A PIEBALD FAMILY

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Piebaldism is an uncommon congenital hypomelanotic disorder characterized by a white forelock and vitiligo like amelanotic macules. We report a family with piebaldism affecting four successive generations. The disease was present in 16 members of the family.

Key Words : Piebaldism, White forelock, Autosomal dominant disorder

Introduction

Piebaldism is an autosomal dominant congenital leucoderma characterized by localized stable hypomelanosis of the skin and hair, and by a characteristic distribution that involves the anterior trunk, extremities, the central portion of eyebrows, and the midfrontal portion of scalp with resultant white forelock.[1] White forelock is the most characteristic feature occurring in 80-90% of piebald individuals.[2] The hypopigmented macules are classically distributed in bilateral pattern.

Melanin and melanocytes are absent from the skin and hair bulbs of involved region.[3] Absence of melanocytes in hypomelanotic areas results from failure of melanoblasts either to migrate into the skin or to differentiate into melanocytes.[1]

Case Reports

A 10-month-old female child was seen with asymptomatic widespread depigmented macules on scalp, trunk and extremities since birth. The patches had remained fixed since their appearance. The examination revealed a well circumscribed white forelock in midfrontal region. Large depigmented macular area was seen on ventral part of mid trunk. Similar large, depigmented macules were present on mid portions of both upper and lower extremities. The entire back, hands and feet, lower portions of forearm and legs and mucosae were completely spared. Discrete, 2-4 mm, skin coloured and hyperpigmented macules were interspersed between the depigmented areas on extremities and trunk. The child had no obvious ocular, hearing or neurological defects. The physical and mental development was normal. There was no history of consanguinity amongst the parents.

Interestingly similar pattern of depigmentation was reported to be present in several members of the family (Fig.1). We could examine 4 of the 16 affected members including father, grandfather and great grand mother of the child. All of them had depigmented lesions and the presence of white forelock in frontal region. All had almost

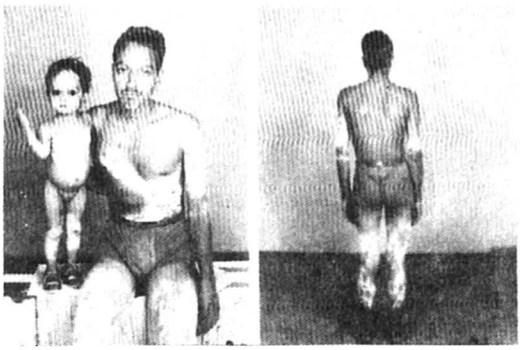

Fig. 1. Pedigree of the family showing piebaldism.

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similar distribution of lesions as in the child. Distal portions of the extremities and back were totally spared. None had mucosal involvement. The lesions in all had remained relatively unchanged throughout their life.

Discussion

The appearance of depigmented macules since birth, presence of white forelock in frontal region, typical distribution of depigmented macules, their relative fixity since the time of their appearance and presence of similar disease in several members of the family was distinctive enough to make a clinical diagnosis of piebaldism. In all out of total 77 members of the family 16 were affected.

In piebaldism, families with affected members in four,[4] five,[5] six[6] or seven[7] generations have been described. The first extensive family study was that of Rizzoli who described 38 piebalds among 111 members of 5 generations of the Bianconini (literally "white forelock") family. In a family claiming descent from Elisabeth Mortimer, a grand daughter of king Edward III, the piebald trait existed for 500 years.[8] The piebaldism trait it transmitted as an autosomal dominant pattern with a very high degree of penetrance.[1]

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