Familial Case of Piebaldism

Ailesel bir Piebaldizm Olgusu

Bir Piebaldizm Olgusu / A Case of Piebaldism

Ersin Aydin[1], Bilal Dogan[2], Ozlem Karabudak Abuaf[2]
[1]Kasimpasa Military Hospital Department of Dermatovenerology,
[2]GATA Haydarpasa Teaching Hospital Department of Dermatovenerology, Istanbul, Türkiye

Previously presented in 22nd World Congress of Dermatology, May 24–29, 2011, Seoul, Korea.

Özet

Piebaldizm, KIT gen mutasyonu sonucu melanoblastların farklılaşması ve deriye göçündeki bozukluktan kaynaklanan, otozomal dominant kalıtılan, ender görülen bir hastalıktır. Frontal bölgedeki saçlarda beyazlama (Beyaz perçem) ve simetrik yerleşen hipo veya depigmente yamalarla karakterizedir. Biz burada saçında beyaz perçem, vücudunda simetrik beyaz yamalar olan, iki kızkardeşinde benzer yakınmalar bulunan bir olguyu sunuyoruz.

Anahtar Kelimeler

Piebaldizm; Beyaz Perçem; Ailesel Piebaldizm

Abstract

Piebaldism is a rare, autosomal dominant disease resulting from mutation in the KIT gene which affects differentiation and migration of melanoblasts. It is characterized by white forelock on central frontal scalp and symmetrical stable hypopigmented or depigmented macules. We report here in a 20-year old male patient who has symmetric white patches on his skin and white hair from birth with similar complaints in his two sisters.

Keywords

Piebaldism; White Forelock; Familial Case of Piebaldism

DOI: 10.4328/JCAM.2175 Received: 21.11.2013 Accepted: 03.12.2013 Printed: 01.09.2013 J Clin Anal Med 2013;4(suppl 1): 90-2
Corresponding Author: Ersin Aydin, Kasimpasa Military Hospital, 34440, Istanbul, Turkey.
T.: +90555562862 E-Mail: drersinaydin@yahoo.com

Introduction

Piebaldism is a rare, autosomal dominant disease characterized by the congenital absence of melanocytes in affected areas of the skin and hair, results in mutations of the KIT gene on chromosome 4q12, which affects the differentiation and migration of melanoblasts from the neural crest during the embryonic life [1]. The skin depigmentation is very characteristic for the disease. Affected individuals always present at birth, white patches are characterized by rigorous symmetry and affect mostly the face, anterior portion of thorax and abdomen, arms, forearms, legs, and thighs [2]. Piebaldism is a permanent condition which mostly has a static course and it may be a socially disabling manifestation for the patient.

Case Report

A 20-year-old male patient referred to our outpatient clinic for white color change in his scalp and face. On dermatological examination, there were a white triangular forelock extending from vertex to frontal scalp and a white plaque on the center of forehead including medial eyebrows (Figure 1). Irregular, sharply demarcated, symmetrical, and wide depigmented patches including some repigmentation islets on front of trunk, neck, arms and legs have also been determined (Figure 2,3). Hands, feet and shoulders were not affected. Family history showed that his two sisters also had similar lesions on their scalp and trunk. Routine laboratory examinations including complete blood count, liver and renal function tests were within normal ranges. In histopathological examination of lesional skin, any melanocytes weren't detected as compatible with piebaldism. There weren't any abnormalities noted in hearing tests and visual examinations which can be associated with piebaldism. The patient was diagnosed as familial piebaldism with these findings.



Figure 1. Irregular, sharply demarcated, symmetrical, and wide depigmented patches neck, front of trunk and extremities. Note the islands of hyperpigmented macules within depigmentation.



Figure 2. Depigmented patches over the extremities



Figure 3. White triangular forelock extending from vertex to frontal scalp and a white patches on the center of forehead. Also note the whitening of hair of the medial eyebrows.

Discussion

First descriptions for piebaldism date back to early Egyptian, Greek and Roman writings [3]. The disease gene causing piebaldism was firstly identified by Giebel and Spritz in a large family in 1991 [4]. Some recent studies have shown that it was mostly caused by mutations in the KIT gene which encodes the cell-surface receptor transmembrane tyrosine kinase for the steel factor [5]. To date, more than 60 mutations of the KIT gene have been reported in piebaldism.

