

# Looking through the dermoscope in a case of piebaldism

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Sir,

Piebaldism is an uncommon autosomal dominant disorder characterized by the congenital absence of melanocytes in localised areas of the skin and hair due to *c-kit* gene mutation, which affects the differentiation and migration of melanoblasts from the neural crest during the embryonic life. This condition is clinically characterised by the presence of localized stable hypo to depigmentation 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, which is also the most common and persisting feature [1].

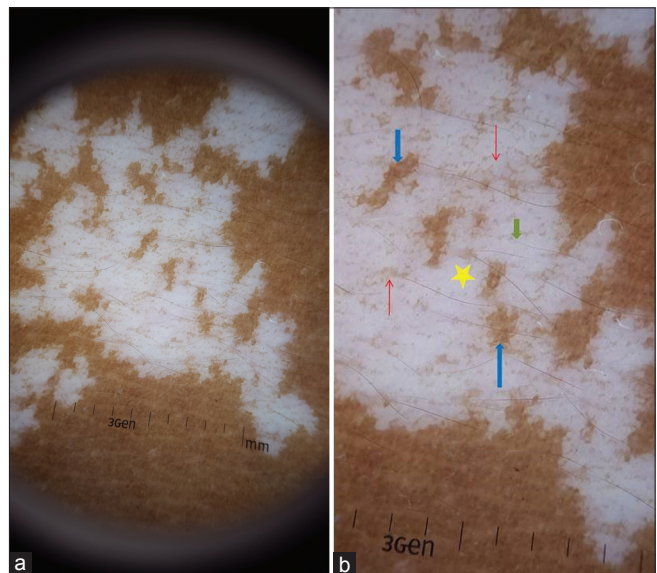
A 6 year old girl, first child born of non consanguineous marriage presented with asymptomatic white patches on the upper chest which remained stable since their appearance (Fig. 1). The child had no obvious ocular, hearing or neurological defects. The physical and mental development was normal. The audiogram of the child was normal. Routine biochemical tests were also normal. Family history was non contributory.

The examination revealed a well circumscribed white forelock in midfrontal region. Depigmented irregularly shaped macules were seen on the right upper chest. The entire back, abdomen, hands and feet, lower portions of forearm and legs and mucosae were completely spared. There was no evidence of involvement of eyebrows or eyelashes. The differentials considered at this point were piebaldism, nevus depigmentosus and congenital vitiligo.

Dermoscopic evaluation of the depigmented lesions revealed distorted melanocytic network with reduced pigment intensity (red arrows), absence of



**Figure 1:** Depigmented white patches over the anterior chest with irregular margins.



**Figure 2:** (a-b) Dermoscopy reveals distorted melanocytic network with reduced pigment intensity (red arrows), absence of perifollicular pigment (green arrow), vast areas of depigmentation (yellow star) and islands of normal skin or hyperpigmentation.

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perifollicular pigment (green arrow), vast areas of depigmentation (yellow star) and islands of normal skin or hyperpigmentation (Figs. 2a and 2b).

Based on clinical and dermoscopic features a diagnosis of piebaldism was established.

Piebaldism is a rare autosomal dominant disorder characterized by the congenital absence of melanocytes in affected areas of the skin and hair due to mutations of the *c-kit* gene, located on Chromosome 4q12, which affects the differentiation and migration of melanoblasts from the neural crest during the embryonic life [2]. Melanocytes are absent or considerably reduced in depigmented patches histologically and ultrastructurally. They are normal in number in the hyperpigmented areas.

Piebaldism results from defective migration of melanoblasts from neural crest to the ventral mid line thus manifesting dermoscopically as vast areas of depigmentation interspersed with islands of normal skin or hyperpigmentation. The second defect in the differentiation of melanoblasts to melanocytes

results in distorted melanocytic network with reduced pigment intensity. We hypothesize that the absence of perifollicular pigment may be due to the defect in transfer of melanosomes from the melanocytes in the hair bud to the surrounding keratinocytes, however further studies and ultra structural analysis will be necessary to substantiate the same.

Herein we report the first ever description of dermoscopic features in a case of piebaldism and state that further continuous documentation of observations in these rare cases will help us in arriving at more specific features.

## REFERENCES

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