

## Piebaldism: A Brief Report

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### Abstract

Piebaldism is an autosomal dominant congenital pigmentary disorder. It is characterised by white forelock and multiple symmetrical stable depigmented macules. We report a familial case of piebaldism affecting a 6 year old female and her father.

**Keywords:** Piebaldism; Autosomal dominant; Pigmentary disorder; Vitiligo

### Introduction

The word Piebaldism is derived from combination of two words Magpie (pie) and Bald eagle (bald) [1]. It is a rare autosomal dominant pigmentary disorder characterised by complete absence of melanocytes in affected areas of skin and hair [2]. Its prevalence is less than one per 20000 individuals. Characteristic triangular or diamond shaped depigmented macule in midline of forehead along with white forelock is present in affected individuals in majority of cases (90%). There is usually symmetrical involvement of ventral surface of trunk and middle part of extremities. These lesions are persistent and have a static course with characteristic distribution. We report this case in which multiple members of the same family were affected with variable expression of the disease.

### Case Report

A 6 year old female was referred from Paediatrics department to us with complaint of white patches over her abdomen and forehead. These lesions were present since birth and were asymptomatic and stable in nature. Examination of the child revealed presence of bilaterally symmetrical depigmented macule on her legs (Figure 1 and 2). It was approximately 5 to 7 cm in size and was of irregular shape. A well-defined white forelock in mid frontal region with underlying depigmented rhomboid macule on forehead was also present. Trunk, hands, feet, mucosa along with eyelashes and eyebrows were found to be spared. There was no associated ocular, auditory or neurological defect. All her development milestones and routine biochemical tests were normal. We advised for a biopsy from the lesion but her father refused. It was also found out that she was being treated as a case of vitiligo. Further evaluation revealed a similar picture of depigmentation on forehead with white forelock in her father (Figure 3). Strong family history with similar complaints was present in 3 generations comprising 12 members (Figure 4). There was history of spontaneous regression/



**Figure 1:** Examination of the child revealed presence of bilaterally symmetrical depigmented macule on her legs.



**Figure 2:** Examination of the child revealed presence of bilaterally symmetrical depigmented macule on her legs.



**Figure 3:** Depigmentation on forehead with white forelock in her father.

repigmentation of lesions in some members of her family including her father. No history of consanguinity was present. Considering the strong prevalence in family history and characteristic features, the patient was diagnosed as a case of piebaldism.

### Discussion

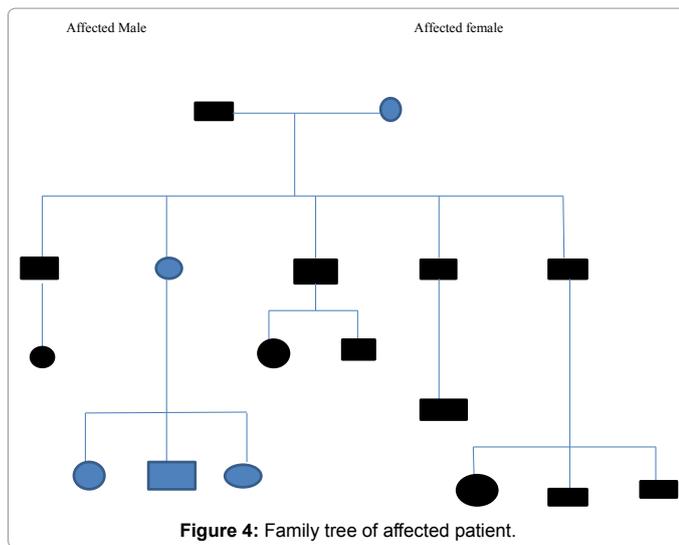
Piebaldism is a rare pigmentary disorder inherited as autosomal dominant condition with equal predisposition in males and females affecting all races. It is linked to mutation in C-KIT proto-oncogene

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**Received:** September 03, 2015; **Accepted:** January 30, 2016; **Published:** February 04, 2016

**Citation:** Kaushik M, Gupta S, Mahendra A (2016) Piebaldism: A Brief Report. Dermatol Case Rep 1: 103.

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mapped to chromosome 4q11/4q12. This gene is responsible for cell surface receptor transmembrane tyrosine kinase for stem cell/mast cell growth factor [3]. There may be Zinc finger neural crest mutation instead. These mutations cause aberrant melanocyte proliferation, survival or migration during embryonic development. It is also associated with hirschsprung disease, neurofibromatosis, congenital dyserythropoietic anaemia, diamond blackfan anaemia or Grover's disease [3]. Piebaldism is characterised by white forelock on white triangular elongated or diamond shaped macule on forehead since birth. Eyebrows and eyelashes may also be involved along with abdominal wall, anterior chest, back, mid-thighs and occasionally lower part of neck, face, middle upper arm and wrist. Islets of hyperpigmented macules in the leukodermic patches and on normally colored skin are other characteristic features. Many members of the same family may be affected with variable expression of disease. Also piebaldism is a cutaneous sign of Waardenburg syndrome along with heterochromia of irides, lateral displacement of inner canthi and deafness. As reported in this case, an elucidative history and examination is very important in diagnosis of piebaldism keeping in mind vitiligo and albinism as an important depigmentary disorder. Vitiligo generally occurs later in life (around 20 years) and the configuration and distribution of lesions is quite different. Hypomelanotic macules are usually first noted on the sun exposed areas of skin, on the face (around ears, eyes or nose) or on dorsum of hands. The macules have a convex outline, increase irregularly in size and fuse with neighbouring lesions to form complex patterns. Melanocytes are destroyed in vitiligo while in piebaldism, they are missing since birth. Also vitiliginous lesion might repigment with characteristic perifollicular pigmentation. Inheritance pattern in

vitiligo is polygenic or autosomal dominant with variable penetrance. Albinism on the other hand is an autosomal recessive disease presenting with widespread hypomelanotic skin due to defective melanin production. It lacks the characteristic hyperpigmented macules within hypopigmented areas of piebaldism. There are also associated hair and eyes anomalies in albinism. Persons with vitiligo or piebaldism usually have normal vision. In this condition, melanocytes are present but there is complete or partial defect in melanin synthesis. Several case reports have been published claiming a nonrandom, casual relationship between piebaldism and neurofibromatosis type 1. An association of piebaldism and vitiligo in two brothers has been reported in Indian subcontinent. However no exact pattern of spread of piebaldism among many family members of same generation could be concluded.

## Management

Treatment of piebaldism is quite challenging with varying results. Sunscreens should be used to protect amelanotic areas from sunburn [4]. Artificial pigmentary agents can also be used. Medical management has been found to be unsatisfactory. The condition can be surgically treated with split thickness skin grafting, punch grafting or suction blister epidermal grafting. Besides, autologous melanocyte transplant after culturing of melanocytes and keratinocytes also been tried with encouraging results. After these procedures, phototherapy should be used for accelerated pigmentation [5]. Treatment with combination of dermaabrasion and grafting or a combination of Erbium:YAG laser surgery grafting on recipient bed too have been reported [6-9].

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