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Piebaldism

- A Case Report -

S.-M. Hwang, M.D., S.-K. Ahn, M.D., W.-S. Lee, M.D., M.-S. Yoo, M.D., E.-H. Choi, M.D.

Department of Dermatology, Yonsei University, Wonju College of Medicine

We report a familial case of piebaldism affecting a 33-year-old man and his 3-month-old son. Typical skin findings, white forelock and amelanotic white macules containing hyperpigmented macules, were characteristically presented on both patients.

Key Words : *Piebaldism, White forelock, Hypopigmentation, Hyperpigmentation.*

INTRODUCTION

Piebaldism, previously described as familial white skin spotting with white forelock, poikilochromia alba, congenital vitiligo, leukoderma, achromia, and partial albinism, is an autosomal dominant, stable leukoderma characterized by a white forelock and vitiligo-like amelanotic macules, usually containing a few normally pigmented or hyperpigmented macules. Piebaldism is thought to result from a defective migration of melanoblast or a failure of survival and /or differentiation from melanoblast to melanocytes (Cook, 1952; Hayashibe and Mishima, 1988; Mosher and Fitzpatrick, 1988; Mosher et al., 1993; Dippel et al., 1995 ; Ward et al., 1995). Melanocytes are completely absent from the regions of hypopigmented skin. Here we describe a family with piebaldism and have differentiated it from other diseases with circumscribed hypopigmentary lesions.

CASE REPORT

A 3-month-old male infant was presented with hypopigmented skin lesions on his face, abdomen and thighs visible from birth. On physical examination, there were elongated cigar shaped hypopigmented patches with poliosis on the center of his forehead and symmetric rectangular whitish hypopigmented patches on the abdomen and thighs (Fig. 2 A,C). Routine laboratory examinations including complete blood count, urine analysis, and liver function test were within normal limits. His medical history showed that he was born at full term by normal spontaneous vaginal delivery. His birth weight was 3.95kg. His family history showed that his father had similar skin lesions. But his father's skin lesion was somewhat different. There were scarring changes on his forehead and some hyperpigmented

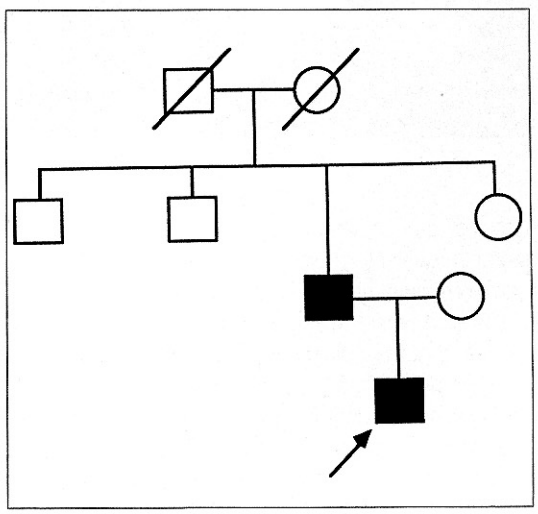

Fig. 1. Familial pedigree. The arrow indicate proband.

Address for correspondence : *Sung-Ku Ahn, M.D., Department of Dermatology, Yonsei University, Wonju College of Medicine, Wonju, Korea, 220-701*
Tel : (0371) 41-1380, Fax : (0371) 48-2650

