

PIEBALDISM: A CASE REPORT

Trinh Ngoc Phat¹

SUMMARY

A 7-year-old female patient presented with leukoderma patches and poliosis on forehead and both legs at birth. The lesions increased in size gradually in proportion to the patient's growth without spreading out. There are no symptoms. The child's mental and motor developments were completely normal. The physical examination did not reveal any other abnormalities. No one in the patient's family or pedigree had the same manifestations.

Keywords: *Piebaldism, leukoderma patch, poliosis, forelock*

INTRODUCTION

Piebaldism is a rare inherited disorder of pigmentation with an estimated incidence of less than 1 in 20000 individuals¹. It is characterised by isolated leukoderma patches (white skin) and poliosis (white hair) present at birth that was caused by mutations of the KIT proto-oncogene account for almost cases^{1,2,3}. The lesions usually remain unchanged throughout life. Management includes sun protection, patient education and repigmentation treatments^{1,2,3}.

CASE REPORT

A 7-year-old female patient presented with leukoderma patches and poliosis on forehead and both legs at birth. The lesions increased in size gradually in proportion to the patient's growth without spreading out. Patient and her parent did not report any symptom.

Physical examination revealed triangular leukoderma patch on central parietal-frontal area with forelock and symmetrical large leukoderma patches with poliosis that extending from the knee to the lower leg of the both sides. The boundary of the lesion is quite well-defined. No precancerous lesions were detected in the depigmented

areas. No other mucosal and skin abnormalities were detected. The child's mental and motor developments were completely normal. The physical examination of other systems did not reveal any other abnormalities.

She is the only child in the family and no one in the her family or pedigree have the same manifestations.

Her parent refused to have a skin biopsy so the histopathological result was not obtained



Figure 1. Triangular leukoderma patch on central parietal-frontal area with forelock in a 7-year-old female patient. (Source: Trinh Ngoc Phat)

¹: Hanoi Medical University



Figure 2. Symmetrical large leukoderma patches with poliosis that extending from the knee to the lower leg of the both sides in a 7-year-old female patient. (Source: Trinh Ngoc Phat)

DISCUSSION

The name piebaldism is derived from a combination of the “pie” as in the magpie (a bird of black and white plumage) and the “bald” of the bald eagle (the US national bird that has a white feathered head). Hence the major characteristic of piebaldism is a white forelock (a patch of white hair directly above the forehead)^{3,4}.

Piebaldism is a rare autosomal dominant genetic condition with an estimated incidence

of less than 1:20000¹. Both males and females are equally affected, and no race is spared^{1,2,5}.

The most common cause of piebaldism is a mutation in the KIT proto-oncogene on chromosome 4 account for 75% of cases^{1,5,6}. During embryogenesis, the KIT ligand produced within a dermatomyotome binds to the KIT receptor (a tyrosine kinase receptor) on melanoblasts and melanocytes, allowing for survival and migration along a given dermatomyotome⁸. Mutations in KIT within the 4q12 locus lead to abnormal melanocyte migration and the absence of melanocytes in the ventral midline of the epidermis. This absence leads to the characteristic central leukoderma and poliosis seen in piebaldism. The site of mutation within the KIT gene correlates with the phenotypic severity of the disease^{1,5,6,7,8}. Additionally, mutations in SNAI2 within the 8q11.21 locus, a transcription repressor of KIT and E-cadherin important in maintaining melanoblast homeostasis, have also been implicated in cases of piebaldism.^{6,7} This patient is the only member of the pedigree that developed the disease, so it seems that the mutation in the patient was a random mutation that had just arisen.