Clinical manifestations of piebaldism strongly correlate with the site of the mutation of the KIT gene. Both males and females are equally affected, and no race is spared [6]. As well as a white forelock of hair may be the only manifestation in 80–90% of patients, both the hair and the underlying forehead, eyebrows, eyelashes, and trunk may be affected as in our case [7]. Depigmented macules are rectangular or irregular in shape and generally have a symmetrical distribution. Characteristically islands of hyperpigmentation are present within and at the border of depigmented areas like our case [6]. Although the disease has a stabile course, a few cases progress or recover partially or completely, especially after injury have also been reported [8].

A number of syndromes like Waardenburg syndrome, Albinism-Deafness syndrome, Tietz syndrome which don't show kit gene mutation but characterized by piebald-like hypopigmentation of skin and hair have also been described [2]. No abnormalities have been found in hearing tests and visual examinations of our case for screening these syndromes.

Piebaldism should be differentiated from some other conditions characterized by depigmentation of hair and skin such as segmental vitiligo and albinism. These diseases in the differential diagnosis of piebaldism are easily excluded. Segmental vitiligo may rarely be present at birth, but is generally acquired later in life and has linear, unilateral distribution. It is not genetically inherited, although it may run in some families. Albinism is a congenital genetically inherited disorder, but is characterized by partial or complete absence of melanin production in skin, hair and eyes, generalized distribution, diffuse hypo/depigmentation. Histopathologically, melanocytes are significantly reduced or totally absent in lesional skin as in our case.

There is no effective treatment method which will provide an optimal repigmentation of the lesions. Sun screens and cosmetic camouflage products are recommended to protect exposed amelanotic areas from sunburns. Use of autologous minigrafts and autologous cultured melanocytes were found potentially useful [9]. Phototherapy alone has a little effect but was found to be effective after the melanocyte transplantation. Surgical procedures are not practically acceptable in patients having wide depigmented areas as in our case because of requiring multiple donor areas and risk of scarring. We recommended our patient to avoid sun exposure and use sunscreens because he didn't accept the other treatment options.

Competing interests

The authors declare that they have no competing interests.

References

1. Agarwal S, Ojha A. Piebaldism: A brief report and review of the literature. Indian Dermatol Online J 2012;3(2):144-7.
2. Thomas I, Kihiczak GG, Fox MD, Janniger CK, Schwartz RA. Piebaldism: an update. Int J Dermatol 2004;43(10):716-9.
3. Spritz RA. The molecular basis of human piebaldism. Pigment Cell Res 1992;5(5 Pt 2):340-3.
4. Giebel LB, Spritz RA. Mutation of the KIT (mast/stem cell growth factor receptor) protooncogene in human piebaldism. Proc Natl Acad Sci U S A 1991;88:(19)8696-9.
5. Wen GD, Zhou C, Yu C, DU J, Xu QX, Liu ZY et al. A novel mutation of the KIT gene in a Chinese family with piebaldism. Chin Med J (Engl). 2013;126(12):2325-8.
6. Ortonne JP, Bahaderan P, Fitzpatric TB, Mosher DB, Hori Y. Hypomelanosis and Hypermelanosis. In: Fitzpatric TB, Freedberg IM, Eisen AZ, Wolff K, Austen KF, Goldsmith LA, et al. editors. Dermatology in General Medicine. 6th ed. New York: McGraw-Hill; 2003. p. 836–81.
7. Ward KA, Moss C, Sanders DS. Human piebaldism: relationship between phenotype and site of kit gene mutation. Br J Dermatol 1995;132:929–935.
8. Matsunaga H, Tanioka M, Utani A, Miyachi Y. Familial case of piebaldism with regression of white forelock. Clin Exp Dermatol 2008;33(4):511-2.
9. Neves DR, Régis Júnior JR, Oliveira PJ, Zac RI, Silveira Kde S. Melanocyte transplant in piebaldism: case report. An Bras Dermatol 2010;85(3):384-8.

How to cite this article:
Aydin E, Dogan B, Abuaf ÖK. Familial Case of Piebaldism. J Clin Anal Med 2013;4(suppl 1): 90-2.