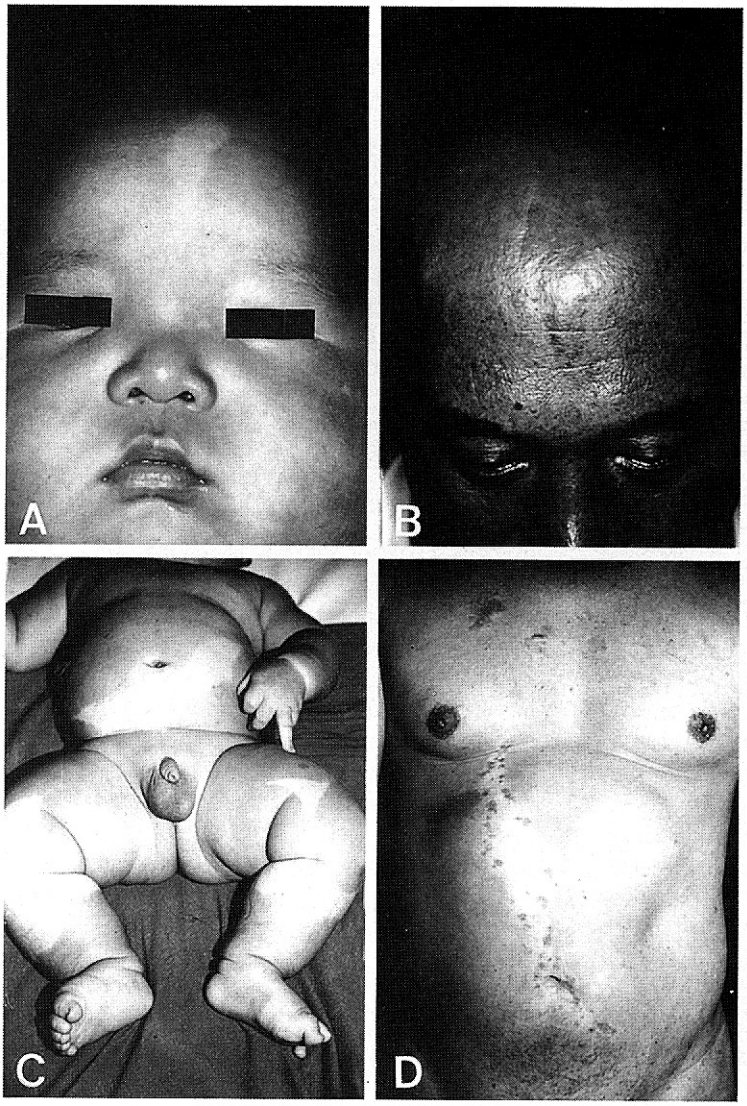

Fig. 2. Clinical features ; (A) white forelock in patient, (B) scarring changes on the forehead of patient's father, (C) vitiligo-like amelanotic macules in patient, (D) in patient's father, there are hyperpigmented macules, embedded and prevalent on the peripheral margins of the hypopigmented patch.

macules, embedded on the hypopigmented patches and/or prevalented on the peripheral margins of hypopigmented patch (Fig. 2 B,D).

The skin biopsy was taken on the hypopigmented patch and normal pigmented skin from the abdomen of his father. Contrary to the normal pigmented skin, the specimen from the hypopigmented patch revealed an absence of melanin pigment and melanocytes by hematoxylin and eosin and Fontana-Masson stain (Fig. 3 A, B). On electron microscopic examination, we could not find melanosomes in the keratinocytes and melanocytes (Fig. 3 C, D).

DISCUSSION

Disorders of hypopigmentation are a major reason for visiting a dermatologist, especially among Asians and Blacks. An exact diagnosis is essential for their prognosis. To make the correct diagnosis require specialized knowledge, a detailed history, careful physical examination and use of special techniques.

