CASE REPORT

# Haim Munk syndrome: A Case Report

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## ABSTRACT

**Introduction:** Haim-Munk syndrome (HMS) is a rare autosomal recessive palmoplantar keratoderma (PPK) combined with severe destruction of periodontium which is differentiated from Papillon-Lefèvre syndrome based on the presence of certain additional features.

**Case report:**A 23 year old female patient reported with the chief complaint of multiple missing teeth and wanted replacement for the same. She had premature exfoliation of primary and many of her permanent teeth. General examination revealed mild, diffuse palmoplantar keratosis with shortening of fingers and curved nails. Severe aggressive periodontitis was evident with multiple missing teeth on intraoral examination. Skin lesions were treated with Vaseline and 6% salicylic acid. Dental treatment included oral prophylaxis with 0.2% chlorhexidine mouth rinse, extractions with systemic antibiotics followed by prosthetic managment.

**Conclusion:**This case of palmaplantar keratoderma with severe early onset periodontitis was differentiated from the more common condition of PLS and diagnosed as HMS based on the additional clinical findings present in this patient.

**Keywords:** Haim-Munk syndrome, Papillon-Lefevre disease, alveolar bone loss, keratosis palmoplantaris, Aggressive Periodontitis.

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## INTRODUCTION

Haim-Munk syndrome (HMS) or Cochin Jewish disorder is a rare autosomal recessive type IV palmoplantar keratoderma (PPK) that involves abnormal hyperkeratosis of the palm of the hands and sole of the feet combined with severe destruction of periodontium leading to premature tooth loss. In addition, other clinical findings seen in these patients include acroosteolysis, arachnodactyly, atrophic changes of the nails (onychogrophosis) and deformity of the fingers, and recurrent pyogenic skin infections.[1]

## CASE REPORT

A 23 year old female patient reported to the department of Periodontics, Mahatma Gandhi Post Graduate Institute of Dental Sciences, Puducherry with the chief complaint of multiple missing teeth and wanted replacement for the same. All her primary teeth erupted sequentially at appropriate age but exfoliated prematurely. She had normal eruption of permanent teeth which started exfoliating prematurely by the age of 16. She also complained of decreased sweating with dryness and itching of palms and soles since childhood. She was born of second degree consanguineous marriage and no similar presentations are seen in any of her family members.

General examination revealed mild, diffuse palmoplantar keratosis with shortening of fingers and curved nails, knuckle pads and lusterless hair (Fig-1,2).

Extraoral examination showed mild maxillary hypoplasia with relative mandibular prognathism. Thinning of upper and lower lipsis seen with slight facial asymmetry and deviation of mandible to the right side.

On intraoral examination patient was partially edentulous with grade III mobility in relation to 38 and grade I mobility in 12. Generalized severe clinical attachment loss was evident but no deep periodontal pockets were present (Fig-3). Based

on the clinical findings a provisional diagnosis of Papillon-Lefevre syndrome was made. The differential diagnoses taken into consideration were Haim-Munk syndrome, Hypotrichosis Acroosteolysis Palmoplantar keratoderma Peridontitis syndrome (HOPP), Weary-Kindler and Kindler syndromes, Variant carvajal syndrome,Clouston syndrome and Mandibulo Acral dysplasia.

*INVESTIGATIONS*

Skin biopsy showed marked hyperkeratosis but no decrease in density of sweat glands and there was no hearing loss. Hand wrist and feet X-raysshowed acral osteolysis (Fig-4). Lateral skull radiograph does not show any calcifications. OPG revealed generalized moderate to severe horizontal bone loss with periodontal ligament space widening in relation to 42 with missing 15,16,17,21,22,34, 35,36,37,41,45,46 and 47.

*TREATMENT*

Full mouth oral prophylaxis was done and oral hygiene instructions and 2% chlorhexidine were given followed by extraction of 38 along with systemic antibiotics. Since no progressive periodontal lesions were seen within 6 weeks follow up, a removable partial denture was fabricated for the patient. Patient was referred to dermatology department for management of skin lesion and was given Vaseline and 6% salicylic acid for a period of one month.

