CASE STUDY

Pachyonychia Congenita With Involvement of the Larynx

Paquioniquia congénita con compromiso laringeo

Hugo Rodríguez, Giselle Cuestas,* Adrián Zanetta, Ziomara Balbarrey

Servicio de Endoscopia Respiratoria, Hospital de Pediatría «Prof. Dr. Juan R. Garrahan», Buenos Aires, Argentina

Received 9 August 2012; accepted 29 October 2012

Clinical Case

We present the case of a 2-year-old male patient, with subungual hyperkeratosis and thickening and yellowish-brown discoloration of the ungual plate of his 20 digits, along with lingual leukoplakia from the first months of life. The patient came for consultation to the Respiratory Endoscopy Service for dysphonia present from the age of 10 months (Fig. 1).

His maternal great-grandmother, maternal grandmother, mother, uncle and sister all present pachyonychia congenita (PC). The patient is the youngest of the 6 members of the family with this disorder, which includes 4 generations.

There is no history of consanguinity. There are no dental or hair abnormalities, nor cystic lesions. The patient does not present other medical problems and his growth and development are normal.

The results of mycological tests of nail scrapings and whitish oral mucosal plaques were negative. Fibre laryngoscopy with topical anaesthesia revealed leukoplakia that involved the posterior commissure, interarytenoid mucosa and upper face of both vocal folds. Direct laryngoscopy was carried out under general anaesthesia general with biopsy of the lesion (Fig. 2A).

The diagnosis of dysphonia secondary to PC was based on the clinical and endoscopic findings, as well as on the anatomical pathology. The latter evidenced thickening of the epithelium with dyskeratosis, parakeratosis and acanthosis papillomatosis, marked spongiosis and slight lymphoplasmacytic infiltration in the chorion (Fig. 2B).

The child’s relatives were free from laryngeal involvement.

The patient’s dysphonia continued, without symptoms of respiratory obstruction, and he remained under follow-up by the endoscopy, dermatology and genetics services.

Discussion

Pachyonychia congenita is a rare genodermatosis that can involve the larynx. It is caused by mutations in 1 of the 4 keratin genes and is generally transmitted as an autosomal dominant feature.1–7 It was described for the first time in 1906.2,3 Approximately 474 families with PC have been documented to date; of these, 224 have undergone genetic tests.1

Although many different classifications have been proposed, there are 2 main subtypes: PC-1 phenotype (Jadassohn-Lewandowsky type), which is the most common variant and is linked to mutations in the genes of the keratins K6a and K16; and PC-2 phenotype (Jackson-Lawler type), related to mutations in K6b and K17.1–5

Traditionally, both PC-1 and PC-2 display subungual hyperkeratosis and thickening of all the nails of the hands and feet as their most notable feature. They also show focal palmoplantar keratoderma and follicular keratosis, principally on knees and elbows. The clinical difference between PC-1 and PC-2 has historically been suggested by the oral leukokeratosis that is more prominent in PC-1, and by the presence of cysts (multiple steatocystoma and hairy cysts), abnormalities in the hair and birth teeth in PC-2.1–3,6,7 Oral leukokeratosis oral is frequently one of the first signs of PC. It can be misdiagnosed as muguet and can cause difficulties...
Figure 1  (A) Endoscopic view of the larynx with leukoplakic lesions in the posterior commissure and vocal folds. (B) Thickening and discolouration of the nails of the patient’s hands and feet. (C) Hands of the patient, his mother and his sister, where hypertrophic ungual dystrophy can be observed.

Figure 2  (A) Enlarged endoscopic view of the larynx. (B) Histopathology: epithelial thickening with marked spongiosis can be seen.
in suction in the newborn.\textsuperscript{1,5,7} Findings typical of the PC-1 type were present in our case.

These clinical characteristics often present at birth or in early childhood, but there are variants that develop later in life (late-onset PC).\textsuperscript{3}

Few cases of laryngeal involvement in patients with PC have been described in the literature.\textsuperscript{2,3,8} In those with a known genotype, laryngeal pathology of a variable degree was seen in the phenotypes 1 and 2, with the most frequent mutation in K6\textsubscript{a}.\textsuperscript{2}

Dysphonia is reported in 16\% of the patients with PC.\textsuperscript{3,4} It is an early sign of laryngeal pathology\textsuperscript{3} and can be the main and only manifestation of laryngeal involvement. More infrequent is the occurrence of laryngeal obstruction due to leukokeratosis, which is a potentially deadly complication, more common at early ages and after an upper respiratory infection.\textsuperscript{2,3,9}

Children with dysphonia and PC should be examined using flexible laryngoscopy. We advise taking a biopsy if suspicious lesions are identified in order to rule out other laryngeal pathology. Endoscopic assessment reveals leukoplakic changes; the lesions range from slight thickening of the mucosa up to exophytic masses, with the posterior commissure being the most frequently involved area in the larynx.\textsuperscript{3,7,10}

The dysphonia associated with PC, especially after excessive use of the voice, normally resolves spontaneously with vocal rest. It can also improve spontaneously in a period of several years.\textsuperscript{1,3,4,8,10} Surgical procedures to improve voice quality should be avoided, because they can make the underlying condition worse.\textsuperscript{1} Keratins 6, 16 and 17 are in response to stress. Surgery and laser techniques can worsen the situation given that they could stimulate the mutated keratin; the treatment of choice is conservative therapy.\textsuperscript{2}

However, the rare occurrence of respiratory difficult requires emergency surgical intervention to re-establish the airway.\textsuperscript{3,5} In the published cases of laryngeal obstruction, microsurgical removal or CO\textsubscript{2} laser vaporization of the laryngeal leukokeratosis areas was successful. The procedure may need to be repeated if there is a relapse.\textsuperscript{1,2,9} We did not operate on our patient given the absence of respiratory obstruction symptoms.

It is important for the otorhinolaryngologists and dermatologists to bear in mind that laryngeal involvement and, on rare occasions, laryngeal obstruction can occur. Dysphonia is an early sign of laryngeal involvement and should provoke endoscopic assessment of the larynx. Early detection and possibly treatment of the laryngeal lesions can prevent obstruction of the airway. Given the unpredictable evolution of laryngeal involvement, patient follow-up is essential.

References