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 few cases, the confidence intervals are large. Averaging their first 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 1·5-4 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 believe 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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phosphatase being a chance finding. However when we reviewed the cases reported in the medical literature between 40-60% of them actually have symptoms of failure to thrive or gastrointestinal disturbance.1,2 This therefore appears to be a common clinical association. In all our cases radiology and other liver function tests were normal. Isoenzyme were measured in six cases and all but one showed either a mixture of liver and bone activity or bands between the two, typical of THI.1 There was a tendency for our cases to cluster during the summer months; this has been previously documented.4 Although the aetiology of THI remains unclear it may be the end result of different insults (infective or otherwise) in different children;2 rotavirus was found in one of our cases and adenovirus in another. The importance of the diagnosis currently is its recognition, and in the avoidance of extensive investigations. Other investigations associated with such raised alkaline phosphatase would have clinical correlates such as deranged liver function tests, or abnormal wrist or hand radiographs. A family history of familial study would detect the rare familial type of raised alkaline phosphatase.5 We suggest that the isolated finding of a massively raised alkaline phosphatase in an infant is assumed to be THI unless clinical circumstances suggest otherwise. Isoenzyme analysis and other investigations of bone and liver function tests were undertaken after eight weeks if the alkaline phosphatase has not begun to fall. Finally as between a half and three quarters of cases present with the related symptoms of diarrhoea, malabsorption or failure to thrive, we suggest that THI should no longer be considered as a pure biochemical phenomenon but rather a condition that does appear to have definite clinical associations.

**Epilepsy in children and the risk of drowning**

**Editor,—**Dr S Kemp and Sibert raised the issue of death by drowning in children with epilepsy.1 We recently had a patient aged 10 years who had had epilepsy for four years and who was on treatment with sodium valproate. He had initially presented with frequent absences and these lasted only for a second or so at a time. He had no other neurological problems. Before his death he had had no fits for the previous three weeks and had never had any generalised tonic-clonic seizures. He was found by his parents drowned in his bath having been in the bath for 5–10 minute period; he was resuscitated and was admitted to this hospital to the intensive care unit. On admission he was comatose, his pupils were dilated and dilated. He was treated by intermittent positive pressure ventilation, fluid restriction, and anticonvulsants. He developed further seizures the next morning, associated with hyperthermia and hypertension, and died 14 hours after the initial drowning episode. Postmortem examination showed presence of cerebral oedema and a few anoxic changes within his brain.

His history is a little similar to subject 1 in Kemp and Sibert’s paper, that is, a child with normal intellect and with no neurological signs who had had only a fairly minor form of epilepsy and no major motor problems. His death underscores the important reminder that Bernstein and colleagues’ work needs to be used as a guide, in such cases, to the situation in the case of drowning is a potential problem. All these cases support the view that all parents who have children, even if they have minor seizures, must either be advised of the risk of their children being in the bath or as suggested by Kemp and Sibert use a shower in an enclosed bath.

**Pressure reduction of intussusception**

**Editor,—**In this January’s issue of the journal you published a paper from this centre