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**Cutis tricolor: congenital hyper- and hypopigmented lesions in a background of normal skin with and without associated systemic features: further expansion of the phenotype**

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**Abstract** The term cutis tricolor describes the uncommon co-existence of congenital hyper- and hypopigmented macules, in close proximity to each other, in a background of normal skin so far seen in a 17-year-old patient with various other congenital defects. The suggested explanation for this phenomenon is allelic twin spotting. We report on two boys, aged 6 and 11 years, with an unusual combination of three different degrees of pigmentation, one of whom had in addition, psychomotor delay, dysmorphic features, musculoskeletal abnormalities and subcortical and periventricular white matter high signal lesions on brain neuroimaging. In both cases a search for mosaicism in peripheral blood lymphocytes and cultured fibroblasts was negative. In contrast to the previously reported case, the two children had large streaks or patches of hyper- and hypopigmented skin lesions, in close proximity to each other, involving large areas of the body. The rest of the skin had a normal intermediate pigmentation.

**Conclusion** This combination of three degrees of pigmentation in association with systemic defects in one child and the lack of such association in the other confirms and further expands the clinical phenotype of cutis tricolor.

**Introduction**

Happle et al. [7] in a recently described new syndrome coined the term “cutis tricolor” for the presence of congenital hyper- and hypopigmented macules on a background of normal skin. The skin changes in their sporadic case were associated with multisystemic birth defects [7].

Here, two children are reported with an unusual combination of three degrees of pigmentation of whom one had, in addition, psychomotor delay, dysmorphic features, musculoskeletal abnormalities and white matter lesions on brain neuroimaging.

**Case reports**

**Case 1**

This 11-year-old boy was the full-term product of unrelated parents born after normal delivery following an uneventful pregnancy. Developmental milestones were normal. He was recalled to have since birth a large, circumferential, streaky segment of café-au-lait pigmentation over the trunk. At age 6 years, on the basis of the above lesion and of three additional café-au-lait spots, the diagnosis of neurofibromatosis type 1 (NF1) was suggested. At that time, screening investigations, including slit lamp and fundus examination, ECG, EEG, abdominal and head ultrasonography and brain MRI were normal.

When first referred to our institution at age 9 years, on examination his height, weight and head circumference were within the 50th percentile. There were no dysmorphic features. On examination of the skin, a peculiar pigmentary disturbance was noted (Figs. 1, 2). A large, spirally shaped, streaky hyperpigmented lesion involved the lower abdomen from the right peri-umbilical area, with a sharp midline cutoff, over the right flank and trunk across the interscapular region and the left scapular area under the axilla and around the left chest and neck towards the mandible and right cheek. In addition, well demarcated areas of hypopigmented streaks were present in the right upper chest (Figs. 1A, B, 2) and hypopigmented patches in the right supraclavicular area and neck (Figs. 1B, 2), in close proximity to the hyperpigmented lesions. The rest of the skin had a normal intermediate pigmentation (Figs. 1, 2). Three café-au-lait macules (1 × 2 cm across) with irregular borders, were noted over the trunk and left thigh. The rest of physical examination was otherwise unremarkable. Absence of
**Fig. 1A-D** Case 1 at age 10 years. A A large streak of hyperpigmentation involves the lower abdomen from the right peri-umbilical area with a sharp midline cutoff. Note the hypopigmented area over the right upper chest in close proximity to the hyperpigmented lesion over the left upper chest and the normally pigmented skin over the upper portion of the abdomen. B The lesion goes over the right flank and trunk. Note the triple coloured skin: the right upper chest is hypopigmented, the upper portion of the abdomen is normally pigmented and the lower abdomen hyperpigmented. C Involvement of the posterior aspect of the trunk across the interscapular area and the left scapular area under the axilla. D Note the hyperpigmented lesion over the left chest, neck, the mandible and the right cheek. A well demarcated area of hypopigmentation is evident in the right supraclavicular area in close proximity to the hyperpigmented lesions. The rest of the skin has a normally pigmented background.

**NF1 and NF2 gene mutations using 15 NF1 polymorphic microsatellite markers [19] and the NF2 polymorphic microsatellite markers was demonstrated in the peripheral blood lymphocytes obtained from the proband and his relatives and in the cultured fibroblasts obtained from the proband’s hyperpigmented and normal skin areas.**

**Case 2**

A 6-year-old boy had psychomotor retardation, multiple dysmorphic features and widespread pigmentedary disturbances. He was born at term after an uneventful pregnancy and normal delivery. His parents recalled that the pigmented lesions were first noticed at age 3 months. When first referred to our institution he was 4 years old. On examination his height was >90th percentile and height and head circumference were within the 50th percentile. He had hypertelorism, epicanthal folds, deep set and forward rotated ears, deep nasal bridge, large and bulbous nose with broad nostrils, large philtrum and prominent lips and short neck (Fig. 3A). There was pectus excavatum (Fig. 3A), mild kyphoscoliosis resulting in scapular deformity (Fig. 3B) and leg length discrepancy with the

**Fig. 2** Diagram showing the arrangement of the three different degrees of pigmentation in case 1.

- **Normal intermediate pigmentation**
- **Hypopigmentation**
- **Hyperpigmentation**