

## Diagnosis of Cystic Fibrosis

Not long ago a diagnosis of cystic fibrosis carried with it in most cases the probability of death within a very few years. The improvement in life expectancy now is largely the result of active treatment and the introduction of more effective antibiotics. There is also strong evidence<sup>1-3</sup> that early diagnosis has improved the chances of survival.

Early memories die hard, and for many a doctor the term cystic fibrosis recalls a wasted, pot-bellied infant with grossly clubbed fingers seen during a ward round as a medical student. The newborn baby with cystic fibrosis looks perfectly normal; so may the toddler and even the older child. It is only after recurrent respiratory infection or prolonged malabsorption that the characteristic appearance is seen. The triad of failure to thrive, an exceptionally good appetite, and offensive, rather loose stools should be sufficient to require laboratory tests for cystic fibrosis to be carried out. The story of persistently abnormal stools, frequent or bulky, greasy, pale, and offensive even when the child is well nourished, should lead the doctor to consider cystic fibrosis as the diagnosis, especially if the child's appetite is said to be good. A cough should be considered a late symptom, though a persistent catarrhal cough or "frog in the throat" may be present from the first weeks of life in babies exposed early to infection. Some infants, however, first present on account of an acute lower respiratory infection, while others are thought to have pertussis because of the paroxysmal cough. In either case a carefully taken history will usually, though not invariably, elicit some evidence of malabsorption. An odd but characteristic comment is that of the mother who notes that her baby tastes salty when kissed or has a line of salt on the forehead after sweating.

Clinical findings, therefore, at the time when diagnosis should be made are remarkable chiefly for their absence. The submaxillary salivary glands may be enlarged,<sup>4</sup> but not as a rule in babies, and they are never easy to feel. The abdomen may be distended and the liver is likely to be more easily palpable than would be expected for the child's age. Signs of early pulmonary disease are shown by some degree of forward bowing of the sternum and perhaps indrawing of the lower ribs, with a mild degree of finger clubbing consisting only of loss of the nail-bed angle.

Nasal polyps are rare in childhood but occur in about 5-10% of children with cystic fibrosis,<sup>5</sup> so the presence of one in a child with pulmonary symptoms should suggest this disease as the cause. Prolapse of the rectum on the other

hand is common enough in infancy, but is even more frequent in the untreated toddler with cystic fibrosis,<sup>6</sup> and so this condition should always be excluded before attempting any form of treatment, particularly surgical.

A persistent or paroxysmal cough, recurrent attacks of pneumonia, yellow or greenish purulent sputum, a voracious appetite, and frequent large, greasy, and foul stools are relatively late symptoms. In the differential diagnosis bronchiectasis may need consideration, rare though it now is in Britain. Wheezing and bronchospasm are uncommon in cystic fibrosis, but may occur, and asthma itself occasionally occurs in association with it. Coeliac disease enters into the differential diagnosis and may be associated with cystic fibrosis. Diabetes mellitus is a complication in about 5% of cases, but it is unlikely that a case of cystic fibrosis would present primarily with diabetes.

In about 10% of cases cystic fibrosis presents at birth with acute intestinal obstruction as the result of meconium ileus and sometimes meconium peritonitis and ileal atresia.<sup>7</sup> The plain x-ray of the abdomen commonly shows, in addition to the usual fluid levels and distended loops of bowel of acute obstruction, an opaque shadow in the pelvis and lower abdomen, with a characteristic bubbly appearance due to air in the meconium-filled intestine. The appearance at laparotomy of the ileum distended with dark green meconium and the narrow string-like colon is unmistakable.

The normal appearance of the infant with cystic fibrosis will always cause difficulties in diagnosis and makes all the more necessary a reliable screening test. The measurement of sodium concentration in parotid (or buccal) saliva<sup>8</sup> by means of a sodium electrode has been shown to be reliable between the ages of 4 months and 5 years. Recent work suggests that a simple test for the albumin content of meconium in the newborn<sup>9</sup> may provide a useful screening test which is easily carried out. The chloride skin electrode has its advocates.<sup>10</sup> Unfortunately it has to be admitted that there is as yet no universally applicable, cheap, harmless, and rapidly executed method of screening newborn babies for the disease.

Confirmatory laboratory diagnosis in all cases over the age of a few days in whom there is the least suspicion of cystic fibrosis depends on electrophoretic stimulation of sweating and estimation of the sodium content. At least 100 mg of sweat is required for accurate estimation by flame photometry and estimates on smaller amounts should be discarded. A level of below 50 mEq per litre excludes the diagnosis and

of 70 mEq or over is diagnostic in children. The concentration of salt in the sweat rises with age and exertion. Concentrations of sodium between 50 and 70 mEq per litre in young children are inconclusive, and the test must be repeated.

