

Clinical findings of the phakomatoses

Hypomelanosis of Ito

Mark Quigg, MD, MSc; Robert S. Rust, MD; and James Q. Miller, MD†

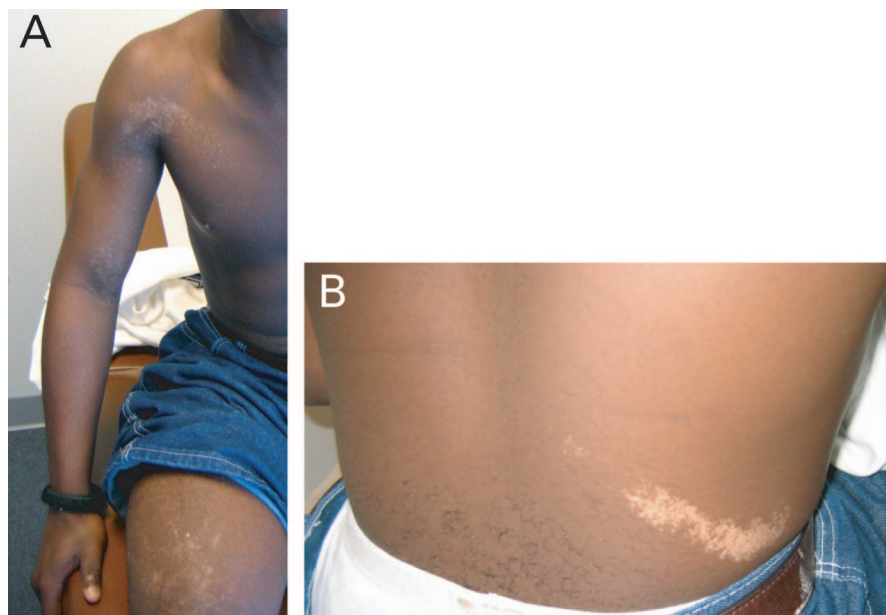


Figure 1. (A) A 15-year-old boy with hypomelanosis of Ito (HI) and seizures demonstrates reticulated white maculae present on the right arm and leg. (B) In the same patient a right lumbar hypopigmented streak clearly demonstrates the characteristic macules along Blaschko lines. These lines indicate the migratory paths of ectodermal precursor cells and are distinct from dermatomes. Errors in chromosomal division during embryogenesis lead to chromosomal mosaicism, reflected among successors to different cell lines. The skin lesions of HI present at birth or early childhood and may fade with age.

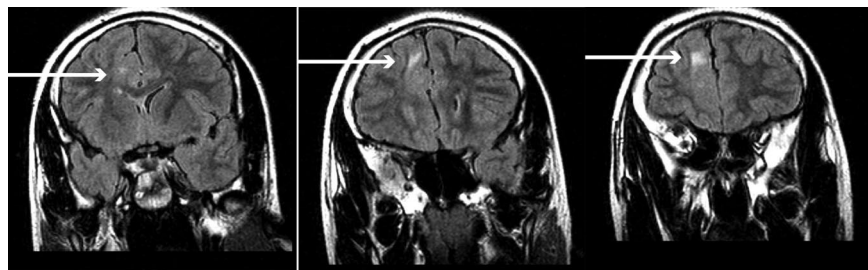


Figure 2. In the same patient as in figure 1, three sequential sections of a coronal fluid-attenuated inversion recovery MRI show a high-intensity lesion in the right cingulate gyrus, ipsilateral to cutaneous findings (arrows). Hypomelanosis of Ito (HI) lacks characteristic neuroimaging findings probably as the result of mosaicism and ranges among cortical dysplasias, heterotopias, and hamartomas.

Hypomelanosis of Ito (HI, incontinentia pigmenti achromians, pigmentary mosaicism-Ito type) is a multisystem disorder with unilateral or bilateral hypomelanotic whorled, streaked, or reticulated macules distributed along Blaschko lines. Extracutaneous congenital abnormalities present in 75% of cases of HI and usually involve the brain;

mental retardation is present in 60%, epilepsy in 50%, and autism in 10%. Dysmorphism (hemihypertrophy or hemiencephalomegaly), dental, cardiac, gastrointestinal, or renal abnormalities may present.¹ Chromosomal mosaicism is the basis of variability in HI² and other disorders with lesions distributed along Blaschko lines; abnormali-

ties in HI include 9q33, 15q11-q13, Xp11, and Xp21.2.² Clinical and neuroimaging findings are shown in Figures 1 and 2.

1. Pascual-Castroviejo I, Roche C, Martinez-Bermejo A, et al. Hypomelanosis of ITO: a study of 76 infantile cases. *Brain Dev* 1998; 20:36-43.
2. Hatchwell E. Hypomelanosis of Ito and X;autosome translocations: a unifying hypothesis. *J Med Genet* 1996;33:177-183.

From the Department of Neurology, University of Virginia, Charlottesville.

† Deceased.

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Address correspondence and reprint requests to Dr. Mark Quigg, Department of Neurology, Box 800394, Health Sciences Center, University of Virginia, Charlottesville, VA 22908; e-mail: Quigg@virginia.edu

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