CASE REPORT

Piebaldism and Vitiligo in Two Brothers

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Abstract

Piebaldism is an autosomal dominant uncommon (<1 in 20,000) congenital pigmentary disorder. Depigmented patches are present since birth. They usually remain unchanged throughout life. Vitiligo is its closest differential diagnosis. We report a unique family in which these two dissimilar depigmentations, i.e. piebaldism and vitiligo (with nevus depigmentosus), were noted in two brothers. To the best of our knowledge, this is the first report of this presentation in the literature. *(Iran J Dermatol 2009;12 (Suppl): S8-S11)*

Keywords: piebaldism, vitiligo, depigmentation

Introduction

The name 'Piebaldism' is derived from a combination of the 'pie' in the magpie (a bird of black and white plumage) and the 'bald' of the bald eagle (the US national bird with a white feathered head). The classical pattern of piebaldism includes a frontal, median or paramedian triangular or diamond patch associated with a mesh of white hair (forelock) in 80-90% of the cases. Piebaldism is an autosomal dominant uncommon (<1 in 20,000) congenital disorder produced as a result of inactivating mutations or deletions of the KIT (receptor tyrosine kinase) or SLUG (zinc finger neural crest transcriptional factor) genes resulting in decreased receptor tyrosine kinase signalling, impaired melanoblast development, and a decreased melanogenesis [1,2,3]. Patches of skin totally devoid of pigment are present at birth and usually remain unchanged throughout life. In a case of piebaldism, vitiligo is the major differential diagnosis that must be excluded (Table 1). We report two brothers of a family affected with dissimilar depigmentations: piebaldism in the elder and vitiligo with nevus depigmentosus in the younger brother.

Case Report

A 19-year-old male presented with depigmented patches on anterior abdomen (extensively involved), mid arms and legs (moderately involved). He had a depigmented patch on his central forehead (extending to scalp) along with a tuft of white hair (forelock) (Figure 1). All lesions were present since birth. Hyperpigmented macules were present in areas lacking pigmentation (Figure 2). Hairs were white over the depigmented lesions. He was diagnosed with piebaldism. Similar features of piebaldism were noted in four generations comprising of ten affected members. Neither of them had involvement of the back. All family members gave the history of partial resolution of the white lesions of their foreheads, hands, and legs with aging.

The youngest member of the family was a 12-year-old male who did not have any lesions of piebaldism since birth. He had a depigmented linear lesion on his anterior right lower abdomen, extending from the lower half of the umbilicus towards his back. It was noted in early childhood and was diagnosed as nevus depigmentosus. Depigmented patches (with no normal skin in between) were noted on bilateral postauricular regions and on his left leg (circular or oval shaped) since last year (Figure 2,3). Late appearance of these lesions, absence of normal skin in between and typical periorificial distribution around the ears suggested the diagnosis of vitiligo. His mother (forty eight years old) also had a lesion of vitiligo for the last eighteen years. Table 2 describes this family. All the diagnoses were made clinically as the

