. Gestation and birth were normal. The patient walked at 14 months. At the age of 9 years, he was admitted to hospital because of mental retardation and ectopia lentis with cataract. He underwent removal of dislocated lenses and homocystinuria was diagnosed. When 19, a seizure occurred and he was treated with phenobarbitone. He was well until October 1979 when mild spasmodic torticollis developed. In January 1980, the patient was admitted to another hospital with pulmonary disease. During the next 9 months, dystonia progressed. He was admitted to Ste Anne Hospital at age 22.

General examination disclosed bilateral genu valgum and arachnodactyly. There was mild scoliosis and marked dystonia of the trunk to the left. The patient walked unsteadily with shuffling irregular steps and a stiff flexed-arm posture. Muscle strength was normal. Muscle tone was slightly increased in the upper and lower limbs, bilaterally, but deep tendon reflexes were not hyperactive and plantar responses were flexor. The knee jerks did not elicit any movement but a short myotonic-like contraction in the quadriceps. There was no passive shortening reaction. All modalities of sensation were intact. Spasmodic torticollis consisted of a tonic attitude of rotation of the head to the right with clonic spasms of the left sternomastoid and right trapezius. Occasionally, rhythmic movements of the left leg were noticed. There was profound, almost unintelligible dysarthria. Eye movements were not limited but were difficult to the right with frequent eyeblinks. Vision appeared normal. There was no optic atrophy. Mental retardation was evidenced by an IQ less than 75 and defective score for Raven matrices. Routine examination of blood and urine

were normal. Homocystine in the urine was $2\,439\,\mu\text{mol/g}$ creatinine. In the cerebrospinal fluid, there was $440\,\text{mg/l}$ protein and a normal cell count. Homovanillic acid was $170\,\text{mmol/l}$ ($N: 230 \pm 65$) and 5 hydroxy-indole acetic acid was $170\,\text{mmol/l}$ ($N: 198 \pm 73$). Electromyographic study showed rhythmic 2-3 Hz contractions of the two sternomastoids and a negative polysynaptic test. There was no evidence of upper motor neuron involvement. Nerve conduction velocities were within normal range. A CT brain scan was not available because of torticollis and the risk of anaesthesia. The patient was started on a therapeutic trial by bromocriptine and pyridoxine.

To our knowledge, the only case of homocystinuria reported with a dystonic syndrome was by Hagberg *et al.*³ After chickenpox, a 15-year-old homocystinuric boy became stiff with unsteady gait and torticollis. There was no spasticity. Two years later, the patient had "an extrapyramidal syndrome of flexion dystonia and rigidity of varying severity" and hyperkinetic movements. A review of the literature reveals many reports of patients with abnormal speech, an abnormal gait or general hyper-tonus. When increased tone was not associated with increased reflexes or extensor planta responses,⁴⁻⁶ it is likely that it was attributable to an extrapyramidal rigidity. In the present case, there was mild rigidity and dystonia was the main symptom.

In a previous report⁷ we described rigidity and dystonia in a case of Leigh's disease with bilateral putaminal lesions and we emphasised the dystonic syndrome in juvenile cases of the disease. In homocystinuria, pathological changes often involve the basal ganglia.⁸⁻¹⁰ It is likely that rigidity and dystonia could develop after multiple minute infarcts in these structures.

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The effect of lumbar puncture on the electroencephalogram

Sir: We have often wondered if a reported electroencephalogram (EEG) abnormality were in whole or part a consequence of a recent lumbar puncture (LP). We can find no reference to this possibility in the literature and sought to clarify the matter by an investigation of 30 patients who as part of their general investigations required both LP and EEG.

