Use of Articles in the Pachyonychia Congenita Bibliography

The articles in the PC Bibliography may be restricted by copyright laws. These have been made available to you by PC Project for the exclusive use in teaching, scholarship or research regarding Pachyonychia Congenita.

To the best of our understanding, in supplying this material to you we have followed the guidelines of Sec 107 regarding fair use of copyright materials. That section reads as follows:

Sec. 107. - Limitations on exclusive rights: Fair use
Notwithstanding the provisions of sections 106 and 106A, the fair use of a copyrighted work, including such use by reproduction in copies or phonorecords or by any other means specified by that section, for purposes such as criticism, comment, news reporting, teaching (including multiple copies for classroom use), scholarship, or research, is not an infringement of copyright. In determining whether the use made of a work in any particular case is a fair use the factors to be considered shall include - (1) the purpose and character of the use, including whether such use is of a commercial nature or is for nonprofit educational purposes; (2) the nature of the copyrighted work; (3) the amount and substantiality of the portion used in relation to the copyrighted work as a whole; and (4) the effect of the use upon the potential market for or value of the copyrighted work. The fact that a work is unpublished shall not itself bar a finding of fair use if such finding is made upon consideration of all the above factors.

We hope that making available the relevant information on Pachyonychia Congenita will be a means of furthering research to find effective therapies and a cure for PC.
1. Introduction

In the first decade case, a 12-month-old girl with pachyonychia congenita and Lauenroth-Lewandowsky syndrome, laryngeal obstruction was noted. The patient's airway due to leukoedema and successful microlaryngeal management of laryngeal obstruction was described. This report confirms the presence of laryngeal involvement in children with pachyonychia congenita. The laryngeal involvement can present as a laryngomalacia or dysplasia. Laryngomalacia and vomiting

Abstract

Received 5 April 1994; revision received 7 June 1994; accepted 16 June 1994

Department of Pathology, University of Ulm, D-89070 Ulm, FRG
Department of Otorhinolaryngology, University of Ulm, D-89070 Ulm, FRG
Department of Pediatrics, University of Ulm, Ulm, FRG

S.A. Wydyrlewicz, H. Tenders, W. Prise', W. Molin, W.M. Teillet

Diagnosis and Management of Laryngeal Obstruction in Childhood Pachyonychia Congenita

International Journal of Pediatric Otorhinolaryngology
Laryngeal obstruction has recently been recognized as another important finding in pachyonychia congenita. Though less frequent, it bears the risk of life-threatening respiratory distress especially in infants and children (1, 3-7). This report focuses on the diagnosis of the disease and surgical management of laryngeal obstruction in a girl, 19 months of age, with pachyonychia congenita.

### 2. Clinical report

The girl was the first and only child of nonconsanguineous caucasion parents. There was no family history for pachyonychia congenita. After an uneventful pregnancy, she was born with normal weight and length. During the first weeks of life, her nails began to discolor and thicken. Hoarseness of the voice was reported to be chronic. Planter callouses had occurred since she started walking at 12 months. Inspiratory stridor occurred around the age of 15 months and had been progressive. In case of infections of the upper respiratory tract, hoarseness and inspiratory stridor increased markedly and led to respiratory distress.

At the age of 19 months she was referred to our department for the evaluation of multiple hyperkeratotic lesions and inspiratory stridor. She had a hoarse, deep voice. A slight, constant inspiratory stridor without respiratory distress was noted.
Fig. 3. Microscopic view of the left vocal cord showing the larger lesion on the left vocal cord.

Fig. 2. Direct laryngoscopic view of the glottis with exophytic lesion in the posterior commissure and on both vocal cords.
Histological examination of the laryngeal lesions (Fig. 5) revealed scattered small capillaries; no inflammatory cellular infiltrate could be noted. Keratinocytes were present in the neighborhood of the papillae. The papillae contained squamous epithelium with superficial parakeratotic keratinisation. Some vascularised subepithelial inflammatory cells were noted.

3. Histopathological Findings

operation.

sided. No sinister had occurred during a wire laryngoscopy injection 4 months after.

Clinical controls 3 and 6 months later revealed that only a slight hoarseness had per-

posteriorly. The patient's side of the vocal cord was excised (Figs. 4).

cases, the surgical site was clear of the larynx vocal cord. Using microscopic laryn-

in the left vocal cord were most prominent (Fig. 3). The lesions were symmetrical and involved the vocal cords and the posterior commissure (Fig. 2). The lesion was most marked in the vocal cords and the posterior commissure. Biopsy performed under general anaesthesia. Laryngoscopic examination revealed keratosis with

Hysteroscopy and instillation of the larynx lesion were the indications for a suspension.

Hyperaemia and irritation of the vocal cords were evident at the pressure points.

multiple reefs were evident at the pressure points.

The larynx persisted at night. All reeds of the hands and feet were opaque; thickened

Figs. 4. Microscopic view of the laryngeal lesions after excision and cauterisation of the left vocal cord.
Figure 5. (A) Stratified squamous epithelium with oblique sections of dermal papillae (hematoxylin-eosin).

Figure 6. Magnification × 330.

Dermal papillae without inflammation (arrow). Dermal papillae with suprabasal parakeratotic keratinisation and cytoplasmic vacuolation of some melanocytes (arrows). Higher magnification ×130. (B) Figure 6a shows vacuolation of the area indicated by arrow in (A).
4. Discussion

Except for a mild inflammatory infiltrate, the pattern of histopathological findings in our case was in accordance with the microscopic appearance of laryngeal lesions present in this entity [1].

In conclusion, treatment of pachyonychia congenita is completely symptomatic, aiming at ameliorating the discomfort produced by the hyperkeratotic skin and nail lesions. However, in infants and children with pachyonychia congenita, hoarseness and stridor should alert the physician to obstructive involvement of the larynx. In these cases, immediate laryngoscopy is indicated. Microsurgical removal of leukoplakia can prevent life-threatening respiratory tract obstruction. Consistent clinical monitoring is necessary, because recurrence of the leukoplakia lesions might occur [1].

References