

## PAPILLON-LEFÈVRE SYNDROME: REPORT OF TWO CASES

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**ABSTRACT :** *Papillon-Lefèvre syndrome is a rare genetic disorder characterised by palmar-plantar hyperkeratosis and severe periodontitis.*

*We report two cases of Papillon-Lefèvre syndrome, aged 16 and 6 years, one of whom was previously misdiagnosed as psoriasis vulgaris and was treated successfully with acitretin. The other responded very well to topical emollients.*

**Key Words:** *Papillon-Lefèvre Syndrome, Palmar-Plantar Hyperkeratosis, Premature Loss of Teeth, Oral Cavity Examination, Acitretin Therapy.*

### INTRODUCTION

Papillon-Lefèvre syndrome is a rare genetic disorder characterised by palmar-plantar hyperkeratosis and premature loss of both the deciduous and permanent teeth, and first described by Papillon and Lefèvre in 1924 (1).

Since this condition is often confused with other hereditary keratodermas, oral cavity examination is mandatory.

Here we report two cases of Papillon-Lefèvre syndrome, one of which was previously misdiagnosed as psoriasis vulgaris because the oral cavity examination was overlooked.

### CASE 1

A 16-year-old girl was referred to our outpatient clinic by the Gazi University Faculty of Dentistry, Department of Pediatric Dentistry because of palmoplantar keratoderma of 15 years

duration. Erythematous plaques on her knees, elbows, palms and soles had been previously diagnosed and treated as psoriasis with topical agents.

Her parents were not consanguineous. No similar condition was observed among the family members and none of the relatives had any skin disease.

Physical examination revealed symmetrical, well-demarcated, rough, erythematous, hyperkeratotic, scaly lesions on her knees, elbows, palms and soles (Fig.1a). Transverse grooves, which were present in most of the fingernails, were most prominent on the thumb nail (Fig.1b). Her general growth, hair growth, sweating and mental development were all normal.

On her oral cavity examination, apart from permanent second molar, no other teeth were



Fig. 1: Erythematous, hyperkeratotic, scaly lesions on the palm of case 1 (A). Transverse groove on the thumb nail of case 1 (B). Teeth loss (apart from permanent second molar) of case 1 (C). Hyperkeratosis, focal parakeratosis and marked acanthosis of case 1 biopsy specimen (HE x 400) (D).

seen (Fig.1c). Excessive alveolar bone loss was detected on radiographic examination .

Histological examination of the foot lesion showed hyperkeratosis, focal parakeratosis and marked acanthosis (Fig.1d). No intracranial calcification was noted on the skull X-rays. Laboratory findings were normal.

Papillon-Lefèvre syndrome was the diagnosis and oral acitretin at 0.6 mg/kg/day was given for two weeks, after which clinical improvement was observed. For her dental condition, a removable prosthodontic appliance was suggested to be used by the Pediatric Dentistry Department. The patient is still under our clinical observation.

## CASE 2

Gazi University Faculty of Dentistry consulted us about a 6 years old child who had teeth loss since 2 years of age and newly developed skin lesions in the form of roughened and thickened palmar and plantar skin. Consanguinity was positive in the family and no similar condition was evident among the family members.

On physical examination there were well demarcated erythematous, hyperkeratotic scaly lesions on the palms and soles. Hair and nails were normal in appearance. No growth or mental retardation was detected.



Oral cavity examination revealed total absence of the deciduous teeth (Fig. 2). Radiographic examination showed that all the permanent teeth were impacted. No intracranial



Fig. 2: Total absence of the deciduous teeth of case 2.

calcification was detected on skull X-rays. No abnormal laboratory findings were seen.

Our provisional diagnosis was Papillon-Lefèvre syndrome.

Topical emollients were used on the lesions after which a marked improvement was detected. Dental prosthetic appliance was recommended by the pediatric dentist.

#### DISCUSSION

Papillon-Lefèvre syndrome is an autosomal recessive disease which develops within the first few months of life. In this entity, the periodontal tissues of the patients reveal severely inflamed gingiva, deep periodontal pockets, diffuse and extensive bone resorption, loosening and exfoliation of the teeth (2, 3). The cutaneous changes are usually manifested when the periodontal abnormalities develop in the first year (4). This cutaneous pathology includes symmetrical, well-demarcated, erythematous, scaly, hyperkeratotic lesions on the palms and soles. Spread of the keratoderma to the dorsa of the hands and to the Achilles tendons, elbows and knees is found in some cases (5).

Nail changes include transverse ridging of nails, onychogryphosis, and sheeted follicular hyperkeratosis (6). Transverse grooves on the fingernails were observed in one of our cases.

Other findings such as intracranial calcification, hyperhidrosis, susceptibility to infections, and mental retardation have been reported (8). We could not observe any of these findings in our two cases.

Histological changes include hyperkeratosis, focal parakeratosis, marked acanthosis, and nonspecific mononuclear perivascular infiltrate. Since dental changes are overlooked by dermatologists, this syndrome is often confused with other hereditary keratodermas, especially those of Unna-Thost and Mal de Meleda where no odontological findings are seen. Indeed, one of our cases was diagnosed and treated as psoriasis for two years and the diagnosis of Papillon-Lefèvre syndrome was only possible after the odontological problems were noticed. For this reason we think that careful oral examination in cases of palmoplantar keratoderma is necessary.

The treatment of Papillon-Lefèvre syndrome is palliative. While one of our patients responded very well to topical emollients the other patient required therapy with acitretin.

Although the association of palmoplantar keratoderma with periodontal disease was first reported by dermatologists, most subsequent publications have appeared in the dental literature. The diagnosis of Papillon-Lefèvre syndrome may require collaboration between the dermatologist and dentist.

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