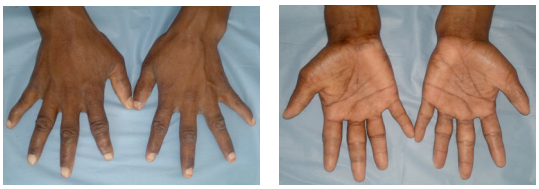

**Figure-1:** Mild, diffuse palmar keratosis with shortening of fingers and curved nails, knuckle pads

## DISCUSSION

Of the many PPK conditions, only Papillon-Lefèvre syndrome (PLS) and HMS are associated with premature periodontal destruction. Although both PLS and HMS share the cardinal features of

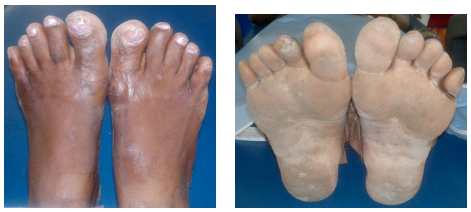

**Figure**-2: Diffuse keratosis with shortening of fingers of feet

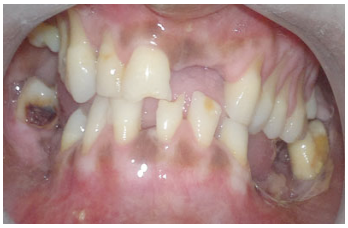

**Figure-3:** multiple missing teeth, severe periodontitis with clinical attachment loss

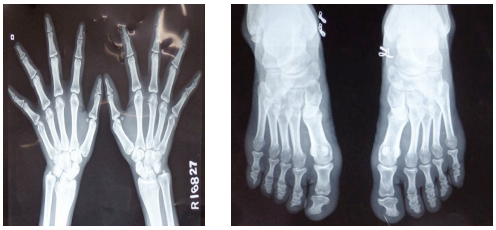

**Figure-4:** Hand wrist and feet radiograph showing

PPK and severe periodontitis, anumber of additional findings are reportedin HMS including acro-osteolysis, arachnodactyly,onychogrophosis, pes planus and recurrent pyogenic skin infections. Skin manifestations in HMS are usually more extensive and severe, and the periodontium is less severely affected, in contrast to PLS.[1] Other syndromes which reported PPK in association with periodontitis are HOPP,[2,3] a case of Weary-Kindler syndrome[4] and a case ofVariant Carvajal syndrome.[5]

HMS is very rare with <100 cases reported in the literature so far. While PLS cases have been identified throughout the world, HMS has mainly beendescribed among descendants of a religiousisolate originally from Cochin, India.[6] One unrelated Brazilian kindred has also been reported.[7] Recently, an isolated case of HMS in a 12 year old girl and her sibling is reported from India in 2014.[8] Also a case report of two Jordanian sisters with suspected HMS has been reported recently who also presented with psoriasiform lesions around their lips.[9] Here is another isolated case of HMS from Tamil Nadu, India. Parentalconsanguinity is a characteristic ofmany cases of both conditions which is true in the present case also.

Recently, mutations of the lysosomal protease cathepsin C gene[10] (CTSC) located in chromosome 11q14-q21[11] region have been identified as the underlying genetic defect in PLS and HMS. To date two mutations in CTSC gene are reported in families affected by HMS: one in codon 286 of exon 6 in an in bred Jewish non-Askhenzai family from Cochin, India and in codon 196 of exon 4 in one Brazilian kindred.[7,12] CTSC gene mutations are known to cause impaired phagocytosis and leukocyte functional disorders.[1] The diagnosis of HMS is based on clinical features and radiographic evaluation of bone deformities of fingers and toes and alveolar bone destruction.[1] Additional to PPK and severe periodontitis this patient had the typical clinical features of HMS like acro-osteolysis and nail deformities. Also absence of, dural calcifications and normal density of sweat glands rule out other possible conditions like PLS and Clouston Syndrome. Also another striking feature is the presence of a stable state in the patient without any progressive periodontal lesions.

Thus it is important to differentiate HMS from PLS from a periodontal point of view as aggressive treatment options like complete extraction and prosthesis will be a crime in such patients. Even the role of systemic antibiotics in the management of this disease is limited. With proper periodontal management and maintenance care HMS patients can be managed like any other periodontitis patient.

## CONCLUSION

This case of palmaplantar keratoderma with severe early onset periodontitis was differentiated from the more common condition of PLS and diagnosed as HMS based on the additional clinical findings present in this patient. These features are less known to periodontitis and are important in determining the treatment plan for HMS patients. The keys to success in the management of HMS are correct diagnosis and properly executed treatment.

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