LETTERS TO
THE EDITOR

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

EDITOR,—Commenting on our paper, which described an apparent halving in the incidence of cystic fibrosis in East Anglia,1 Dr Coles and colleagues suggest that during the last three years of the reported study, a substantial number of patients with cystic fibrosis might have remained undiagnosed because of over reliance on the efficiency of the screening test.2 This is an implausible explanation for our observations. Such an occurrence would require either a drastic deterioration in the analytical performance of the assay, which is not supported by the results of the external or internal quality control procedures conducted throughout this period, or an abrupt change in the pathophysiology responsible for the raised blood immune reactive trypsin (IRT) concentrations in the newborn who have inherited cystic fibrosis during this later period. Furthermore, there is no evidence to suggest that East Anglia’s paediatricians have, over the years, become complacent about achieving the earliest possible diagnosis in that small percentage of cystic fibrosis patients not detected by screening. We are confident in this assertion from the number of inquiries received from every ward and clinic in the region seeking to confirm that routine neonatal IRT screening has been carried out on infants and children presenting with symptoms, in conjunction with the continuing usage of the excellent sweat testing facilities throughout the region, and more recently with the increasing use of the facility to test for the more common cystic fibrosis genotypes on DNA extracted from stored neonatal blood spots.

Secondly, all the cases were diagnosed by IRT assay. Dr Coles and colleagues refer to the figure of 68% of cases being identified by IRT assay alone, implying that the remainder were missed by clinical features which might have led to the diagnosis, in addition to an abnormal IRT assay.

There is thus no evidence to suggest that under ascertainment explains the declining incidence. We agree that it will be important to continue monitoring the incidence of cystic fibrosis in East Anglia.

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Parental participation in case conferences

EDITOR,—In child protection work the focus of professional endeavour should be the welfare of the child and as children live (usually) in families, their careers must clearly be involved in any assessment and longer term plans. So what is the function of the child protection conference? In the past information was collected concerning possible abuse and an assessment of the child and the family at least started. Now the conference has altered, it is held at the end of an investigation usually jointly planned by the police and social services and reports are focused and tend to be more preventive rather than an attempt to look at the concerns in the context of the family. The presentation may be further modified by previous strategy meetings where the professionals have met and discussed in some detail pre-planning of the conference. It then the task of the case conference to consider whether inclusion on the child protection register is to be recommended, a key worker and core group is identified. The papers will usually work with the various professionals and their attendance at the conference is a logical extension of this work. But what of the difficult cases where parents strongly deny the concerns and do not accept the need for professional involvement? As colleagues increasingly recognise child sexual abuse they will meet more denial. A 3 year old girl told her mother of the ‘games’ she plays with Daddy on her weekend access. Social services and the police investigate, the medical reveals a torn hymen and Daddy denies any wrong doing. Although the professionals feel there is substance in the story the Crown Prosecution Service do not take the case forward. At the conference are the estranged parents, the mother may have worked closely with the professionals, the father asserts it’s all an attempt by his mali-cious ex-wife to destroy him. Do we have the skills to handle all the issues presented here in the conference, attended by the parents’ two solicitors who may advise their client not to talk?

It may be argued that the above case is unusual: this depends upon the geographical area and the willingness of professionals to engage in complex work. Each is different, if the girl in the scenario above was 13 years would she accept her abuser, who may be her abuser, to have details of her medical report (and her teacher, etc...).

Working Together does not expect that professionals can always work with parents to achieve child protection.1 Cooperation may develop but true cooperative ‘partnership’ is not achieved instantly. A working relationship with parents may be a better description of the longer term expectation.

