CASE REPORT

Hermansky Pudlak Syndrome Type 2: A Rare Case Report

Yogesh Chhaparwal[1], Mathangi Kumar[1], Shubha Chhaparwal[2]*

[1]Department of Oral Medicine and Radiology, [2]Department of Conservative Dentistry and Endodontics, Manipal College of Dental Sciences, Manipal, Manipal Academy of Higher Education, Manipal-576104 (Karnataka), India

Abstract:

Hermansky-Pudlak Syndrome (HPS) is a rare autosomal recessive disorder characterized by Oculocutaneous Albinism (OCA), platelet disorder, and ceroid accumulation. It is common in North West Puerto Rico region, and the incidence reported is 1/500000. It is a rare genetic disorder with platelet dysfunction resulting in bleeding diathesis. Here we report one such rare case of HPS type 2 in a 7-year-old boy with difficulty in chewing.

Keywords: oculocutaneous albinism, Hermansky Pudlak syndrome, platelet disorder

Introduction:

Hermansky–Pudlak Syndrome (HPS) is a rare autosomal recessive disorder characterized by a triad of oculocutaneous albinism, bleeding diathesis and pulmonary fibrosis. The condition was first described by Hermansky and Pudlak in 1959 [1]. It is common in North West Puerto Rico region and the incidence reported is 1/500000 [2]. However, this syndrome is extremely rare in the Indian population. Genetically, there are at least eight distinct types of HPS that have been identified with HPS-1 being the most common subtype. The syndrome is associated with a mutation in the HPS 1 gene, which is mapped to the chromosomal site at 10q23. HPS gene is involved in the formation and trafficking of lysosome-related organelles, which have been identified in melanocytes, platelets, and epithelial cells [3]. The structures which are commonly affected include skin, eye and oral mucosa. This syndrome is characterized by oculocutaneous albinism, bleeding diathesis, and pulmonary fibrosis [4]. The diagnosis is essentially based on unique clinical features that are characteristic of this genetic disorder. This is the first case of HPS type 2 reported in a young child of Indian origin.

Case Report:

A 7-year-old boy presented to the Department of Oral Medicine and Radiology, Manipal College of Dental Sciences, Manipal, with a complaint of pain in multiple decayed teeth, which interfered with normal chewing. A detailed medical history revealed that the child was under treatment for miliary tuberculosis for the last two years. Parents reported that the developmental milestones were attained at an appropriate age. The patient's parents were second degree consanguineously married, and his elder brother died due to pneumonia (Fig. 1).

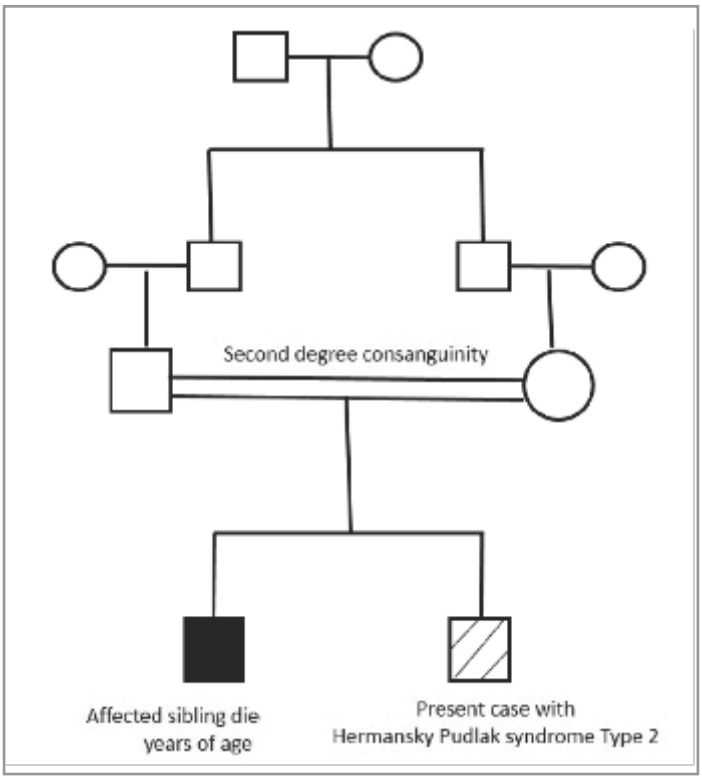

Fig. 1: A Pedigree Chart Showing Parental Consanguinity, Affected Elder Sibling and Present Case with Hermansky Pudlak Syndrome Type 2

Extraoral examination revealed blonde hair, micro-cephalic head, and facial features showed protruding midface with long, flat philtrum, low set posteriorly rotated ears. Broad nasal root and thin upper lips. There was short stature, and the height was in the third percentile, which was below the average range for his age (Figs. 2, 3). Ophthalmology examination revealed oculocutaneous albinism, horizontal nystagmus, and strabismus. The intraoral examination revealed a poor oral hygiene status with multiple carious teeth with pulpal exposure. No soft tissue abnormalities were noted. Panoramic radiograph showed widespread deep dentinal caries involving the entire dentition (Fig. 4). A genetic work-up was advised and performed at the Genetics unit of our institute. Genetic studies revealed a mutation in the adapter-related protein complex beta-1 subunit gene (AP3B1, 603401.0001), which was suggestive of HPS type 2. Here we are reporting a rare case of HPS in a 7-year-old boy with characteristic oculocutaneous albinism and oral findings.

