NEUROLOGICAL PICTURE

doi: 10.1136/jnnp.2005.084723

Hypomelanosis of Ito

Hypomelanosis of Ito (HI) is characterised by hypochromic unilateral skin lesions, hemi-hypertrophy, mental retardation (MR) and seizures.¹ We describe a young woman with HI with interesting and hitherto unreported magnetic resonance imaging (MRI) changes.

A 22 year old woman presented with a history of uncontrolled generalised tonic clonic seizures and myoclonic jerks since the age of 2 years. She had hypopigmented macular lesions in streaks and whorls on her right side in a dermatomal distribution (fig 1 A–D). There was hemihypertrophy of the right upper extremity, and it measured 15 mm longer than the left (fig 1C).

She had moderate to severe MR with an intelligence quotient of 33.3 on the Binet-Kamat test of intelligence, and a social quotient of 32.3 on the Vineland Social Maturity scale. MRI brain revealed right hemimegalencephaly, temporoparieto-occipital pachygyria, and absence of the anterior limb of the sylvian fissure (fig 2A–D). Scalp EEG revealed slowing of background activity and bihemispheric epileptiform discharges.

The term "hypomelanosis" was introduced by Ito and is perhaps the fourth most common neurocutaneous syndrome.^{2–3} Characteristically these patients have hypochromic unilateral or bilateral lesions in whorls, patches, and streaks, with a midline cutoff. Neurological involvement is noted in 76% of patients, with seizures and MR being common. Other features include ataxia, neuropathy, distal spinal muscular atrophy, torticollis, deafness, and spina bifida occulta. Hemimegalencephaly on either side of skin lesions, hemihypertrophy often ipsilateral to hypomelanotic lesions,⁵ arm and leg length discrepancy, and scoliosis are also noted. Neuroimaging may reveal hypoplastic corpus callosum, heterotopias, periventricular white matter changes, vascular malformations, medulloblastoma, and choroid plexus papilloma.^{4–5}

Our patient had the classical phenotype of HI and a few novel findings such as ipsilateral pachygyria and absence of the anterior limb of the sylvian fissure.

N Shobha, A B Taly, S Sinha, G R Arunodaya

Department of Neurology, NIMHANS, Bangalore, India

S G Srikanth

Department of Neuroradiology, NIMHANS, Bangalore, India

Correspondence to: Professor A B Taly, Department of Neurology, NIMHANS, Bangalore: 560 029, Karnataka, India; abtaly@yahoo.com

References

- 1 Berg B. Neurocutaneous syndromes: phakomatoses and allied conditions. In: Swaiman K, ed. *Pediatric neurology: principles and practice*, 3rd ed, 1999:544–545.
- 2 Schwartz MF, Esterly NB, Fretzin DF, et al. Hypomelanosis of Ito (Incontinentia pigmenti achromians): a neurocutaneous syndrome. *J Pediatr* 1977;**90**:236–240.
- 3 Sybert VP. Hypomelanosis of Ito: a description, not a diagnosis. *J Invest Dermatol* 1994;**103**(suppl):141–3.
- 4 Ardinger HH, Bell WE. Hypomelanosis of Ito. Wood's light and magnetic resonance imaging as diagnostic measures. *Arch Neurol* 1986;**43**:848–50.
- 5 Sotero de Menezes M. Hypomelanosis of Ito. www.emedicine.com/ped/topic1123.htm.

Figure 1 Phenotypic features of hypomelanosis of Ito. (A) Hypopigmented macular lesions in streaks and patches over the right side of trunk; (B) striking whorl over the right half of the abdomen; (C, D) hemihypertrophy of the right upper and lower extremities with hypopigmented macular lesions. Informed consent was obtained for publication of this figure.

Figure 2 MRI of the brain showing right sided hemimegalencephaly and pachygyria. (A) T1 weighted axial image in addition showing absence of the anterior limb of the sylvian fissure with hemimegalencephaly and pachygyria on right side. (B) T2 weighted axial image and (C, D), coronal images showing distinct right sided hemimegalencephaly and pachygyria.