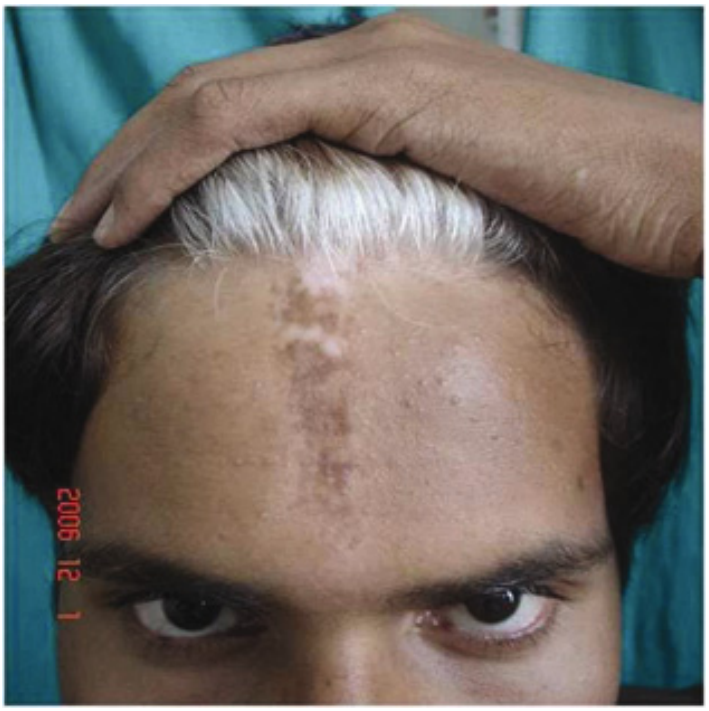

Figure 1. *Classical forelock in the elder brother*

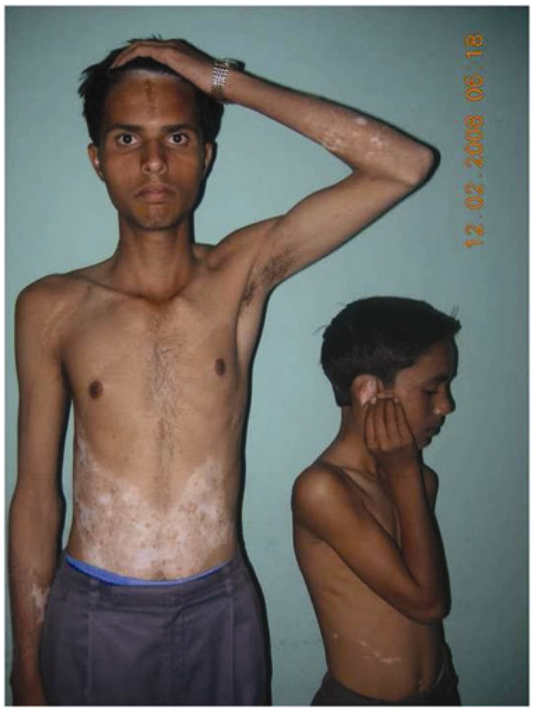

Figure 2. *Piebaldism and vitiligo affecting two brothers.*

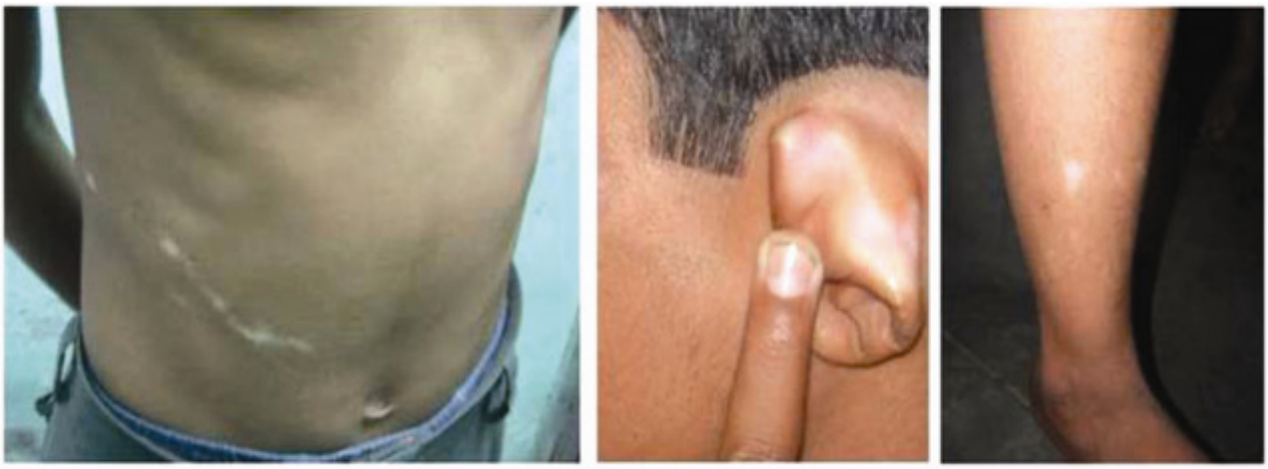

Figure 3. *Younger brother exhibiting nevus depigmentosus on his abdomen and vitiligo lesions on postauricular areas and right leg.*

advantage of genetic study was not available in our set up.

Discussion

In piebaldism, the involved areas are as follows: anterior abdomen extending to chest, lateral trunk sparing dorsal spine, mid arms and legs sparing hands, feet. The periorificial areas (the characteristic site of vitiligo) are not generally involved. The presence of normal or hyperpigmented macules within amelanotic patches is an important characteristic. Skin biopsy from white areas in piebaldism reveals the absence of melanocytes.

Typical vitiligo macules may be peiorificial and involve the skin around the eyes, ears, nose etc.