Informed consent to EEG both before and 24 hours after LP was obtained in 30 consecutive in-patients. Before LP a 16 channel EEG was recorded which included a resting phase, 3 minutes of hyperventilation and a period of intermittent photic stimulation. In 20 patients LP was followed by 24 hours bed rest and a second EEG was performed. In 10 patients the second EEG was undertaken immediately after the LP. Seventeen women (age range 19-69 years, mean 32 years) and 13 men (age range 20-57 years, mean 34 years) were assessed. At discharge 10 patients had no neurological disease; 11 had probable or definite multiple sclerosis; three had benign intracranial hypertension; three had tension headaches and three had presenile dementia. In the 20 patients in whom bed rest was maintained for 24 hours after LP, posturally related headaches, nausea and

vomiting occurred to a variable degree in six. When bed rest was omitted, two out of 10 patients had similar symptoms. This frequency of post-LP symptoms is similar to that found by Carbaat and van Crevel *et al.*¹ EEG was within normal limits in 15 patients. There were minimal non-specific theta wave abnormalities in a further seven patients. The EEG's of the remaining eight patients showed focal or diffuse theta and/or delta wave activity. We found no apparent differences between the first and second EEG in any of the patients examined. We feel it is worth reporting that LP produced no significant effects on EEG's performed up to 24 hours thereafter.

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¹ Carbaat PAT, van Crevel H. Lumbar puncture headache: controlled study on the preventive effect of 24 hours' bed rest. *Lancet*, 1981; 2, 1133-5.

Giant aneurysm of the intracavernous carotid artery and bilateral carotid fibromuscular dysplasia

Sir: Intracranial aneurysms often occur in patients with fibromuscular dysplasia of the cephalic arterial system,¹⁻⁴ their frequency being as high as 51%.⁵ Nevertheless, the association of a giant aneurysm (largest diameter over 25 mm)⁶ of the intracavernous carotid artery and fibromuscular dysplasia, such as the case described here, has not been reported previously.

A 56-year-old woman was admitted with a history of five months of progressive horizontal diplopia, without headache or visual loss. She had no previous history of head injury or hypertension. Examination revealed only a left III and VI cranial nerve paresis; pupil reactions to light were normal, but there was a slight anisocoria (pupil diameter was 3 mm on the left and 2 mm on the right). Blood pressure was 120/70 mm Hg. Four-vessel angiography showed a typical "string of beads" appearance of both internal carotid arteries (fig A, B) and a giant aneurysm (26 × 23 mm) of the left intracavernous carotid artery (fig B). CT scan before operation showed a circular area of increased density without calcification in the left cavernous region (fig C), which rapidly and markedly enhanced as a dense and homogeneous mass (fig D) with

Figure The mid and distal cervical segments of the right (A) and left (B) internal carotid arteries exhibit alternating bands of circumferential narrowing and widening of the arterial lumen ("string of beads" appearance). (B) Giant aneurysm of the left internal carotid artery in the cavernous sinus. CT scan: preoperative pre-contrast (C) and post-contrast (D), and postoperative after contrast injection (E) (for details see text).

the same diameters as those obtained by angiography. Routine analysis, EEG, ECG and conventional radiographs were normal. Carotid artery ligation in the neck area was performed without complications. On follow-up examination four months later, there was an improvement in the diplopia and eye movements and the pupils were normal. On a repeat pre-contrast scan the aneurism was slightly hyperdense and, following infusion of contrast, there was a degree of non-homogeneous enhancement (fig E), which suggested thrombosis.^{7 8}

Intracranial aneurysms associated with fibromuscular dysplasia have the macroscopic appearance of "berry aneurysms".^{5 9 10} In fact, an association between giant intracranial aneurysm and fibromuscular dysplasia, as observed in our patient, has only been previously reported in one case.¹¹ This was an 11-year-old girl who died suddenly as the result of a sub-arachnoid haemorrhage. Postmortem examination showed a ruptured giant fusiform aneurysm of the basilar artery and fibromuscular dysplasia in the muscular arteries of all major organs; however, angiographic studies were not carried out and the histological examination of the

craniocervical arteries is not mentioned. Although the aetiology of a giant intracranial aneurysm is no different from that of other aneurysms, fibromuscular dysplasia is not mentioned as a cause.⁶ We are therefore of the opinion that when dealing with a giant intracranial aneurysm the possibility of fibromuscular dysplasia of the craniocervical arteries should be considered.

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