- <sup>1</sup> Mantle, D. J., and Norman, A. P., *British Medical Journal*, 1966, 2, 1238.
- <sup>2</sup> George, L., and Norman, A. P., *Archives of Disease in Childhood*, 1971, 46, 139.
- <sup>3</sup> Warwick, W., and Monson, S., in *Cystic Fibrosis*, ed. E. Rossi and E. Stoll, p. 365. Basel, Karger, 1967.
- <sup>4</sup> Barbero, G. J., and Sibinga, M. S., *Pediatrics*, 1962, 29, 788.
- <sup>5</sup> Shwachman, H., Kulczycki, L. L., and Mueller, H. L., *American Journal of Diseases of Children*, 1961, 102, 768.
- <sup>6</sup> Kulczycki, L. L., and Shwachman, H., *New England Journal of Medicine*, 1958, 259, 409.
- <sup>7</sup> Holsclaw, D. S., Eckstein, H. B., and Nixon, H. H., *American Journal of Diseases of Children*, 1965, 109, 101.
- <sup>8</sup> Lawson, D., Sagers, B. A., and Chapman, M. J., *Archives of Diseases in Childhood*, 1967, 42, 689.
- <sup>9</sup> Green, M. N., and Shwachman, H., *Pediatrics*, 1968, 41, 989.
- <sup>10</sup> Kopito, L., and Shwachman, H., *Pediatrics*, 1969, 43, 794.

## Hospital Staffing

If volume of paper was a measure of success then a satisfactory structure for hospital medical staffing would have been established long ago. Regrettably, it is not, and yet, another report has now emerged on this topic the outcome of discussions between the Joint Consultants Committee and the Government which started in 1967 after publication of the B.M.A.'s "Charter"<sup>1</sup> for hospital doctors. This report, the second on the progress of the discussions is printed with its appendix containing "certain reservations expressed by the profession" in the *Supplement* at p. 119 and it marks a step towards resolving the refractory problem of hospital staffing. But there is still a long way to go.

The difficulties of balancing the service requirements of hospitals against doctors' training and off-duty needs, combined with the uncertainty of planning a career in the hospital service have all contributed to the widely publicized unrest in recent years among senior and junior medical staff alike. The N.H.S. inherited an unequal distribution of resources and doctors in the hospital sector—among different regions, between teaching and non-teaching hospitals, and among the different specialties. Such maldistribution is not exclusive to medicine or to Britain.<sup>2</sup> Despite progress towards a more even deployment of specialists, the N.H.S. still faces a situation where there is no shortage of applicants for a consultancy in general surgery in a London teaching hospital, but a vacant consultant post in a less popular specialty in a northern industrial town will attract few, if any, suitable candidates. This situation, naturally, has repercussions among junior staff thus creating a vicious circle.

A high standard of treatment for patients everywhere, together with proper training, and a rewarding professional career for doctors is what the hospital service needs. How can it be obtained? Certainly not by direction of doctors—one solution rightly rejected by the Central Committee for Hospital Medical Services. While more money might help to redistribute doctors, unattractive areas cannot be made attractive overnight nor can less popular disciplines suddenly be acclaimed as medical meccas. Yet a practical start must be made somewhere and the proposals in the latest report promise this.

A major frustration of some hospital doctors has been the feeling that they play a small part in deciding their own

destinies, that decisions affecting their professional future seem to be taken in remote and, in their view, unrepresentative committees. The new scheme will provide an opportunity for medical staff at all levels and in all areas to take part in the monitoring and planning of future staffing. It is an opportunity which must be taken, for even though the Central Manpower Committee and its network of regional committees will have an advisory role and will not supplant existing executive procedures or professional negotiating machinery, their views are bound to be heeded. The central committee, containing representatives from the Department as well as the professions, will replace the present Advisory Committee on Consultant Establishments for England and Wales though its functions will be wider. The Scottish A.C.C.E. is to continue for the present. The regional committees, with an entirely professional membership, are a new idea. Hospital staff should ensure their viability so that comprehensive information and advice from the profession (the proposals are for a general practitioner and a community physician to be included in each committee) on local as well as national patterns of hospital staffing will be readily available to the Department of Health and its agencies.

The proposed increase of consultant staff by 4% per annum with the junior staff growth rate being restricted to 2½% should improve career prospects in hospital medicine over the next 10 years (the period of projection), by correcting the present imbalance between the number of doctors in training and consultant vacancies. With the steadily rising output from medical schools, this target should be achieved without any fall in standards. Nevertheless, the C.M.C. will face two big imponderables in its forecasting. Firstly, what will be the future rate of medical immigration and how can the intentions of overseas doctors in the hospital service be anticipated? Secondly, how many British doctors will emigrate? Despite the hazards inherent in making these forecasts the committee should certainly be able to present a clearer picture of long-term prospects in the various specialties for aspiring consultants. Eventually, this may even reduce the overabundance of potential surgeons and general physicians to the advantage of other disciplines.

The fact remains, though, that no amount of monitoring or planning will guarantee that doctors will go where they are needed. Removal of the wider social disincentives of unattractive areas is outside the scope of the N.H.S., so the profession and the Department of Health must press on urgently with discussions on other ways of overcoming the maldistribution. Short-term incentives such as adjustments in pay are one possibility. In the longer term, other incentives will be needed—for example, improved hospital buildings and equipment, good postgraduate facilities, and better accommodation. There are other major issues, too, which influence the recruitment, balance, and distribution of staff, such as the type of contract for junior doctors, pay—including distinction awards—the place of the general practitioner and the part-time doctor in hospital, and the extent of private practice. These matters are the responsibility of the profession's representatives, who will also agree with the Department "strategic objectives" for the C.M.C. If its promise is fulfilled the new scheme should certainly facilitate discussions on resolving all these interrelated problems which affect hospital staffing and thus help to ensure a high standard of patient care together with a satisfying career for the doctors providing it.

<sup>1</sup> *British Medical Journal Supplement*, 1967, 2, 93.

<sup>2</sup> *British Medical Journal Supplement*, 1971, 3, 116.