Correspondence

Pyridoxine responsive epilepsy: expanded pyridoxine dependency?

Sir,

Bankier *et al.* suggest¹ that the syndrome of autosomal recessive pyridoxine dependency should be expanded and that all infants with refractory epileptic seizures must be given an adequate trial of pyridoxine. On the other hand, there are many case reports of infantile spasms of diverse aetiology (Down's syndrome,² haemophilia A,³ etc) that have stopped after pyridoxine. After a dramatic response it may be difficult to withdraw and reintroduce pyridoxine to confirm its specific therapeutic effect.⁴ We present a case of childhood epilepsy, which is unequivocally pyridoxine responsive but highly atypical, to open the general question of what is the basis of this responsiveness.

A girl, now aged 3 years, had twitching episodes at age 6 days and was given phenobarbitone and antibiotics for five days. At 5 months she had one brief rigid-shaking convulsion with fever. From 7½ months she had escalating, frequent refractory seizures commonly right or left hemiclonic, long bouts of left upper limb tremor without EEG change, generalised clonic seizures and two episodes of clonic status epilepticus finally controlled by chlormethiazole. Interictal EEG's contained slow or ragged generalised spike wave. Phenobarbitone, carbamazepine, clonazepam, valproate, phenytoin, and phenytoin plus phenobarbitone were ineffective. After 3 months almost constantly spent in hospital she was given pyridoxine (100 mg a day). Seizures stopped after the first dose, but she was also on phenobarbitone 60 mg and clonazepam 0.5 mg (!). After two months without fits, pyridoxine was stopped and three days later she had a flurry of seizures. Pyridoxine was started, but after two or more months free of fits it was again stopped. Hemiclonic and generalised seizures recurred after three days. During a generalised clonic seizure 50 mg of intravenous pyridoxine stopped limb twitching in three minutes but facial twitching continued and diazepam was given. Oral pyridoxine was restarted for the third time and clonazepam tailed off. Phenobarbitone was withdrawn once her mother had successfully concluded her next pregnancy. After a stable interval on pyridoxine alone, this was stopped in hospital with continuous ambulatory EEG monitoring at the age of 2 years. Four days later seizures increased over a few hours with generalised high voltage spike and waveform. Intravenous pyridoxine was restarted for the fourth and last time and will be continued indefinitely. No seizures have been observed since, and at follow up, despite having had no therapeutic pyridoxine in infancy, she is developmentally normal, with a normal EEG.

There is a recent report⁴ of 'pyridoxine dependency' in a child with minor motor seizures from 14 months, and

Dr Sydney Gellis⁵ has referred to an unpublished case of a 3 year old with mixed seizure disorder that responded to pyridoxine. Even more so than in our patient, it is stretching classical pyridoxine dependency too far to force the inclusion of these two. The need may not be so much for recognition of the wider clinical spectrum of this condition but rather for a rethinking of the meaning of pyridoxine dependency. How does it differ from dependency on one of the classic antiepileptic drugs in mechanism and ontogeny? Well done to the Melbourne group for stimulating renewed interest in this.

References

- 1 Bankier A, Turner M, Hopkins IJ. Pyridoxine dependent seizures—a wider clinical spectrum. *Arch Dis Child* 1983;58:415-8.
- 2 Wolcott GJ, Chun RWM. Myoclonic seizures in Down's syndrome. *Dev Med Child Neurol* 1973;15:805-8.
- 3 Tajima T, *et al.* The effect of Vitamin B6 in a boy with infantile spasms. (abstract). *Brain Dev* 1983;5:62.
- 4 Krishnamoorthy KS. Pyridoxine-dependency seizure: report of a rare presentation. *Ann Neurol* 1983;13:103-4.
- 5 Gellis S. *Pediatric Notes* 1983;7:101.

J B P STEPHENSON
Fraser of Allander Unit and EEG Department,
Royal Hospital for Sick Children,
Yorkhill, Glasgow G3 8SJ

K E BYRNE,
Hawkhead Hospital,
Hawkhead Road,
Paisley PA2 7BL

Prelaparotomy diagnosis of extrahepatic biliary atresia

Sir,

We are surprised that in their search for a 'simple non-invasive test to differentiate between extrahepatic biliary atresia and other causes of conjugated hyperbilirubinaemia', Manolaki *et al.*¹ do not discuss the use of ultrasonography. A report from Gates *et al.*² evaluated 33 children with cholestasis with grayscale sonography and ¹³¹I rose bengal scintigraphy. They found that 17 of 19 children with extrahepatic cholestasis had abnormal sonograms; five had choledochal cysts, 9 had acquired biliary obstruction, while three of five patients with isolated extrahepatic biliary atresia had dilated intrahepatic ducts. All 14 patients with intrahepatic cholestasis had normal sonograms. ¹³¹I rose bengal excretion was abnormal in all cases of extrahepatic biliary atresia