PAPILLON - LEFÈVRE SYNDROME: A CASE REPORT OF TWO SIBLINGS

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Papillon- Lefèvre Syndrome (PLS) is an autosomal recessive disorder characterized by severe periodontitis and palmoplantar hyperkeratosis. Microbial lipopolysaccharides and mutations of cathepsin C gene has been suggested as the most probable etiological factors for PLS. This article presents a case of a thirteen year old female and her sibling suffering from the disease.

Keywords: Papillon- Lefèvre Syndrome, palmoplantar hyperkeratosis, early onset pèriodontitis.

The Papillon- Lefèvre syndrome (PLS) was first described by Papillon and Lefèvre in 1924[1], is characterised by pathognomic palmoplantar hyperkeratosis and rapidly progressive periodontitis. Gorlin et al also suggested, the calcification of the duramater as the third component of this unusual syndrome.[2] There is no significant sex predeliction and both the dentitions are equally affected without any racial predominance,[3] however incidence in Saudi Arabia is higher for which cluster marriages may be the reason.[4]

The dermatological lesions begin to appear during the infantile period of life. The oral symptoms of the disease start with the eruption of primary teeth. The characteristic periodontal manifestations of the syndrome are gingival inflammation, rapid destruction of the periodontal tissues with deep periodontal pockets and mobile teeth which subsequently may lead to complete loss of primary dentition. After the loss of primary teeth the inflammatory symptoms subside.[5] The symptoms reappear with the eruption of the permanent dentition and results in the premature loss of all or most of the teeth

CASE REPORT

A 13 year old female patient was referred to the department of periodontology with the complaints of generalized severe teeth mobility and gingival inflammation. Clinical examination revealed generalized teeth mobility, several missing teeth and gingival bleeding on slight provocation (Fig-1). General examination of the patient revealed hyperkeratotic skin lesions on palms, soles, elbows and on knees (Fig-2). All these clinical findings were suggestive of Papillon- Lefèvre syndrome (PLS).

Detailed case analysis of the patient revealed that she had experienced the identical oral problems in her early childhood and finally lost all of her deciduous teeth and soon after eruption of permanent teeth she started experiencing the similar problems. Her family history disclosed that her elder brother (now 17 years of age) showed similar clinical findings and was edentulous by the age of 16. (Fig-3)

Radiographic findings were suggestive of generalized severe bone loss around all her teeth with widened periodontal ligament space (Fig-4). There were no signs of calcification of the duramater in CT scan examination. Histological examination of the skin biopsy demonstrated hyperkeratosis and acanthosis with lymphocytic infiltration (Fig-5).

Periodontal treatment- Treatment was started with scaling and root planing and oral hygiene instructions were given. Systemic antibiotics (250 mg tetracycline qid for 30 days) and chlorhexidine mouth wash were also prescribed. After a month when her periodontal condition got stable, erupting teeth (15, 25, 34, 35, 44, 45) was submerged beneath the mucoperiosteum by surgery with the hope of preservation of them for future. But on follow-up visits periodontal problems reoccurred in spite of extensive maintenance program. Teeth with hopeless prognosis were extracted and a

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removable partial denture was fabricated for rehabilitation (Fig-6). Patient was quite satisfied with denture on follow-up visits.

DISCUSSION

PLS is a rare hereditary disorder which affects the patient in early stages of life (with the eruption of deciduous teeth). The etiology of PLS has not been confirmed. However gram negative lipopolysaccharides are considered as the primary etiological factor in periodontitis including PLS.[6] Another important etiological factor, recognised for PLS is cathepsin C gene mutations. Cathepsin C gene mutations have been found to be associated with other forms of periodontitis (like Haim-Munk syndrome and prepubertal periodontitis) and have been known to be responsible for the C enzyme activity.[7] The activity of this enzyme is required for proteolytic activation of neutrophil serine proteases, cathepsin G, elastase and proteinase 3. Inactivation of these neutrophil serine proteases results in the dysregulation of the host immune response to microbial infection. Local tissue degradation may be exacerbated by the accumulation of inflammatory infiltrate at the sites of infection.[8] Ryu et al in their biochemical study suggested that cathepsin C mutation and failure in the activity of neutrophil serine proteases may lead to improper localized microbial induced inflammatory response in inflamed periodontal tissues, leading to severe periodontal tissue destruction.[9] Cagli et al proposed that due to this aberrant inflammatory response, the infiltrate of inflammatory cells at sites of periodontal infection is not under normal regulatory control and the increased influx of neutrophils and the retention of inflammatory infiltrate and their proteases may be responsible for continued periodontal destruction, which may be responsible for uncontrolled periodontitis in established cases of PLS.[8]

Various treatment modalities have been advocated previously for the treatment of PLS. Early extractions of deciduous teeth with the intention of complete elimination of periodontopathic bacteria has been suggested to prevent subsequent infection of permanent dentition.[10]

Use of systemic antibiotic agents in conjunction with periodontal therapy and extraction of periodontally involved deciduous teeth were reported effective in treatment of PLS cases.[11-13] Other chemotherapeutic agents which have been suggested effective in the treatment of PLS are synthetic retinoids and the derivatives including isotretinoin, etretinate and acitretin combined with conventional periodontal therapy.[13-15] Successful osseointegration and proper functional loading of dental implants have been reported as successful method of rehabilitation in PLS patients after teeth removal.[16]

Present case initially showed response to the conventional periodontal therapy (scaling and root planing and systemic antibiotic administration) but the periodontal problems reoccurred and patient had to be rehabilitated with removable partial denture. Patient was referred to the department of dermatology for the treatment of skin lesions and retenoids were prescribed which were not useful. There is further need of studies, directed towards the search of effective and stable treatment options of PLS so that the natural teeth can be preserved of the patients with PLS for better life style.

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Government College of Dentistry Indore celebrates Golden Jubilee

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It was in 1961 when this college was started as a small sapling with untiring efforts of Dr S L Mangi, Shri Mishrilal Gangwal and the then Director health Service Dr G L Sharma. Today, the institution has brought out about 46 batches of graduates who have settled not only in Madhya Pradesh but all over the country as well as abroad and have earned good name for the institution

It is our privilege to welcome all the ex-students of this college and extend our heartiest congratulations on this golden jubilee occasion of this institution of which I, Dr V P Jalili, have been associated with it since its infancy.

Wishing the golden jubilee function a grand success.

Dr V P Jalili
Chief Editor