

LETTERS TO THE EDITOR

Crigler-Najjar syndrome type I: management with a phototherapy crib mattress

EDITOR,—Crigler-Najjar syndrome type I is a rare autosomal recessive disorder characterised by absence of uridine diphosphate glucuronic acid transferase activity. Affected newborns develop severe unconjugated hyperbilirubinaemia and are at risk of death or permanent neurological damage from kernicterus. Phototherapy is the basis of treatment until the child grows to a more favourable size for liver transplantation, the only cure presently available.^{1 2}

A girl, born at term to non-consanguineous Italian parents, presented at 8 days of age with a total bilirubin concentration of 419 $\mu\text{mol/l}$ and no conjugated bilirubin. Investigations were normal including complete blood count, Coombs test, liver function tests, urine and blood cultures. Family history was negative for jaundice. After four days of phototherapy the baby was discharged and then readmitted two days later because of unconjugated hyperbilirubinaemia. Thyroid function tests, pyruvate kinase, and glucose-6-phosphate dehydrogenase activities were normal. A percutaneous liver biopsy specimen showed mild non-specific changes including steatosis of some hepatocytes and canalicular cholestasis. A diagnosis of Crigler-Najjar type I was based on the analysis of bile obtained by duodenal aspiration which contained unconjugated bilirubin only (Dr Roy Chowdhury, personal communication).

The baby was treated with phenobarbitone (5 mg daily) and with single and double banks of phototherapy lights (Sylvania F20T12/CW cool white and Philips TL20W/03T special blue) and discharged with a portable Ohmeda 'Bili Blanket' at 7 weeks of age. The bilirubin concentration increased by approximately 5 $\mu\text{mol/l}$ per day so additional phototherapy using two banks of billights was given twice a week for nine hours each time in a medical daycare unit. She was readmitted on three occasions for continuous double phototherapy when her bilirubin concentration rose above 250 $\mu\text{mol/l}$, associated with viral illnesses or vaccination. The phenobarbitone did not induce bilirubin conjugation.

A fan cooled phototherapy lamp was designed to replace a standard crib mattress (130 cm \times 70 cm) (figure). The 'treatment' surface consisted of a 9.5 mm thick trans-

parent acrylic lid. The lamp held ten 120 cm fluorescent tubes (Philips FT40T12/BB). The five ballasts (Philips Mark III, R2S40-TPC, sound rating A) to power the lamps were housed in a separate fan cooled enclosure, all necessary interconnections being made by a multicore cable and connector. Radiation levels at 450 nm, measured at the treatment surface, exceeded 25 $\mu\text{W/cm}^2$ (Bio-tek phototherapy radiometer, model 74345). A transparent layer of plastic air bubble sheeting (Astro Polyfoam, Toronto) was held in place on the treatment surface with a white crib sheet, lowering the light intensity to approximately 15 $\mu\text{W/cm}^2/\text{nm}$ and reducing the glare from the lamps to a more comfortable level. The baby slept with her head on a small pillow and was covered with a standard crib sheet and quilt.

The baby was treated initially with eight hours of phototherapy at night using standard 'daylight' fluorescent lamps but had significant skin tanning over a three week period. After the bed was refitted with the Philips F40T12/BB lamps the bilirubin concentration decreased and was maintained around 200–250 $\mu\text{mol/l}$ with eight hours of nightly phototherapy. No further admissions were required and the baby's development was appropriate at 16 months of age.

Side effects associated with phototherapy include headache, nausea, vertigo, painful sensations in the eyes and photophobia,³ and retinal damage.⁴ The eyes were covered during treatment with a black eye shield and later protected by her pillow. Ophthalmological examination was normal at 15 months of age.

There has been one other report of a phototherapy bed used to treat a 10 year old girl who remained well without neurological sequelae until lost to follow up at age 17 (J M Littlewood, personal communication).⁵

As the child gets older a new phototherapy mattress with more lights will be made to fit a standard twin bed frame. This way we hope to control the hyperbilirubinaemia for as long as possible while awaiting the development of new therapeutic techniques such as gene transfer⁶ thus avoiding the need for liver transplantation.

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Cystic fibrosis identified by neonatal screening: incidence, genotype, and early natural history

EDITOR,—The finding by Green *et al* of an apparent halving of the incidence of cystic fibrosis in East Anglia is a surprising one,¹ although with so few cases the confidence intervals are large. Averaging their first three and last three years shows a reduction from 5.5 per 10 000 to 3.4 per 10 000. Our own UK-wide survey covering two decades showed a relatively constant rate of 4.0 per 10 000, albeit with an apparent reduction in the last three years due to delayed ascertainment.²

The authors suggest a number of reasons why this apparent reduction in incidence might not be real. To these we would add two more. Firstly the screening test used does not always show a very high sensitivity, indeed only 68% of their cases were detected by screening alone. This then could lead to under ascertainment in the later years of the study of the same sort that we have observed. Secondly, the existence of the screening programme may have made clinicians less likely to diagnose cystic fibrosis in the mistaken belief that it would already have been diagnosed from the neonatal heel prick specimen. This also could lead to lower numbers in the later years.

We hope that the incidence of infants being born with cystic fibrosis will indeed decline as a result of early diagnosis, antenatal screening and genetic counselling, but we feel that it is premature to claim such a reduction in East Anglia on these data.

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Transient hyperphosphatasemia of infancy and failure to thrive

EDITOR,—Transient hyperphosphatasemia of infancy (THI) is well described in biochemical journals but in spite of reported incidence figures of 1.1-3.5%¹ does not appear in standard paediatric texts. THI is an apparently benign condition characterised by a marked transient increase in serum alkaline phosphatase lasting several weeks in the absence of any clinical, radiological, or biochemical evidence of bone or liver pathology.² The rise in alkaline phosphatase is quite dramatic, often exceeding 10 times the upper limit for the laboratory.

We have seen 13 further cases of THI with an age range of 5-16 months, with peak values of alkaline phosphatase ranging from 1518-14 230 IU/l, all returning to normal within nine weeks. Three quarters of our cases (10/13) presented with failure to thrive or weight loss, and 60% (8/13) had diarrhoea, which was recurrent or persistent in five cases. Previous cases have been said to have no consistent clinical features with the raised alkaline

