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. Major clinical manifestations are mental retardation, seizures, ectopia lentis, skeletal deformities and occlusive vascular disease. 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 μmol/l creatinine. In the cerebrospinal fluid, there was 440 mg/l protein and a normal cell count. Homovanillic acid was 170 mmol/l (N: 20 ± 65) and 5 hydroxyindoletol and acetic acid was 170 mmol/l (N: 198 ± 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. 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 hypertonus. When increased tone was not associated with increased reflexes or extensor plantar responses, 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 report 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. It is likely that rigidity and dystonia could develop after multiple minute infarcts in these structures.

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References

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
Homocystinuria and dystonia.

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