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 and the remaining cases with 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 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 written rather than 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 depth how they 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 more often. A 3 year old girl told her mother of the ‘games’ she played 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 may 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 mili-pate, the ex-wife, to control 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 still have been recorded as 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, the journal Sheety Skefington is right to suggest caution and keep the focus on the child’s needs and welfare; Hutchinson is overly optimistic of parenting behaviour given our 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