

# Haim-Munk syndrome with erythroderma - A case report

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**Abstract** Haim-Munk syndrome (HMS) is a rare autosomal recessive disorder of keratinization clinically characterized by early onset severe periodontitis, palmoplantar keratoderma, onychogryphosis and arachnodactyly. Pes planus, acro-osteolysis and recurrent pyogenic infections are the frequent findings. Lysosomal protease cathepsin C gene mutation is the key etiological factor. Diagnosis is established on the basis of clinical features. We described a case of HMS presented in our OPD, with an unusual presentation i.e. erythroderma.

**Key words**

Aggressive periodontitis, Haim-Munk syndrome, Palmo-plantar keratoderma, Papillon Lefèvre syndrome.

## Introduction

Palmoplantar keratoderma (PPK) is a heterogeneous condition characterized by hyperkeratosis and erythema of the palms and soles. Papillon-Lefèvre syndrome (PLS) and Haim-Munk syndrome (HMS) are rare autosomal recessive PPK characterized by early onset periodontal disease.<sup>1,2</sup> Patients with HMS also show hypertrophy and curving of nails (onychogryphosis), flat foot (pes planus), extreme length and slenderness of fingers and toes (arachnodactyly) and osteolysis involving the distal phalanges of fingers and toes (acro-osteolysis).<sup>3</sup> We report a case of HMS presented in our OPD, in a 32-year-old woman who suffered from dry and thickened skin with recurrent skin infections.

## Case report

A 32-year-old female from Sialkot with a consanguineous family presented in outpatient department of Dermatology, Mayo Hospital Lahore. Written informed consent was taken from the patient.

She was evaluated for palmoplantar hyperkeratosis, multiple psoriasiform plaques on trunk and extremities, periodontitis and recurrent skin and soft tissue infections. The patient started developing thickening of the skin of her palms and soles and shedding of primary dentition as a result of periodontitis at the age of 4 years. Secondary dentition was also affected by periodontal disease, with subsequent premature shedding of permanent teeth. At the age of 16, she lost all her permanent teeth. She also gave history of multiple episodes of erythroderma.

The physical examination showed severe yellowish hyperkeratosis of the palms and soles along with fissures (**Figure 1**). The keratotic lesions of the palms extended to the dorsal

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**Figure 1** Plantar keratoderma



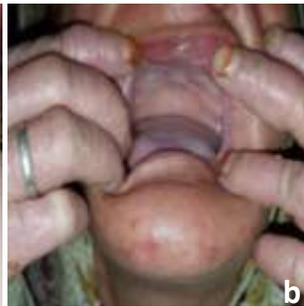
**Figure 2** Feet of the patient



**Figure 3** Palms and dorsal aspects of hands



**Figure 4** Dorsal aspect of hands, slender fingers, onychogryphosis and lower limbs of the patient



**Figure 5** Pes planus and anodontia



**Figure 6** Soft tissue infection

aspects of the fingers (**Figure 3**); besides the lesions on the soles, which were more severe, erythema and scaling extended up to the upper arm and mid-thigh respectively (**Figure 4, 6**).

In addition, the patient had multiple, scaly, and erythematous patches on elbows, arms and shins. Few of her finger nails showed onychogryphotic changes (**Figure 3, 4**). She also had pes planus (**Figure 5**), and the X-ray of hands revealed typical arachnodactyly and acroosteolysis.

Blood cell count, erythrocyte sedimentation rate, liver function transaminase levels, alkaline phosphatase, total bilirubin, renal functions, serum electrolytes were normal. Rheumatoid factor and Antinuclear antibodies were not detected. Based on the patient's history, clinical and radiographic findings, the diagnosis of Haim-Munk Syndrome (HMS) was made.

The patient's family members were unaffected and no abnormal signs were present on physical examination.

The patient was treated with oral retinoids, topical diluted steroids, keratolytics and systemic antibiotics. Oral antihistamines were also given. She was asked to visit after a week and then after four weeks. The follow-up visits showed complete remission of skin infection along with improvement in her symptoms.

## Discussion

HMS is a rare autosomal recessive congenital disorder which was named after the investigators Salim Haim and Dr. J. Munk who reported it among members of Jewish family from Cochin, India.<sup>4</sup> It affects males and females in equal number.<sup>5</sup>

HMS presents with severe and extensive cutaneous manifestations. In addition to the marked palmoplantar hyperkeratosis, patients may present with erythematous, scaly and well circumscribed patches on the knees, elbows and dorsal aspect of the hands and feet. Severe, early-onset progressive periodontal disease affects both the deciduous and permanent dentitions and presents with gingival inflammation and alveolar bone destruction.<sup>6</sup> Onychogryphosis, arachnodactyly, acro-osteolysis and pes planus are additional features that help to distinguish HMS from other forms of PPKs.<sup>7</sup> A peculiar claw-like volar curve of the fingers with tapered and pointed phalangeal ends is a hallmark of HMS. Destructive arthritis of the wrist and shoulder joints has been reported in isolated cases.<sup>8</sup> Patients with HMS have increased susceptibility to bacterial skin infections.<sup>1</sup>

