Case Report

Hermansky–Pudlak Syndrome a Case Report

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Abstract

We report a case of Hermansky –Pudlak syndrome in a 34 year old lady who was born of non consanguineous marriage The incidence of HPS is highest in Puerto Rico.There are increasing reports of HPS among Indians. There are only rare case reports where the mode of inheritance is not autosomal recessive.

Keywords: Dyspnea, Pulmonary Fibrosis, Oculocutanoues Albinism

Introduction

Hermansky-Pudlak syndrome is an autosomal recessive genetic disorder characterized by oculocutaneous albinism, platelet dysfunction. Most of the patients in whom the disorder has been identified and characterized are of Puerto Rican descent. It is possible that HPS in India is under-diagnosed. There have been case reports of this disease in people of Indian origin of the eight subtypes HPS -1 was identified in the patients of Indian origin.

Case Report

A 34 year old lady presented with sudden onset of breathlessness of one week duration. There was no associated cough, fever, palpitation, chest pain. She had occasional episodes of epistaxis and bleeding from the skin. She was born of non consanguineous marriage. On examination she had oculocutaneous albinism, cutaneous telengiectasias and nystagmus. Her blood pressure was 120/70 mm of Hg and her pulse rate was 92/minute. Her respiratory rate was 36/minute and she was cyanosed. Cardiovascular system was normal. Auscultation of chest showed few scattered crepitations. Investigations showed normal hemogram, blood sugar, renal and liver function tests. X-ray chest and echocardiogram was also normal. CT scan chest showed evidence of diffuse pulmonary fibrosis. In view of the oculocutaneous albinism, nystagmus and CT chest showing diffuse pulmonary fibrosis, a diagnosis of Hermansky Pudlak syndrome was entertained. She was started on steroids and pirfenidone and antibiotics. Steroids was initially given in a dose of 40 mg/day and then gradually tapered. Pirfenidone was initially given 276 mg one tab three times daily and plan to increase it to three tabs tid. She improved and was put on continuous pirfenidone treatment.

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Discussion

Hermansky Pudlak syndrome (HPS) is a rare autosomal recessive disorder, characterized by oculocutaneous albinism, bleeding diathesis due to platelet dysfunction, pulmonary fibrosis, granulomatous colitis, cardiomyopathy and renal failure. The global incidence of HPS is 1 in 500,000-10, 00,000 [1]. HPS is now being increasingly recognized in people of Indian descent, as well as in countries like Pakistan, Holland and Japan [2]. There are case reports of HPS in children born of unrelated parents [3].

Of the eight known subtypes only HPS -1 and HPS-4 develop pulmonary fibrosis [4]. Pulmonary fibrosis manifests in the 4th and 5th decades of life. Pulmonary fibrosis is due to deposition of ceroid like substance in liposomes of reticuloendothelial cells, bone marrow and lung macrophages [5]. Pulmonary fibrosis contributes significantly to the mortality of these patients. Average life span is around 30-50 years. The definitive treatment for pulmonary fibrosis in HPS is lung transplantation. Corticosteroids are ineffective. Pirfenidone, an antifibrotic agent has been shown to be effective in

slowing down the progression of fibrosis in type 4 HPS [6]. Other aspects of treatment include avoidance of smoking, early treatment of respiratory infections and regular pneumococcal and influenza vaccination.