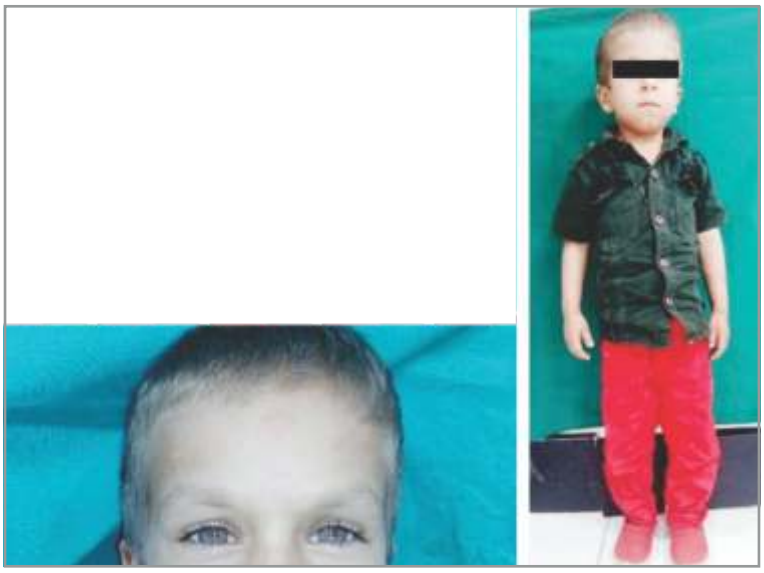

Fig. 2 And Fig. 3: Typical Oculocutaneous Albinism with Blonde Hair and Short Stature

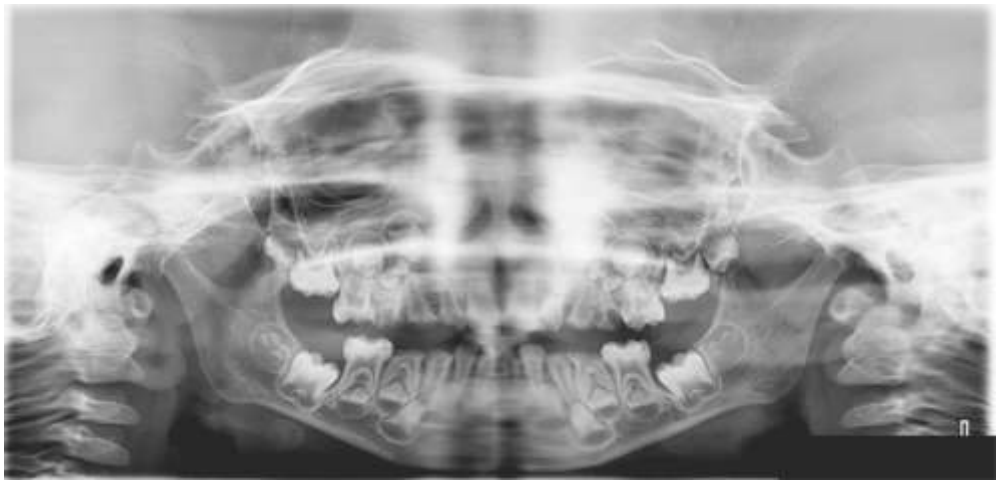

Fig. 4: Panoramic Radiograph Showing Widespread Carious Involvement of Teeth

Discussion:

HPS is a rare autosomal recessive disorder characterized by a triad of oculocutaneous albinism, bleeding diathesis, and pulmonary fibrosis. The condition was first described by Hermansky and Pudlak in 1959. It is common in North West Puerto Rico region, and the incidence reported is 1/500000. However, this syndrome is extremely rare in other parts of the world. The syndrome is associated with a mutation in the HPS1 gene, which is mapped to the chromosomal site at 10q23HPS gene is involved in the formation and trafficking of lysosome-related organelles, which have been identified in melanocytes, platelets, and epithelial cells. The diagnosis is

essentially based on unique clinical features that are characteristic of this genetic disorder [5].

Clinical manifestations of HPS are secondary to the inherent neutropenic disorder, which makes the individual susceptible to opportunistic infections. Likewise, the present case also presented as pulmonary tuberculosis, which was one of the components of HPS. This syndrome is characterized by the defect in the genes responsible for the breakdown of liposomes, thus resulting in the accumulation of ceroid lipofuscin (mucopolysaccharidosis) in the organs leading to systemic complications like pulmonary fibrosis [6].

