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 The fact that a work is unpublished shall not itself bar a finding of fair use if such finding is made upon consideration of all the above factors.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.
Case Reports

Pachyonychia congenita with laryngeal involvement

Bruce Benjamin ¹, David S. Parsons ² and H.F. Molloy ³

¹ Department of Laryngology, Sydney University, Sydney (Australia); and Departments of ² Pediatric Otolaryngology and ³ Dermatology, Royal Alexandra Hospital for Children, Sydney (Australia)

(Received 23 January 1987)
(Received 22 March 1987)

Key words: Pachyonychia; Larynx

Summary

Pachyonychia congenita (Jadassohn–Lewandowsky Syndrome) is a rare autosomal dominant disorder characterized by nail dystrophy, hyperkeratosis of the palms and soles, leukoplakia of the mucosa of the upper respiratory tract and anus, follicular keratoses especially about the knees and elbows, and palmar and plantar hyperhidrosis. We present a patient with pachyonychia congenita and an exophytic lesion in the larynx at the posterior commissure. He is the youngest of 4 family members with this disorder covering 3 generations. Each of the 4 patients also exhibited both oral leukoplakia compatible with the Jadassohn–Lewandowsky syndrome (Ikonograph Dermatol. Lab., 1 (1906) p. 29), and subcutaneous cysts of the face and scalp as described by Jackson and Lawler (Ann. Eugenics (1951) 142.

Introduction:

Pachyonychia congenita was first described in the literature by Jadassohn and Lewandowsky [5] in 1906. Since that time over one hundred cases have been reported but only a few with laryngeal involvement [1,2,4].

Kumer and Loos [6] classified the disorder into 3 types. Type I presented with symmetric hyperkeratoses of the hands and feet and follicular keratoses of the body. Type II combined these findings with leukoplakia of mucous membranes; patients with laryngeal involvement should be classified with this group. Type III patients have corneal dyskeratosis in addition to the manifestations of type I.

Correspondence: B. Benjamin, 229 Macquarie Street, Sydney 2000, Australia.

0165-5876/87/$03.50 © 1987 Elsevier Science Publishers B.V. (Biomedical Division)
A single autosomal dominant gene with variable penetrance appears to be responsible for the disorder [8]. Family members' manifestations may vary but the basic Kumer and Loos classification appear to be consistent throughout the family line.

Diagnosis with certainty can often be made within the first 3 months of life especially in those patients with a positive family history for pachyonychia congenita. The earliest presentation of the disorder may be with any number of clinical features. The nails are usually reported to be normal at birth but quickly become abnormal in the first 1–12 weeks. Teeth have been present at birth in many patients. Oral leukoplaikia has been described as the first visible abnormality in many neonates, but may be misdiagnosed as thrush [8].

As the patient grows, skin involvement may be noticeable as keratotic patches on the palms and soles, and areas of the skin may appear to be thickened. Follicular hyperkeratosis with well developed keratin plugs may occur on the extensor surfaces around the large joints. There may be bullae, which are usually associated with hyperhidrosis of the palms and soles. Bullae may develop over calluses and may be painful. Hair manifestations have been reported from dry and kinky, to thick and exuberant, to thin with alopecia. Nodular epithelial cysts of the scalp, face, and neck are not uncommon.

Oral leukoplaikia is not found in all patients but for those who have it leukoplaikia may be present at birth. The most common site is along the line of dental occlusion. These lesions usually appear as whitish plaques frequently involving the dorsum of the tongue. The lateral borders of the tongue may appear especially thickened. Other mucous membranes that have demonstrated leukoplaikia include nasal and perianal mucosa.

The laryngeal lesions described have all involved the posterior commissure [1,2,4]. These have usually been described as white in color. In the masses which were biopsied, the histopathology has revealed thickening of the epithelium with acanthosis and parakeratosis, extensive vacuolization as seen in white sponge nevus, and without evidence of dyskeratosis. Hoarseness, the clinical sign of laryngeal involvement, has been reported in less than 10% of patients with this syndrome.