In recent years in the journal Sheeby Skeffington is right to suggest caution and keep the focus on the child’s needs and welfare; Hutchinson is overly optimistic of the potential that might be gained from an increasing knowledge of the dynamics of abusive relationships and behaviours.3

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Sudden infant death syndrome

EDITOR,—A high degree of heat production has been identified as a risk factor in the causation of sudden infant death syndrome (SIDS) by several studies.1 It has been suggested that one reason why sleeping prone may increase the risk of SIDS is that it restricts the loss of heat from the infant to the environment.1 Concern has thus focused on factors that may increase heat production, such as febrile illness, or limit heat loss, such as excessive thermal insulation of clothing or high environmental


Differences between breast fed and formula fed infants in metabolic rate, which is directly related to heat production, have already been reported, and we have recently found that the difference in metabolic rate is significantly higher in formula fed compared with breast fed infants at age 12 weeks. Furthermore, total daily energy expenditure (TDEE) measured by isotopic methods, has also been shown to be significantly greater in formula fed than breast fed infants. In a longitudinal study in the first year of life, TDEE was found to be significantly greater in formula fed infants at 6 weeks and 12 weeks but to be similar between the diet groups at 6 months and 9 months (P S Davies, unpublished data). This pattern correlates with the reported distribution of SIDS by age.4

A number of studies of SIDS have reported higher rates of formula feeding in cases compared with controls. Because SIDS does occur in breast fed infants, formula feeds have not been considered a major risk factor. Some authors have suggested that the relationship between formula feeding and SIDS incidence is an artefact of the relationship between SIDS and social class. However, it is more likely that the reverse is true, and that variables such as family size, social class, maternal age and interpregnancy gap are related to SIDS incidence because of their effect on aspects of infant care, of which formula feeding might be one.

If this hypothesis is correct, one explanation might be that the amount of energy in contemporary formula is high in comparison to the mean energy content of breast milk. This view has been increasingly supported from recent studies of nutrition, growth, morbidity, and development of breast fed and formula fed infants. These findings suggest the need for a thorough review of energy requirements in infancy and especially the adequacy of the energy density of infant formulas.

The physician’s hands and early detection of neuroblastoma

EDITOR.—The data from some developed countries show that within the past 25 years, the five year survival rates in neuroblastoma have increased two-fold from the initial 25%. The outlook for patients presenting over 1 year of age with stage IV remains dismal (20% five year survival). The drop in mortality rates is related to more frequent detection of this tumour in children under 1 year of age and especially in those with obvious organomegaly, especially below 1 year of age, during obligatory frequent check up visits to which almost 100% of parents faithfully report, allowed the incidental detection of neuroblastoma in every sixth patient with this tumour. Out of 65 children at stage I and 60 patients at stage II, incidental detection of neuroblastoma occurred, respectively, in almost every second and third patient with this disease. This may suggest that a systematic approach, a greater awareness of the relatively high incidence of this tumour (the most common solid tumour in children), and the need for good abdominal examination may increase the detection of children with neuroblastoma before the onset of symptoms.6 The shift to diagnosis at earlier ages and stages may result from more frequent chest radiographs and use of ultrasound, in the diagnostic workup of neuroblastoma. In a study of 293 infants suspected of neuroblastoma on the basis of urinay screening, physical examinations revealed a tumour in more than one half of the patients. A careful abdominal examination is of great importance in neuroblastoma (the primary tumour is in 75-95% of the cases located within the abdomen).

The hands and eyes of a physician have always been and continue to be the most important tools in detecting diseases. A physician may notice slight, rare symptoms of low stages, and the totality of a new neuroblastoma, such as Homer’s syndrome and associated heterochromia, the watery diar- rhoea syndrome and the dancing eyes and dancing feet syndrome. Never in his entire life is a human being subjected to medical examinations as often as in early childhood. This is dictated by obligatory periodic check ups, physical examinations before vaccinations, and a mother’s loving care, prompting her to seek medical assistance any time she sees a sign or symptom which makes her anxious. The skilful hands of the physician, examining the abdomen on these occasions, can contribute to early detection of the low stages of neuroblastoma and to the decrease of mortality rates in this tumour. It is possible to perform this type of screening throughout the world.


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