Genetically, there are at least eight distinct types of HPS that have been identified with HPS-1 being the most common subtype. Genetic testing is required to establish various types of HPS. The present case was a rare type 2 of HPS. The most commonly reported complication reported with this condition is a respiratory difficulty, pulmonary fibrosis, and bleeding tendencies [7-8].

The diagnosis is established by typical skin features, hair, and ocular hypopigmentation. In the present case, consanguineous marriage between parents was reported, which increases the risk in offspring. The present case showed characteristic features of HPS.

The most common oral manifestation is bleeding from the oral cavity and gingival enlargement [9-10]. Individuals with HPS require special dental considerations and treatment modifications. Oral examination for such individuals requires avoidance of the usage of routine dental chair operatory light as these individuals are photophobic and may perceive abnormal stimulus from such a light source, thereby causing visual disturbances. The use of protective eyewear with ultraviolet filters may be a safe approach to prevent them from such vision problems. Since individuals with HPS have platelet dysfunction, they are at increased risk of bleeding while performing dental procedures. The use of local hemostatic agents to control bleeding should be employed while giving dental care. The various local hemostatic agents used include oxidized cellulose, absorbable gelatin sponge, and topical thrombin. To protect these patients from unnecessary risks, systemic medication with an antifibrinolytic agent may also be considered. For performing any invasive dental procedure, consultation with a hematologist should be taken. Investigations such as bleeding time and platelet function test should be undertaken before any invasive treatment procedures.

Prophylactic use of desmopressin may be considered during any invasive procedure to manage bleeding diathesis [11]. For achieving pain control, the use of aspirin and nonsteroidal anti-inflammatory drugs should be avoided, and acetaminophen is the drug of choice. The role of preventive care in these patients should be emphasized with regular recall visits every three months for oral hygiene reinforcement.

Conclusion:

Dental care in patients with HPS requires special considerations. The presence of oculocutaneous albinism causes societal problems and can result in treatment delay. The present case signifies the need for dental clinicians to be well versed with general features and be competent enough to manage any potential complications and thereby provide comprehensive dental care. These children require restoration of emotional and social care apart from regular treatment needs.

References

1. Feliciano NZ, Rivera E, Agrait E, Rodriguez K. Hermansky-Pudlak syndrome: dental management considerations. *J Dent Child (Chic)* 2006;73(1):51-56.
2. Vani R, Keertihvasan S, Anbananthan K. Hermansky-Pudlak syndrome: A case report. *J Assoc Chest Physicians* 2014;2:78-80
3. Hurford MT, Sebastiano C. Hermansky-Pudlak Syndrome: Report of a case and review of the literature. *Int J Clin Exp Pathol* 2008;1(6):550-554.
4. Murthy V, Yuvraj V, Thomas S, Nair P. Prosthodontic management of an albinism patient-dental implications and management. *BMJ Case Reports* 2013:1136-1137.
5. Vicary GW, Vergne Y, Santiago-Cornier A, Young LR, Roman J. Pulmonary fibrosis in Hermansky–Pudlak syndrome. *Ann Am Thorac Soc* 2016;13(10):1839-46.
6. Huizing M, Malicdan MC, Wang JA, Pri-Chen H, Hess RA, Fischer R *et al.* Hermansky–Pudlak syndrome: Mutation update. *Human Mutation* 2020;41(3):543-80.
7. Vincent LM, Adams D, Hess RA, Ziegler SG, Tsilou E, Golas G *et al.* Hermansky–Pudlak syndrome type 1 in patients of Indian descent. *Mole Genet Metab* 2009;97(3):227-233.
8. Panchadhyayee P, Saha A, Saha K, Ta RK, Barma P. Hermansky-Pudlak syndrome. *Muller J Med Sci Res* 2014;5(1):74.
9. Sharma D, Jhingta PK, Bhardwaj VK, Rasila D, Negi N, Vaid S. Oculocutaneous albinism and the oral manifestation in a 14-year-old boy - A case report and review of literature. *J Dent Allied Sci* 2017;6:93-7
10. Harrison C, Khair K, Baxter B, Russell-Eggitt I, Hann I, Liesner R. Hermansky–Pudlak syndrome: infrequent bleeding and first report of Turkish and Pakistani kindreds. *Arch Dis Child* 2002 1;86(4):297-301.
11. Wijermans PW, van Dorp DB. Hermansky-Pudlak syndrome: correction of bleeding time by 1-desamino-8D-arginine vasopressin. *Am J Hematol* 1989;30:154-157.

Author for Correspondence:
Dr. Mathangi Kumar, Department of Oral Medicine and Radiology, Manipal College of Dental Sciences, Manipal, Manipal Academy of Higher Education, Manipal –576104, Karnataka Email: mathangi.kumar@manipal.edu Cell: 7892221629

How to cite this article:
Chhaparwal Y, Kumar M, Chhaparwal S. Hermansky Pudlak Syndrome Type 2: A Rare Case Report. *J Krishna Inst Med Sci Univ* 2020; 9(3):97-101

Submitted: *04-Oct-2019* ***Accepted:*** *03-Mar-2020* ***Published:*** *01-July-2020*