PLS is a rare syndrome characterized by certain features similar to those seen in HMS. Genetic analysis of many kindreds suggest these two syndromes to be due to mutation of a gene CTSC, known as cathepsin C.<sup>9-14</sup> This gene encodes the production of an enzyme, lysosomal protease and it is expressed in various organs and tissues of our body. It also plays a role in the differentiation of epidermis and binding gingiva. Mutation of the CTSC gene may result in reduced levels of cathepsin C or defective cathepsin C that cannot perform its normal functions in the body. Although both PLS and HMS share the cardinal features of PPK and severe periodontitis, a number of additional findings are reported in HMS including

arachnodactyly, acro-osteolysis, atrophic changes of the nails, and a radiographic deformity of the fingers.<sup>7</sup>

Management of HMS needs a multidisciplinary approach however dermatological therapeutic options include oral retinoids, cotrimoxazole and topical keratolytics.<sup>15,16</sup>

## Conclusion

We are presenting this case as it is a very rare disease and not many cases have been reported and our case is the first one to be reported with history of recurrent episodes of erythroderma. HMS requires a multidisciplinary approach to achieve better management and control and our patient will be managed accordingly in future.

## References

1. Oji V, Metze D, Traupe H. Inherited Disorders of Cornification. In: Griffiths C, Barker J, Bleiker T, Chalmers R, Creamer D, eds. *Rook's Textbook of Dermatology*. 9th edn. John Wiley & Sons Ltd. 2016; pp. 65.61.
2. Wei H, Wee LWY, Born B, Seang S, Koh MJA, Yee R, Lin G, Rafi'ee K, Mey S, Tan EC. Palmoplantar keratoderma, oral involvement, and homozygous CTSC mutation in two brothers from Cambodia. *J Med Genet A*. 2020; **182(2)**: 296-302.
3. Janjua SA, Iftikhar N, Hussain I, Khachemoune A. Dermatologic, periodontal, and skeletal manifestations of Haim-Munk syndrome in two siblings. *J Am Acad Dermatol* 2008; **58(2)**: 339-44.
4. Aboud KA, Aboud DA. Salim Haim and the syndrome that bears his name. *Dermatol Online J*. 2011; **17(8)**: 15.
5. Pahwa P, Lamba AK, Faraz F, Tandon S. Haim-Munk syndrome. *J Indian Soc Periodontol*. 2010; **14(3)**: 201-3.
6. Erciyas K, Inaloz S, Erciyasc AF. Periodontal Manifestations in a patient with Haim-Munk syndrome. *Eur J Dent*. 2010; **4(3)**: 338-40.
7. Hart TC, Hart PS, Michalec MD, Zhang Y, Firatli E, Van Dyke TE *et al*. Haim-Munk syndrome and Papillon-Lefevre syndrome

- are allelic mutations in cathepsin C. *J Med Genet.* 2000; **37**: 88-94.
8. Lidar M, Zlotogorski A, Langevitz P, Tweezer-Zaks N, Zandman-Goddard G. Destructive arthritis in a patient with Haim-munk syndrome. *J Rheumatology.* 2004; **31(4)**: 814-17.
  9. Pap ÉM, Farkas K, Tóth L, Fábos B, Széll M, Németh G, Nagy N. Identification of putative genetic modifying factors that influence the development of Papillon-Lefèvre or Haim-Munk syndrome phenotypes. *Clin Exp Dermatol.* 2020; **45(5)**: 555-59.
  10. Sulák A, Tóth L, Farkas K, Tripolszki K, Fábos B, Kemény L, Vályi P, Nagy K, Nagy N, Széll M. One mutation, two phenotypes: a single nonsense mutation of the CTSC gene causes two clinically distinct phenotypes. *Clin Exp Dermatol.* 2016; **41(2)**: 190-5.
  11. Cury VF, Gomez RS, Costa JE, Friedman E, Boson W, De Marco L. A homozygous cathepsin C mutation associated with Haim-Munk syndrome. *Br J Dermatol.* 2005; **152**: 353-6.
  12. Rai R, Thaigarajan S, Mohandas S, Natarajan K, Sekar CS, Rama lingam S. Haim Munk syndrome and Papillon Lefevre syndrome – allelic mutations in cathepsin C with variation in phenotype. *Int J Dermatol* 2010; **49(5)**: 541-3.
  13. Lamort AS, Hamon Y, Czaplewski C, Geldon A, Seren S, Coquet L, Lecaille F, Lesner A, Lalmanach G, Gauthier F, Jenne D, Korkmaz B. Processing and Maturation of Cathepsin C Zymogen: A Biochemical and Molecular Modeling Analysis. *Int J Mol Sci.* 2019; **20(19)**: 4747.
  14. Molitor A, Prud'homme T, Miao Z, Conrad S, Bloch-Zupan A, Pichot A, Hanauer A, Isidor B, Bahram S, Carapito R. Exome sequencing identifies a novel missense variant in CTSC causing nonsyndromic aggressive periodontitis. *J Hum Genet.* 2019; **64(7)**: 689-94.
  15. Mohan RS, Verma S. Haim Munk syndrome: Report of two siblings of Northern India treated with acitretin. *Indian J Dermatol Venereol Leprol* 2011; **77**: 252.
  16. Khan U, Iqar S, Khalid L. "Haim Munk Syndrome: two siblings of Southern Pakistan treated with acitretin." *Pak Oral Dental J.* 2012; **32(1)**.