

An infant with epilepsy: don't forget the importance of skin examination

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DESCRIPTION

A 4-month-old, developmentally normal girl presented with four episodes of right focal seizures. She had an uneventful perinatal period and no significant family history. Clinical examination revealed normocephaly, and hypopigmented whorls along Blaschko's lines over left anterior trunk, back (left >right), both upper limbs and left leg (figure 1). Brain MRI revealed left hemimegalencephaly (figure 2). There was no preceding history of vesicular or verrucous lesions. She was diagnosed as hypomelanosis of Ito (HI) and initiated on oral phenytoin and multisensory stimulation.



Figure 1 Skin findings in the index child. Note the hypopigmented whorls following Blaschko's lines (A) over the back (right >left), (B) abdomen, chest (left-sided), (C) left leg and (D) left arm.

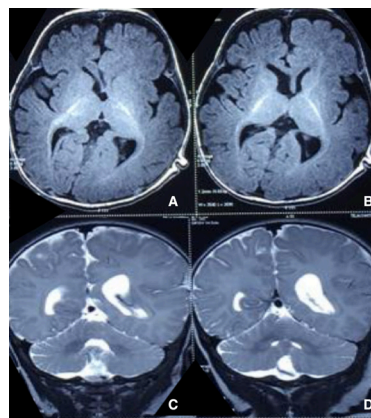


Figure 2 Brain MRI of the index child. (A,B) Axial T1-weighted and (C,D) coronal T2-weighted images showing enlarged left hemisphere and prominence of occipital horn of the left lateral ventricle suggestive of left hemimegalencephaly.

HI (also known as incontinentia pigmenti achromians) is neurocutaneous symptom complex characterised by the presence of whorled hypochromic skin lesions often associated with systemic manifestations.^{1 2} Incontinentia pigmenti (with preceding vesicular or verrucous phases) is an important differential diagnosis. HI is often associated with systemic abnormalities such as ophthalmologic and skeletal abnormalities, brain and cardiac malformations.^{1 3} Neurological manifestations include epilepsy, developmental delay, autistic features, brain malformations such as agenesis or dysplasia of the corpus callosum, hemimegalencephaly and arteriovenous malformations.^{1 3} Epilepsy has been reported in up to 50% of patients.

Even in the absence of epilepsy, a neuroimaging may be warranted in a florid dermatological presentation. Also, the importance of skin examination cannot be understated even if the dermatological findings are subtle such as ash leaf macules and so on.

Learning points

- ▶ Hypomelanosis of Ito (HI) is characterised by whorled hypochromic skin lesions along Blaschko's lines.
- ▶ Incontinentia pigmenti is an important differential diagnosis.
- ▶ HI may be associated with systemic abnormalities such as brain malformations and so on, and these should be actively looked for.

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