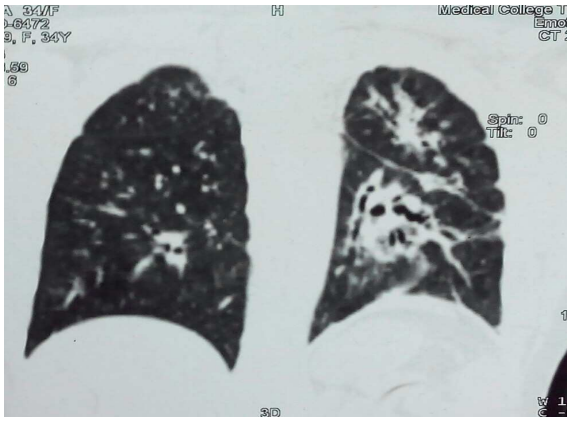

Fig 1: CT scan chest showed evidence of diffuse pulmonary fibrosis

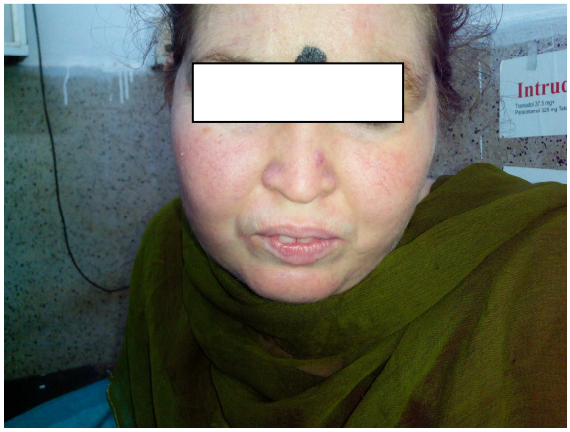

Fig 2: oculocutaneous albinism

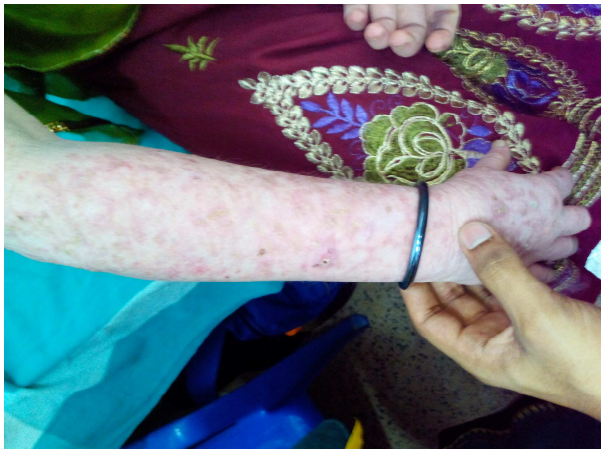

Fig 3: Cutaneous albinism

Conclusion

It is possible that HPS in India is under-diagnosed. High degree of suspicion in patients with clinical symptoms with laboratory nvestigation is important for diagnosis.

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References

1. Pagon RA, Bird TD, Dolan CR, et al., editors. GeneReviews Seattle (WA):University of Washington, Seattle; 1993-2000.

2. Vincent LM, Adams D, Hess RA, Ziegler SG, Tsilou E, Golas G, O'Brien KJ, White JG, Huizing M, Gahl WA. Hermansky-Pudlak syndrome type 1 in patients of Indian descent.Mol Genet Metab. 2009 Jul;97(3):227-33.

3. 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;86(4):297-301.

4. Hermansky-Pudlak Syndrome Type 1 in Patients of Indian Descent Lisa M. Vincent,David Adams, Richard A. Hess, Shira G. Ziegler, Ekaterini Tsilou, Gretchen Golas,Kevin J. O'Brien, James G. White, Marjan Huizing, and William A. Gahl Mol Genet Metab. 2009; 97(3): 227–233.

5. Di Pietro SM, Falcón-Pérez JM, Tenza D, Setty SR, Marks MS, Raposo G, Dell'Angelica EC BLOC-1 interacts with BLOC-2 and the AP-3 complex to

facilitate protein trafficking on endosomes.Mol Biol Cell. 2006;17(9):4027-38. Epub 2006 Jul 12.

6. Hermansky Pudlak syndrome type 4 with interstitial pneumonia Yoshihiko Saketa Kodai Kawamura,Kazuya Ichika Resp Medicine Case Reports 2013;9:38-41.

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