PIGMENTARY MOSAICISM ALONG BLASCHKO'S LINES: CLASSIFICATION CHALLENGES

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Case reports. *Case 1*. A 26-year-old man presented with asymptomatic white lesions on the right side of the abdomen and thigh, present since birth. There was no history of prior inflammation, trauma, or similar lesions in the family. Cutaneous examination revealed well-demarcated hypopigmented to depigmented macules arranged in a spiral and whorled pattern on the flank, with focal areas of pigmentation and poliosis within the white patches. The lesions followed Blaschko's lines, extending from the right abdomen to the thigh, gluteal region, and knee (Fig. 1). The general physical examination was within normal limits.

A 4-mm punch biopsy showed a marked decrease in melanocytes. This finding was neither specific for hypomelanosis of Ito nor sufficient to distinguish it from systematized nevus depigmentosus. Because the available clinical and histopathological information did not allow for a reliable distinction between these entities, the condition was preferentially described as hypopigmentation distributed along Blaschko's lines within the spectrum of pigmentary mosaicism.

Case 2. A 10-day-old, full-term male neonate, large for gestational age, born to a 20-year-old primigravida via spontaneous conception from a non-consanguineous marriage, was referred for the evaluation of hyperpigmented lesions on the trunk. No antenatal or perinatal complications were reported. Cutaneous examination showed multiple, well-demarcated, and hyperpigmented macules arranged along Blaschko's lines on the trunk and extremities (Fig. 2).

With this extensive pattern, it was not possible to establish with certainty whether the darker areas represented altered skin against a lighter background or whether the lighter areas represented the altered component relative to a more extensive darker skin background. The greater extent of the darker pigmentation might favor the latter interpretation, but body surface area alone cannot determine the baseline phenotype. The case was therefore described from a morphological standpoint without presupposing that either color represents normal skin. Further clinical follow-up, careful examination of apparently uninvolved skin and familial pigmentation, as well as complementary evaluation when indicated, may help refine the interpretation.

Discussion. Pigmentary mosaicism describes patterned alterations of cutaneous pigmentation that can follow Blaschko's lines. These alterations may be hypopigmented, hyperpigmented, or a combination of both. Recognizing the distribution is important, but the pattern alone does not establish a specific diagnosis (1, 2).

In the first case, the congenital onset and whorled hypopigmentation raised suspicion for both hypomelanosis of Ito and nevus depigmentosus. Nevus depigmentosus (or hypomelanic nevus) typically appears at birth or in early childhood and remains relatively stable in distribution; it may be localized or segmental (3). Hypomelanosis of Ito is characterized by more extensive blaschko-hypopigmenta-



Fig. 1



Fig. 2

Fig. 1, 2: Pigmentary mosaicism distributed along Blaschko's lines. In Fig. 1 (case 1), extensive areas of hypopigmentation span multiple segments. In Fig. 2 (case 2), the generalized distribution makes it difficult to identify the abnormal color.

tion, which may be associated with extracutaneous anomalies, particularly neurological and musculoskeletal ones. However, the boundaries between these conditions are not sharply defined. Associated anomalies have been reported in both groups, supporting their inclusion within the broader spectrum of pigmentary mosaicism (4). Although our patient's general physical examination was normal, this finding alone does not allow for a distinction between the two conditions. We therefore favored the descriptive diagnosis of pigmentary mosaicism distributed along Blaschko's lines.

The reduction in melanocytes observed on biopsy does not independently confirm hypomelanosis of Ito. In nevus depigmentosus, a reduced amount of melanin with a preserved number of melanocytes has been demonstrated, suggesting a defect in melanocyte function or melanosome formation (3). Histopathological findings must therefore be interpreted in conjunction with clinical and anamnestic data, rather than being used in isolation to formulate a specific diagnosis.

The second case illustrates the difficulty of identifying normal skin color in extensive pigmentary mosaicism. The darker areas were initially interpreted as hyperpigmented lesions. However, their greater extension raises the possibility that they represent the background skin, with the lighter areas constituting the pathological condition. This remains a clinical interpretation, as the relative extent of each color cannot establish which one is abnormal. Comparison with apparently unaffected skin and evaluation under appropriate illumination can be useful; Wood's lamp examination can render pigmentation differences more apparent but does not inherently identify the normal skin population (2).

Evaluation for neurological, developmental, musculoskeletal, and ocular anomalies remains important in patients with pigmentary mosaicism (2, 5). In neonatal-onset cases, follow-up is beneficial because some of the associated findings may manifest at a later stage.

Conclusion. These two cases demonstrate the classification challenges within the spectrum of pigmentary mosaicism.

Conflicts of interest

The authors declare that they have no conflicts of interest.

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