Piebaldism: A Cutaneous Pigmentation Defect

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A female newborn presented at birth with an achromic patch at the frontal region with associated local hair depigmentation (Fig. 1) and leukoderma areas in the pre-tibial regions (Fig. 2), forearms, and abdomen (Fig. 3). Physical examination was otherwise unremarkable. She was born of an uneventful first gestation of 39 weeks, had good adaptation to extrauterine life, and an appropriate somatometry for gestational age at birth. Family history was positive for piebaldism (mother and maternal aunt). Ophthalmological and auditory evaluation were normal. Currently, she is 5 months old and has adequate psychomotor development and growth.

Piebaldism is a rare inherited autosomal dominant disease caused by a different mutation in the *C-KIT* gene located on chromosome 4q12-13.[1] In the presence of this mutation, a defect occurs in the migration and subsequent proliferation of melanoblasts in some body areas during the embryonic development, causing achromic patches due to the absence of melanin production.[2,3]

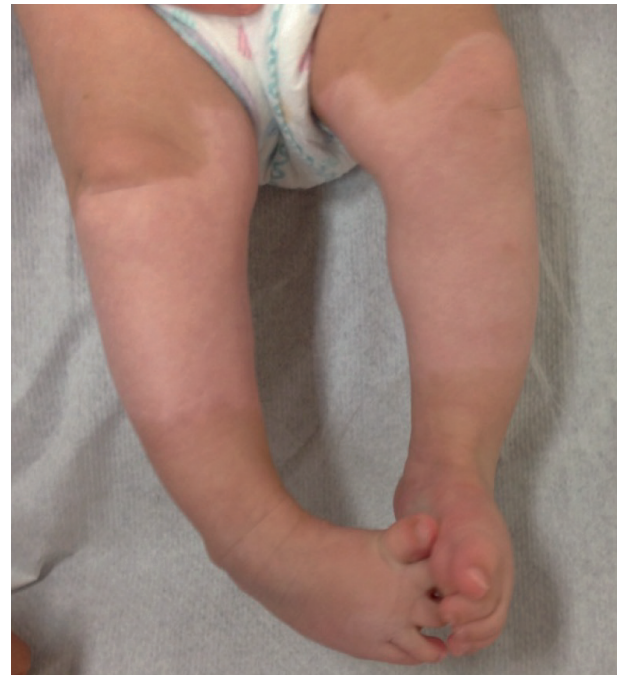

Figure 2. Leukoderma in the pre-tibial region.

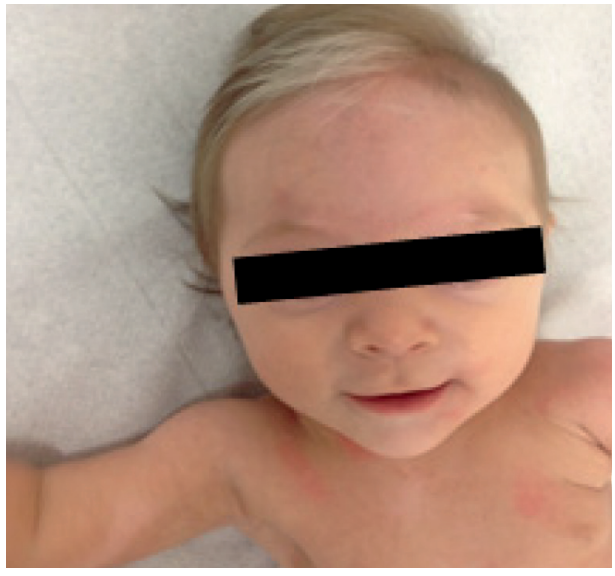

Figure 1. Achromic area in the frontal region, associated with hair depigmentation.

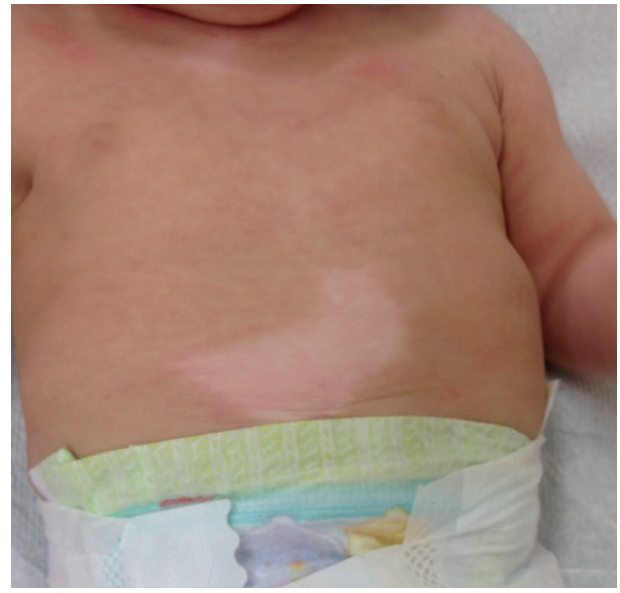

Figure 3. Hypochromic patches in the abdominal region.

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The most suggestive clinical finding is the white forelock, a triangular achromic area in the frontal region associated with hair depigmentation (poliosis), present in 80%-90% of the cases.[1] Other areas, such as the limbs, chest, and abdominal region, may also be affected. Skin changes are present from birth and remain stable over time.[2-4]

Typically, islands of hyperpigmentation are present within and at the border of depigmented areas.

The differential diagnosis is made with other cutaneous pigmentation disorders, such as vitiligo, total albinism, Tietz syndrome, and Waardenburg syndrome.[5]

In the case described, the presence of circumscribed poliosis associated with areas of depigmentation in the limbs and abdomen since birth, the stable course and positive family history, ensured the differential diagnosis with vitiligo. The remaining entities were excluded, considering the physical examination, hearing assessment, and ophthalmologic evaluation.

Therapy relies essentially on daily preventive photoprotection.

The association of clinical history, genealogy and physical examination is crucial to the diagnosis of this pathology, obviating the need for complementary tests.

Keywords: Diagnosis, Differential; Infant, Newborn; Piebaldism/diagnosis; Skin Diseases, Genetic

WHAT THIS REPORT ADDS

- The presence of poliosis should make us suspicious of Piebaldism.
- Skin changes are present from birth and remain stable over time.
- The association of clinical history, genealogical tree, and physical examination is crucial to the diagnosis.

Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

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Consent for publication

Consent for publication was obtained.

Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

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