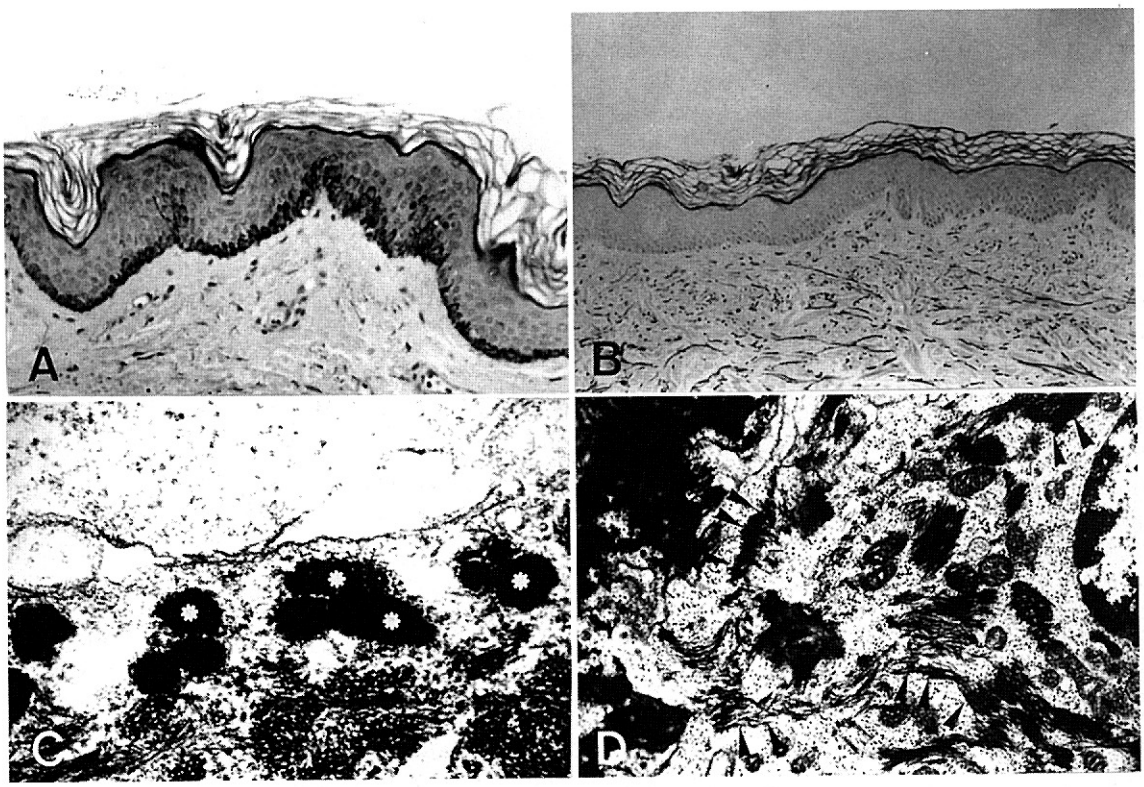

Fig. 3. Histopathologic and electron microscopic findings ; (A,C) specimen from normal pigmented skin, (B,D) specimen from hypopigmented patch. Specimen from hypopigmented patch revealed an absence of melanin pigment and melanocytes. In electron microscopic examination, there are no melanosomes in the keratinocytes and melanocytes. asterisk ; melanosomes, arrow head ; tonofilament (A, B ; Fontanna - Masson stain × 100, C,D ; electron microscopic feature × 17000).

In piebaldism, there are characteristic lesions such as a white forelock, hypopigmented patches symmetrically involving the ventral surface of the trunk and the middle part of the extremities, and hyperpigmented macules and so it can be diagnosed easily. A white forelock occurs in 80 to 90 percent of piebaldism cases and is a triangular, elongated, or diamond-shaped hypopigmented macules in the midline of the forehead. There are white to silvery hairs. Hypopigmented patches are apparent in 10 to 20 percent of patients with piebaldism. These hypopigmented macules are characteristically symmetric involving the anterior abdomen to chest, mid-upper arm to wrist, mid-thigh to mid-calf, but sparing hands, upper arms, shoulders, upper thighs, and mid-calf to feet. In elderly patients with piebaldism, there are other characteristic hyperpigmented macules which are particulary prevalent on the periphery and/or within the hypopigmented patches. Most of them are less than 1 cm in diameter (Cooke, 1952 ; Mosher and Fitzpatrick, 1988 ; Pinto and Bolognia, 1993).

The molecular defect in piebaldism has been identified recently. Human piebaldism was linked to mutations of the kit protooncogene which is mapped to the proximal long arm of chromosome 4 (4q12) (Spritz et al., 1993 ; Spritz, 1994 ; Tomita, 1994). Mutations with the kit protooncogene which encodes the tyrosine kinase transmembrane cellular receptor for mast/stem cell growth factor result in defects in the migration of melanoblast or a failure of survival and/or differentiation from melanoblast to melanocyte on the ventral aspect of the skin (Hayashibe and Mishima, 1988 ; Mosher et al., 1993 ; Dippel et al., 1995).

Peibaldism should be differentiated from circum-

scribed hypomelanosis such as segmental vitiligo, ash-leaf shaped white macules of tuberous sclerosis, nevus depigmentosus, and nevus anemicus. These entities in the differential diagnosis of piebaldism are rather easily excluded: (1) segmental vitiligo, which appears later in life and has a linear, unilateral distribution, and may increase in size and extent (Pinto and Bolognia, 1991; Mosher et al., 1993). (2) ash-leaf shaped white macules of tuberous sclerosis, which occur most commonly on the posterior trunk. The lesions are well circumscribed loss of pigmentation, related to a decrease in size, synthesis, and melanization of melanosome. In addition, multiple neurocutaneous abnormalities such as adenoma sebaceum, ungual fibroma, fibrous plaque on forehead or scalp, retinal hamartoma, cortical tubers, and subependymal glial nodules can be used to make the presumptive clinical diagnosis of tuberous sclerosis (Jimbow et al., 1975; Pinto and Bolognia, 1991; Mosher et al., 1993). (3) nevus depigmentosus, which is a congenital and stable hypopigmentation that has irregular, serrated or geographic margins. There are defects in the transfer of melanosomes from melanocytes to keratinocytes and abnormally aggregated melanosomes (Jimbow et al., 1975; Pinto and Bolognia, 1991; Mosher et al., 1993). (4) nevus anemicus, which is characterized by pale macules and patches of varying sizes and shapes with irregular margins, occurring predominantly on the trunk. There is a localized area of vasoconstriction that is easily differentiated because its border can be obscured by diascopy (Ross et al., 1977; Mountcastle et al., 1986; Pinto and Bolognia, 1991).

Treatment of piebaldism is rarely effective in inducing pigmentation. Photoprotection preparations with sunscreen and cosmetics should be prescribed to obscure and protect the amelanotic areas from burning with sun exposure. Autologous minigrafts, 2 to 4mm punch grafts or use of autologous cultured melanocytes can be potentially helpful (Mosher et al., 1993).

In our patient, it is worth while to notice the scarring change of forehead of the patient's father who complained of a burning sensation with sun exposure. Though induction of pigmentation was disappointing, we would like to stress the need to take a positive attitude toward treatment for protection from sunburn, social embarrassment and cosmetic problems, espcially on the face. Most Asians and Blacks are troubled by pigmentary changes and have more complaints with hypopigmented lesions than hyperpigmented ones. Awareness of characteristic features of piebaldism can lead to the correct diagnosis and treatment and to differentiation from other circumscribed hypopigmentary disorders.

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