The Leopard Kid with White Mane

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Quiz Case

A 6-year-old girl and her 28-year-old mother presented with chalky to creamy white patches of skin and white tufts of hair on the frontal hairline, present since birth. Dermatological examination of both patients revealed white forelock on the frontal scalp [Figure 1] and depigmented patches interspersed with hyperpigmented macules on the abdomen, mid-legs, [Figure 2] and mid-arms. There was a positive history of a similar congenital disorder in the mother's siblings. Both patients were otherwise healthy.

What is the Diagnosis?

Piebaldism.

Discussion

As the diagnosis was straightforward, no biopsy was attempted.

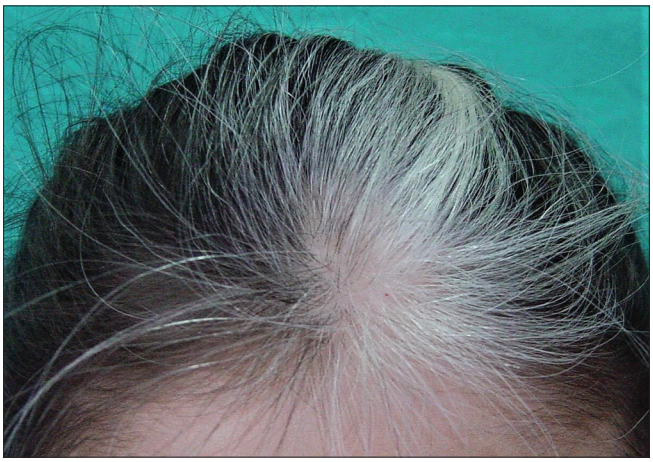

Figure 1: A white forelock on the frontal hairline

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Piebaldism (MIM172800) is a rare autosomal dominant genodermatosis, characterized by congenital patchy absence of melanocytes within the skin and hair.[1] The disorder has incomplete penetrance, variable expressivity, and a prevalence of less than 1 in 20,000 live births.[2]

Piebaldism is manifest at birth and results from germline inactivating (loss-of-function) mutations or deletions of the *c-KIT* (receptor tyrosine kinase; chromosome 4q12) or *SLUG* (zinc finger neural crest transcription factor; chromosome 8q11) genes.[2,3] Almost 75% of cases harbor mutations in the *c-KIT* gene, while the rest 25% result from *SLUG* gene mutations.[1,2] *C-KIT* proto-oncogene encodes the tyrosine kinase cell-surface receptor for mast/stem cell growth factor, and it is responsible for the proliferation, survival, differentiation, and migration of melanoblasts during embryogenesis.[1] A consistent genotype–phenotype relationship exists in piebaldism, that is, the phenotypic expressivity and severity correlate with the exact site of the mutation within the *c-KIT* gene.[1,2,4] The most severe phenotypes involve mutations of the intracellular tyrosine kinase domain, whereas mild piebaldism stems from mutations occurring

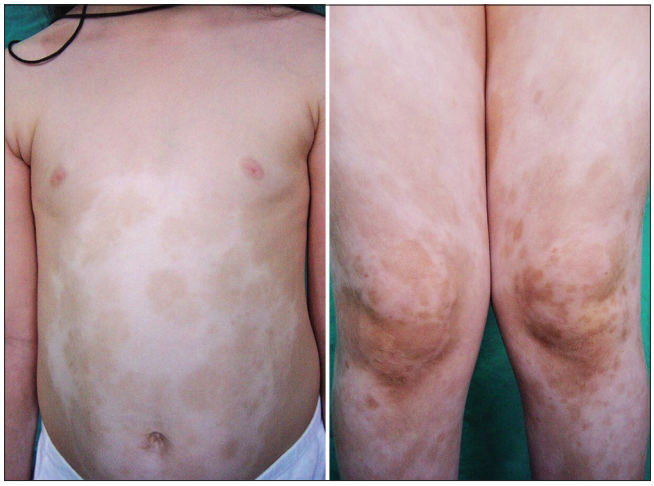

Figure 2: Depigmented patches interspersed with hyperpigmented macules on the abdomen and over the mid-legs

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in the amino terminal extracellular ligand-binding domain.[1]

Clinically, piebaldism is characterized by depigmented patches of skin (*leukoderma*), typically confined to the anterior and lateral trunk, mid-arms and legs, central forehead and the midfrontal region of scalp (*poliosis/ leukotrichia white forelock*).[1,2,4] Depigmented patches are rectangular, rhomboid, or irregular in shape and usually arranged symmetrically over the body. Classically, islands of normal to hyperpigmented skin, resembling lentigines, or freckles, are scattered within and at the border of depigmented areas.[1] White forelock may be the single clinical manifestation of the disorder in 80–90% of the cases and symbolizes a frontal, triangular, or diamond-shaped patch associated with a mesh of white hair.[1,2] Along with the midline scalp, the medial third of eyebrows and eyelashes may be affected as well.[1]

Histopathologically and ultrastructurally, depigmented patches reveal complete absence (or considerable reduction) of melanocytes within the skin and hair follicles.[1,2] Although the melanocyte number is normal in hyperpigmented areas, melanosomes are increased in number.[1]

Piebaldism is usually a benign isolated skin condition, without extracutaneous manifestations. The clinical picture tends to remain stable throughout life.[1,4] However, there are reports of both progression and spontaneous partial repigmentation of depigmented areas.[1,2,4] Contraction of leucodermic patches and complete regression of white forelock have been documented.[2]

Treatment is a challenge. Sun protection is essential to reduce carcinogenesis risk. Camouflage by temporary pigmenting agents (hair dyes, make up, or tanning product dihydroxyacetone) may be a judicious tactic. Depigmented areas of skin are refractory to medications and phototherapy protocols utilized for vitiligo.[1] The best therapeutic approach is surgical skin transplantation (split thickness skin grafting, punch grafting, or suction blister epidermal grafting).[1,4] Recently, excellent results have been achieved through transplantation of autologous melanocytes or autologous cultured epidermis.[1] Gene therapy will be the best hope for the future.

Learning Points

- Piebaldism is an autosomal dominant genodermatosis, characterized by white forelock and depigmented patches of skin (leukoderma) interspersed with hyperpigmented macules on the torso and extremities.
- The main differential diagnostic consideration is vitiligo.
- Piebaldism will be resistant to all classical therapeutic approaches for vitiligo. The best therapeutic approach is surgical skin transplantation.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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