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Letter to the Editor

Hemihypertrophy with hemimegalencephaly associated with type 1b (broad pattern) pigmentary mosaicism

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Dear Editor,

Pigmentary mosaicism represents patterns of pigmentation in the skin and is observed with the defects of the neurological and musculoskeletal systems.[1] Hemimegalencephaly is the enlargement of one hemisphere of the brain that may occur with neurocutaneous syndromes.[2] A 2.5-year-old girl presented with developmental delay and right focal seizures from 7 months of age. On examination, she was found to have normal head circumference (47 cm), multiple hypopigmented streaky macules along the lines of Blaschko-type 1b: Broad pattern on the trunk and upper and lower limbs, hypertrophy of the right upper and lower limb [Figure 1]. The magnetic resonance imaging brain showed left hemimegalencephaly [Figure 2].

Hemimegalencephaly with skin lesions is also seen in linear epidermal nevus syndrome, neurofibromatosis, Proteus syndrome, and Klippel–Trenaunay syndrome. The classical cutaneous lesions along Blaschko lines are helpful to differentiate them. Pigmentary mosaicism should be suspected in any child presenting with refractory focal seizures, hemihypertrophy with classical skin findings and hemimegalencephaly.

Figure 1: Multiple streaks of hypopigmentation along the lines of Blashcko present on the trunk, upper and lower limbs (arrows).

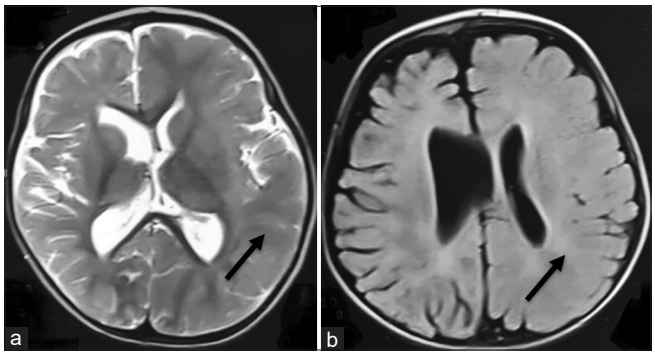

Figure 2: Brain magnetic resonance imaging-T2W (a) and fluid-attenuated inversion recovery (b) showing hypertrophy of the left cerebral hemisphere (arrows).

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

REFERENCES

1. Kromann AB, Ousager LB, Ali IK, Aydemir N, Bygum A. Pigmentary mosaicism: A review of original literature and recommendations for future handling. Orphanet J Rare Dis 2018;13:39.
2. Kentab AY. Hypomelanosis of ito with partial motor seizure and hemimegaloencephaly: Case report. Open Access J Neurol Neurosurg 2017;2:36-9.

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