Bronchiectasis and oligospermia: two families

Sir,—We were interested to read the paper by Dr Pamela B Davis and colleagues describing the association of oligospermia and bronchiectasis (May 1985;40:376-9) in two families, which they suggest is a new syndrome. It is, however, open to question whether the condition described really is new or, as seems possible, details two further families with primary ciliary dyskinesia (PCD). The clinical histories, including neonatal pneumonia, lifelong productive cough, and recurrent otitis media requiring repeated gromet insertion, against a background of general good health and reasonable lung function, are typical of primary ciliary dyskinesia. It is unfortunate that no measurements of nasal or tracheobronchial clearance were undertaken, because severely impaired clearance would have been suggestive of the diagnosis and a normal result would have effectively excluded it. It is stated in the paper that ciliary beat pattern was similar to that of normal controls but it is regrettable that there were no quantitative measurements of ciliary activity such as ciliary beat frequency or percentage immotility, or both, because mild degrees of ciliary dyskinesia may be difficult to identify in vitro, even by experienced observers.

There are now several reports of ciliary dyskinesia occurring with normal ultrastructural appearances.1-3 We have recently studied a family consisting of the index case (a 23 year old woman), her 21 year old brother, and her 51 year old mother. All had normal cardiac situs, recurrent upper or lower respiratory tract infections (or both), reduced nasal ciliary beat frequency (4.5, 5.8, and 0 Hz respectively; normal range 11–16 Hz), and normal ultrastructure. Although the commonest ultrastructural appearances in patients with primary ciliary dyskinesia are those of dynein arm deficiency, it is apparent that abnormalities of ciliary motility for which there is no abnormal morphological counterpart can also occur. These abnormalities can be transmitted genetically, as our cases show. Reliance on ultrastructural appearances clearly has many pitfalls and its seems likely that more cases of primary ciliary dyskinesia with normal ultrastructure will be recognised when in vitro observation and objective measurement of ciliary activity become more widespread.

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**This letter was sent to the authors, and Dr Davis replies below.

Sir,—Dr Greenstone and colleagues suggest that the patients we reported have a variant of “primary ciliary dyskinesia” with normal ultrastructure and that this diagnosis could have been made by measurements of mucociliary clearance. We agree primary ciliary dyskinesia can occur without visibly abnormal morphology; indeed, we referenced a report of a child with Kartagener’s triad and normal ciliary ultrastructure. However, the causes of abnormal mucociliary clearance are legion, and an abnormal result could not confirm the diagnosis. Our patients differ from the usual patients with primary ciliary dyskinesia in that the men have oligospermia. Our patients have a clinical syndrome which resembles primary ciliary dyskinesia in some respects but Young’s syndrome in others, and is identical to neither. When the details of the disordered biochemistry are known, the aetiological relationships among these disorders will be clarified.

The family mentioned by Dr Greenstone and colleagues is intriguing. Parent-child transmission is quite rare in uncommon autosomal recessive genetic disorders and, to our knowledge, autosomal dominant forms of PCD have not yet been reported. Analysis of semen from the 21 year old brother would be helpful in confirming that ciliary dyskinesia occurs outside areas of inflammation. Detailed evaluation of this family might prove useful, for until the biochemistry of this group of disorders is known, we are reduced to careful clinical descriptions in an attempt to understand their pathogenesis.

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Notices

British Thoracic Society: meetings in 1986
18–20 June Cheltenham (NB abstracts required by 27 March 1986)
11–12 December Kensington Town Hall, London

Scadding-Morrison Davies Joint Fellowship in Respiratory Medicine
The Scadding-Morrison Davies Joint Fellowship in Respiratory Medicine is available to medical graduates practising in the United Kingdom, including consultants, irrespective of the number of years in that grade. A sum of up to £10 000 will be awarded to support travel and subsistence as needed, to attend medical centres in the UK, Europe, or elsewhere in the world for studies related to respiratory medicine. Applicants should submit a curriculum vitae together with a detailed account of the duration and nature of the work, and the centres to be visited, confirming that these have agreed to provide the facilities for the work. Applications should be sent before 26 May 1986 to the secretary, Dr KM Citron, Brompton Hospital, Fulham Road, London SW3 6HP.