Case presentation

A 15-year-old male with previously diagnosed pachyonychia congenita, presented with a chronic history of a gruff voice. The mother reported that the voice had always been abnormal but seemed to worsen with puberty. At age 10, at tonsillectomy, a laryngeal abnormality was detected by the anesthesiologist but was not pursued. At age 15, the laryngeal abnormality was again seen by the anesthesiologist during a cosmetic ear operation. The patient was subsequently referred to an otolaryngologist.

Pachyonychia congenita was present in the patient's maternal grandmother, mother, and older brother. In our patient the nails were carefully examined at birth and found to be normal. A whitish plaque was visualized in the mouth on the buccal mucosa.
ears to be wary but the family

mucosa with what mother described as "a patchy, furry tongue." This was treated as thrush. Within 5 days nail changes had begun to occur and "an infection" was diagnosed. Topical antibiotics were applied but treatment for both the oral lesions and fingers did not reverse the clinical picture. There were no neonatal teeth.

By age 3 months the yellowish brown discoloration of the nails had progressed to nail thickening; the characteristic nail pattern was present by age 5 years. Multiple episodes of purulent nail drainage had been observed and treated during this time period.

Additional manifestations of the disease included hyperkeratosis of the knees, elbows, and soles. Hyperhidrosis with painful bullae of the soles represented the single most annoying complaint of the patient. Scalp café au lait spots and subcutaneous cysts were present. The patient had a normal ophthalmologic examination. The hair was particularly thick and abundant.

The 3 other family members with the syndrome have virtually identical histories including subcutaneous cysts and oral leukoplakia but without the voice abnormalities.

Indirect laryngoscopy revealed an obvious posterior commissure mass. At direct laryngoscopy, there was a pink exophytic mass in the midline of the posterior commissure slightly above the level of the vocal cords and extending down to the cords (Fig. 1). No leukoplakia was seen on the lesion. A sub-total excisional biopsy

Fig. 1. Direct laryngoscopic photograph under general anesthesia. There is a multilobed midline mass in the posterior commissure.
Fig. 2. Photomicrograph of the laryngeal mass (H and E $\times 25$).
was performed. Care was taken to preserve normal laryngeal structures. The pathological findings are discussed below. Post-operatively the patient's voice had improved to near normal.

Repeat endoscopy was performed 3 months later. The lesion was minimally present in the posterior commissure and was removed by laser vaporization. There will be further regular follow-up.

Histopathology

The histopathologic evaluation (Fig. 2) was compatible with previous reports of oral and laryngeal biopsies of pachyonychia congenita [2,3,7]. The squamous mucosa was hyperplastic and showed pronounced focal intracellular vacuolation with sparing of the basal layer. There was no epithelial atypia. A patchy chronic inflammatory cell infiltrate was present in the subepithelial connective tissue.

Discussion

Despite the limited number of laryngeal cases which have been reported [1,2,4], use of the operating microscope and surgical excision of the mass appears to be an acceptable treatment for those patients with a voice handicap. Cohn et al. [2] have suggested that this disorder of the larynx has its worst manifestations in childhood and may regress in adulthood; however, Clementi et al. [1] have presented two adult patients with voice abnormalities, the oldest being 61 years. Post-operative recurrence appears to be minimal and aggressive surgical resection with potential injury to the laryngeal structures is not indicated.

Hoarseness or huskiness of voice may be more common than has previously been reported. Our experience suggests all patients with this syndrome and voice abnormality should be evaluated by an ENT surgeon. Endoscopic surgical debulking using forceps and/or laser is appropriate realizing periodic follow-up endoscopies may be necessary if the lesion recurs.

References

8 Nyhan, W.L., Genetic and Malformation Syndromes in Clinical Medicine, Yearbook Medical Publ., Chicago, 1976, p. 223.