

Familial nasal acilia syndrome

R Soferman, Z Ne'man, M Livne, A Avital, Z Spirer

Abstract

Mucociliary dysfunction can be caused by the absence of cilia or by abnormalities in their ultrastructure. A family in which some members were suffering from respiratory morbidity due to lack of cilia is described.

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The familial nasal acilia syndrome is very rare.¹ We describe four children from one family in which the lack of cilia caused respiratory morbidity.

Case report

The index patient was a full term white girl (birth weight 2650 g) who was born without complications (Apgar score 10) after an

uneventful pregnancy of 39 weeks. She had respiratory distress at 48 hours, and a chest radiograph revealed an atelectasis of the right upper lobe. The oxygen saturation was 84%, oxygen tension 6.65 kPa, carbon dioxide tension 6.00 kPa, and pH 7.39. She was treated with 40% oxygen for 24 hours, with antibiotics (ampicillin and gentamycin), and postural drainage. There was a gradual improvement and at the time of discharge from the newborn department, at the age of 12 days, there were no pathological findings in the chest radiograph. At the age of 17 days she was hospitalised because of dyspnoea, and physical examination disclosed retractions of the intercostal spaces and diffuse rhonchi and rales throughout the lungs. No abnormality was detected on the chest radiograph. Her condition improved spontaneously and she was discharged. Three weeks later, she was hospitalised again because of dyspnoea, appearance of cutis marmorata, and high fever. This time the chest radiograph demonstrated infiltration in the right lower lobe. The blood culture was negative, and the white cell count was $15.0 \times 10^9/l$. Both her clinical condition and chest radiograph improved after erythromycin treatment. Repeated sweat tests demonstrated normal chloride concentrations (30-37 mmol/l), and repeated nasal culture revealed normal flora. No immunoglobulin assay was obtained because of her young age. Her height and weight centiles were 25% and 5% respectively. Ultrastructural studies of the respiratory epithelium² were performed on specimens obtained by a double nasal brush biopsy from the inferior turbinate when the patient was clinically well.³ In each sample a minimum number of 100 epithelial cells were randomly selected and analysed and found not to be ciliated.

The same procedure of nasal brush biopsy was repeated three months later and the same picture of unciliated epithelial cells was demonstrated (fig 1). It is noteworthy that during this period of time she continued to suffer from recurrent upper and lower respiratory tract infections.

The index child's parents were second degree relatives (fig 2). Her sister and two cousins (siblings), suffered from recurrent upper and lower respiratory tract infections including chronic sinusitis. One of them had recurrent episodes of lobar atelectasis and all the three had evidence of lung bronchiectasis. The girl cousin had demonstrated a pathological saccharine test. They were diagnosed at different laboratories as suffering from congenital nasal acilia syndrome.

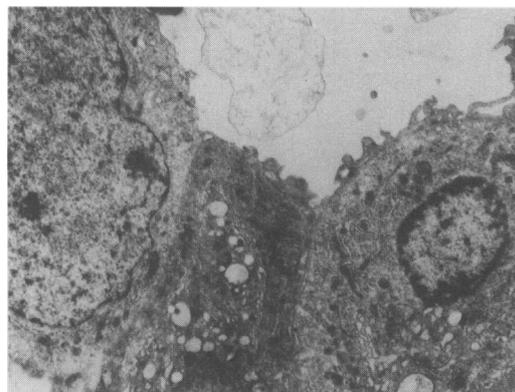


Figure 1 Electron micrograph of nasal aciliated epithelial cells.

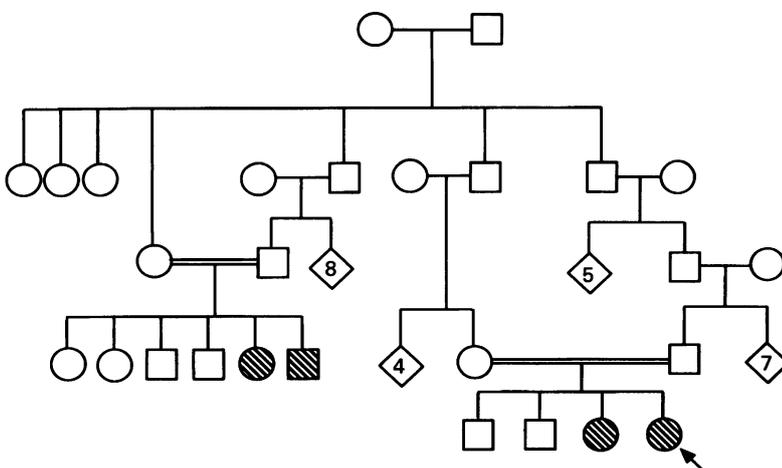


Figure 2 Family pedigree.

Dana Children's Hospital, Sourasky Medical Center, Tel-Aviv
R Soferman
Z Spirer

Department of Pathology, Hadassah Mount Scopus, Jerusalem
Z Ne'man

Pediatric Allergy and Pulmonology Clinic, Assaf Harofeh Medical Center
M Livne

Institute of Pulmonology, Kiryat Hadassah, Hebrew University, Jerusalem, Israel
A Avital

Correspondence to:
Dr R Soferman,
Pediatric-Pulmonology
Clinic, Dana Children's
Hospital,
Sourasky Medical Center,
Tel-Aviv, 64239, Israel.

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Discussion

The dyskinetic cilia syndrome, which predisposes the respiratory tract to many insults, is considered to be an autosomal recessive inherited disease.⁴ A wide spectrum of ciliary defects and concomitant mucociliary dysfunction has been documented. Various congenital and environmental factors have been shown to influence the process of ciliogenesis.⁵ Children who lack cilia are functionally similar to those with dyskinetic cilia syndrome. We believe that the deficiency of nasal cilia in this patient is a primary defect and not a transient denudation of the cilia that may occur after infection or as a result of some other insult, because the first episode of lung atelectasis occurred at the age of 48 hours, and the nasal ciliary brush biopsy specimen was obtained at a period of time when there was no evidence of upper respiratory tract infection.

There are some reported cases of nasal acilia syndrome,^{6,7} and a description of a family in which the father and all three children

appeared to have this syndrome.¹ This present case report emphasises the importance of the nasal ciliary brush biopsy in evaluating a patient suffering from respiratory morbidity.

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