Affected individuals present at birth with a relatively stable and persistent depigmentation of the hair and skin, although in a number of patients, repigmentation may occur spontaneously, either partially or completely, especially after injury^{1,2,4,5,7}. A white forelock of hair arising from a triangular, elongated or diamond-shaped, midline, depigmented macule on the forehead may be the only manifestation in 80 - 90% of cases. Eyebrows and eyelashes may also be affected. The characteristic distribution of depigmented macules includes central macule on forehead with white forelock, anterior abdomen extending

to the chest, the lateral trunk sparing the dorsal spine, the mid-arms and legs sparing the hands, and feet. Depigmented macules are rectangular, rhomboid or irregular in shape and usually have a symmetrical distribution. Typically, islands of hyperpigmentation are present within and at the border of depigmented areas. The mild form may present with only small patches of leukoderma, whereas in more severe forms have the white forelock and larger white patches over the trunk and limbs^{1,2,4,6,8,9}. Café-au-lait macules and flexural freckling may develop and piebaldism is also one of the skin manifestation of Waardenburg syndrome^{1,2,10}. Clinical manifestation of this patient is very typical and have moderate severity^{1,2,8,9}.

The diagnosis is usually made by clinical findings but a biopsy can help in equivocal cases. Skin biopsy from the leukoderma in patients with piebaldism demonstrates complete lack of melanocytes and melanin pigment. Genetic testing on peripheral blood can confirm the diagnosis^{1,3,4,9}.

The management of piebaldism includes education about sun protection and self

skin examination to detect skin cancer and repigmentation treatments. Repigmentation treatments include dermabrasion of areas of depigmentation followed by the application of melanocyte-enriched cell suspensions, melanocyte transplant by shaving off the top layer of skin (epidermis) and replacing it by a shave of skin from a pigmented site, suction epidermal grafting or full-thickness punch grafts, a combination of erbium:YAG laser and autologous cultured epidermis^{1,2,6,8,11}. A combination of these methods may be required and can be augmented by the addition of UV light therapy. Cosmetic camouflage techniques can cover the pigment changes of hair and skin.

Individuals with piebaldism usually have stable leukoderma and poliosis. Unlike vitiligo, these areas of depigmentation do not progress after birth. However, the leukoderma may be progressive in rare cases. There are reports of rare cases of spontaneous repigmentation at the margins or within the patches of leukoderma in the form of dots and macules^{1,2,3,4,9,10}.

REFERENCES

1. Shah M, Patton E, Zedek D. Piebaldism. [Updated 2022 Apr 14]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan-. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK544238/>
2. Agarwal S, Ojha A. Piebaldism: A brief report and review of the literature. *Indian Dermatol Online J.* 2012 May;3(2):144-7.
3. Sewon Kang et al, 2019. Piebaldism, Melanocytic Disorders, Fitzpatrick's Dermatology, Mc GrawHill, volume 1, 9th edition, 1321-1323.
4. Oiso N, Fukai K, Kawada A, Suzuki T. Piebaldism. *J Dermatol.* 2013 May;40(5):330-5.
5. 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):8696-8699.
6. Sanchez-Martin M, Perez-Losada J, Rodriguez-Garcia A, et al. Deletion of the SLUG (SNAI2) gene results in human piebaldism. *Am J Med Genet A.* 2003; 122a(2):125-132.



7. Hamadah I, Chisti M, Haider M, et al. A novel KIT mutation in a family with expanded syndrome of piebaldism. *JAAD Case Rep.* 2019;5(7):627-31. doi:10.1016/j.jdc.2019.01.021. PubMed Central.
8. Saleem MD. Biology of human melanocyte development, Piebaldism, and Waardenburg syndrome. *Pediatr Dermatol.* 2019 Jan;36(1):72-84.
9. Grob A, Grekin S. Piebaldism in children. *Cutis.* 2016 Feb;97(2):90-2.
10. Akarsu S, İlknur T, Avcı C, Fetil E. Piebaldism associated with café-au-lait macules and intertriginous freckling: a case report and review of the literature. *Ann Dermatol.* 2019;31(5):567-70. doi:10.5021/ad.2019.31.5.567. Journal.
11. Guerra L, Primavera G, Raskovic D, Pellegrini G, Golisano O, Bondanza S, Kuhn S, Piazza P, Luci A, Atzori F, De Luca M. Permanent repigmentation of piebaldism by erbium:YAG laser and autologous cultured epidermis. *Br J Dermatol.* 2004 Apr;150(4):715-21.