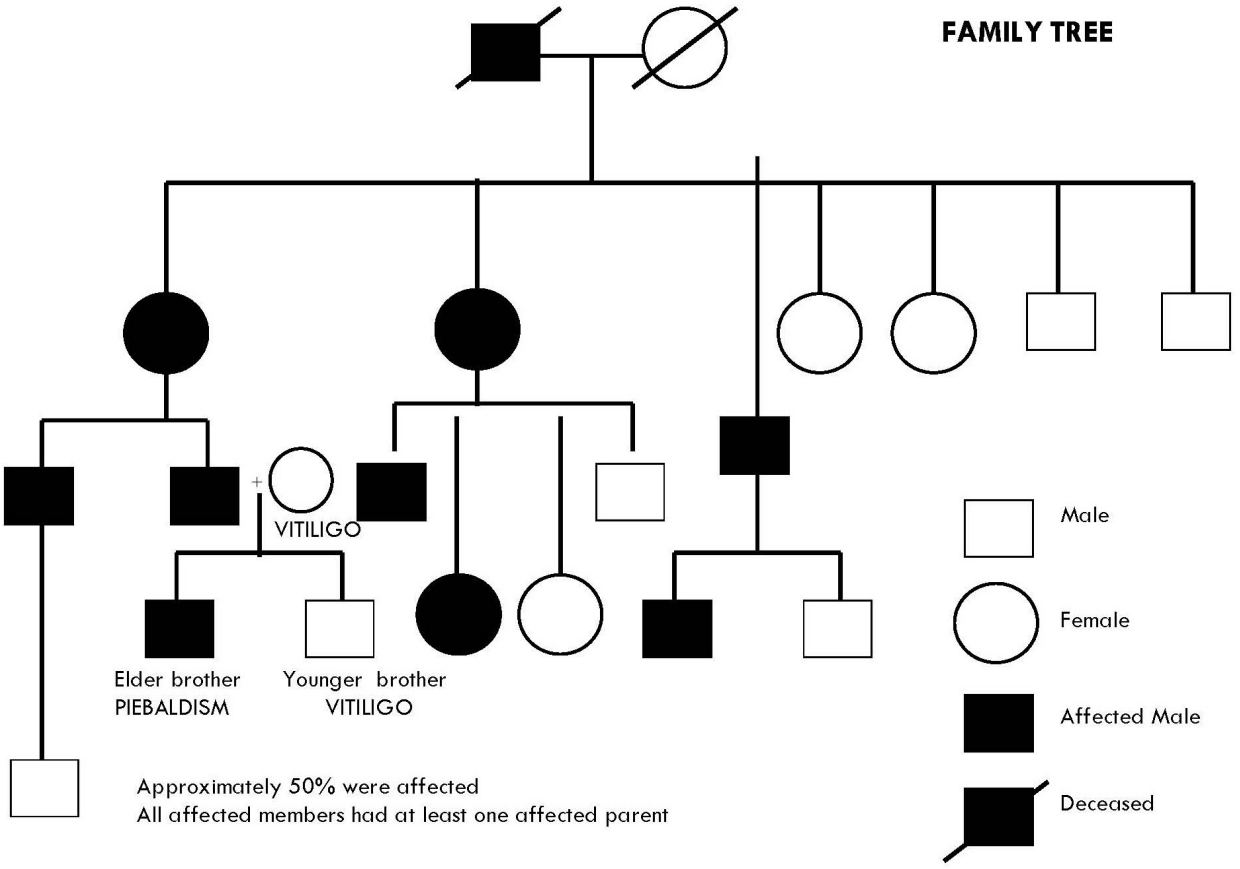

Figure 4. *Family tree of reported patients*

Piebaldism is one of the cutaneous signs of Waardenburg syndrome, along with heterochromia of the irides, lateral displacement of inner canthi and deafness [4]. Deafness and piebaldism without other features of Waardenburg's syndrome is known as Woolfe's syndrome [5]. So, audiometry should be considered in all cases of piebaldism. Naevus depigmentosus is usually a single lesion although it can be large and assumes a dermatomal distribution, usually located on trunk or proximal extremities. It may be congenital or may become prominent during first few years of life as normal pigmentation darkens.

In piebaldism, depigmentation is generally static in nature. But progression of white areas as well as spontaneous partial repigmentation [6,7] are described. Two cases of piebaldism have been described in literature where both the mother and the daughter had a novel Val620Ala mutation in their KIT gene and showed progressive depigmentation [8].

Several case reports have been published claiming a non-random, causal relationship between piebaldism and neurofibromatosis type 1(NF1) [9,10]. The diagnosis was based on two established NF1 diagnostic criteria of six or more café-au-lait spots and axillary or inguinal freckling [11] and none of these patients had any of the nonpigmentary features of NF1 such as cutaneous neurofibromas, etc. Spritz et al, argued that café-au-lait spots and axillary or inguinal freckling are common features of piebaldism. So, they were actually cases of piebaldism who were misdiagnosed as NF1 [12].

We report these two brothers affected with two dissimilar pigmentary disorders. The elder brother had piebaldism while its closest differential diagnosis, vitiligo, was noted in the younger sibling. To the best of our knowledge, such a unique presentation has not been reported before.

Table 1. *Difference between vitiligo and piebaldism*

	VITILIGO	**PIEBALDISM**
AGE	Birth to old age; half by 20 years; rarely congenital	Birth
COURSE	Chronic progressive; occasional limited improvement	Chronic stable (most cases)
SIZE/SHAPE	Mm to cm. Round , scalloped margin	Few to several cm; irregular
DISTRIBUTION	Symmetric, periorificial; Extensor areas of limbs and digits	Central forehead with forelock; mid trunk, sparing dorsal spine; mid arm/ leg sparing hands or feet
SPECIAL FEATURE	Trichrome; occasional segmental pattern	Hyperpigmented macules with in white patches or on normal skin

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