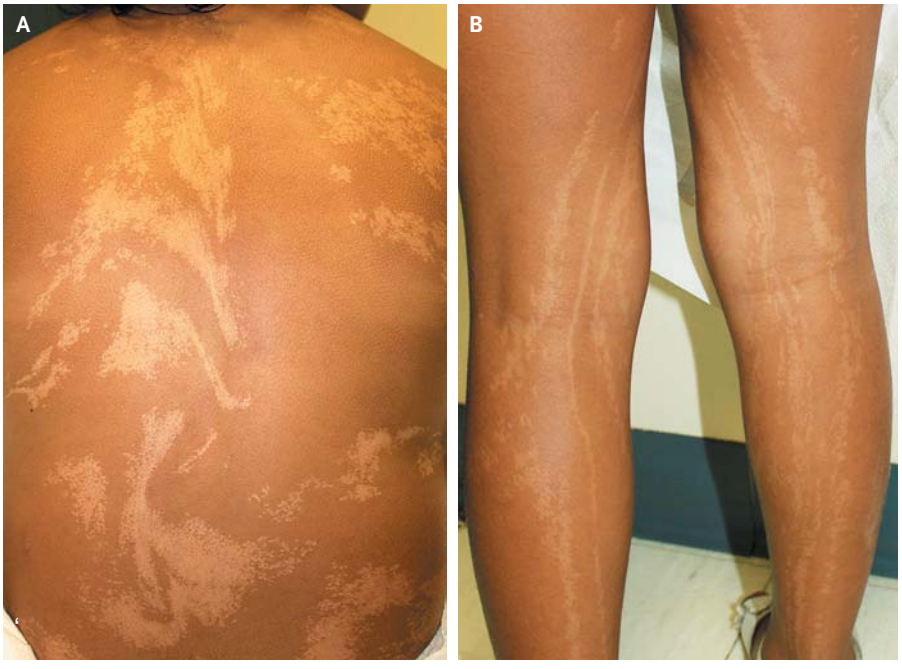
IMAGES IN CLINICAL MEDICINE

X-Chromosomal Translocation and Segmental Hypopigmentation



A Hispanic girl who had been born at term to healthy nonconsanguineous parents was affected by brachycephaly, developmental delay, a mild cognitive deficit, and behavioral disturbances. Cytogenetic testing revealed an apparently balanced translocation between an X chromosome and a chromosome 2 (breakpoints, Xp11.2 and 2q35). When the girl was seen for dermatologic consultation at 8 years of age, we observed hypopigmentation streaks in a wavy pattern on the trunk (Panel A) and a linear pattern on the limbs (Panel B). Lines of Blaschko, believed to be the expression of clones of abnormal cells migrating toward the periphery during embryonic life, are visualized in a wide variety of genodermatoses. An enormous range of cytogenetic abnormalities can cause pigmentary mosaicism. In particular, Blaschkolinear hypopigmentation (also called hypomelanosis of Ito) is typically linked to translocations between an X chromosome (loci Xp11.21 and Xp11.22) and an autosome.

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