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,1,2 in view of the few calendar and confidence intervals being 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 program 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.


Transient hyperphosphatasemia of infancy and failure to thrive

EDITOR.—Transient hyperphosphatasemia of infancy (THI) is well described in bio-chemical journals but in spite of reported incidence figures of 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 132 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 (103/132) presented with failure to thrive or weight loss, and 60% (81/132) 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