Blaschkoid hypermelanosis in a patient with ring 18 chromosome

Sir,

Linear and whorled nevoid hypermelanosis is a rare sporadic pigmentary anomaly occurring in blaschkoid distribution. It has been associated with several chromosomopathies including trisomy 7, 14, 18, 20, inversion of chromosome 9 and X-chromosomal mosaicism.[1-5]

Ring chromosomes usually occur "de novo" due to deletion and fusion of both ends of chromosome. Ring chromosome 18 is a rare disorder characterized by severe mental retardation, microcephaly, brain and ocular malformations, hypotonia, skeletal abnormalities and immunological disorders.[6,7]

A 4-year-old boy, after a full-term birth from eutocic delivery was diagnosed with microcephaly, psychomotor retardation, generalized hypotonia and impaired ambulation. His father had a factor V Leiden heterozygous mutation. The comparative genomic hybridization of peripheral blood demonstrated a ring 18 chromosome with 17% mosaicism. He was referred to the dermatology department due to a 3-year history of stable and asymptomatic hyperpigmented lesions which appeared within the first month of life. Upon examination, we observed a bilateral and symmetrical thoracoabdominal macular hyperpigmentation in blaschkoid distribution with midline demarcation. There were no associated hypopigmented macules or other cutaneous primary lesions [Figure 1]. On complete examination, the patient was found to have slightly low-set ears with no other adnexal, mucosal or musculoskeletal associated abnormalities [Figure 2].

Linear and whorled nevoid hypermelanosis, a rare sporadic pigmentary anomaly is characterized by swirls and streaks of macular hyperpigmentation in blaschkoid distribution, as described by Kalter *et al*. in 1988.[1] Classically, it is seen within the first 2 years of age and is distributed mainly

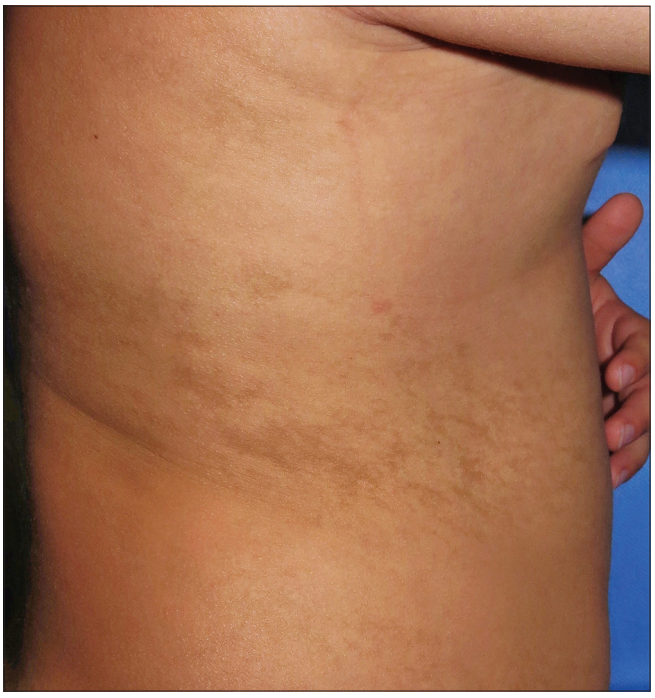

Figure 1: Macular linear hyperpigmentation on the right thoracoabdominal area with midline demarcation

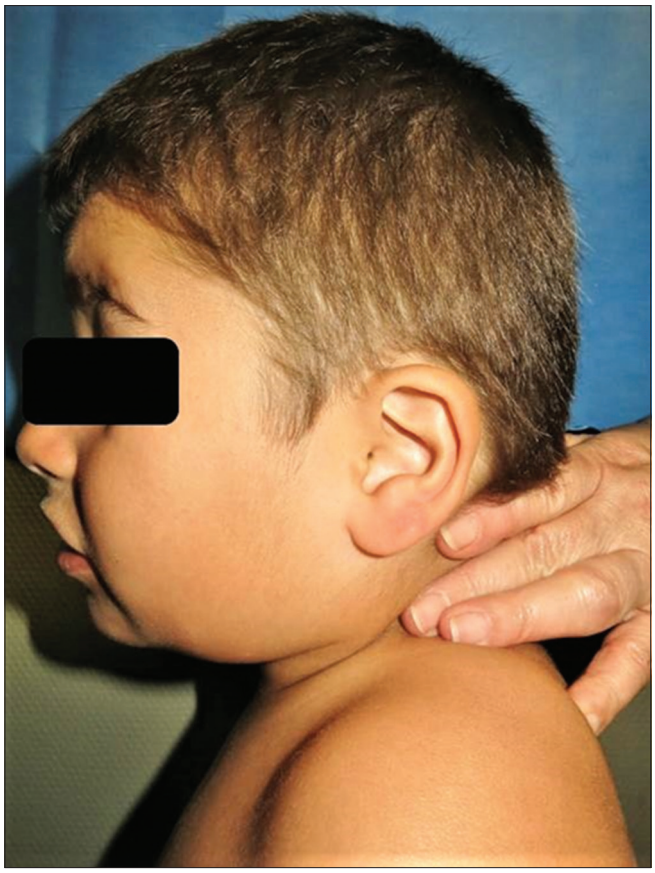

Figure 2: Low-set ear

over the trunk, neck and extremities, sparing the palms, soles and mucosa. Lesions appear without preceding inflammation, rash or injury and progress for a year or two before stabilization.[2-5] No satisfactory treatments are currently available but in some patients, macules become less prominent with age. The histopathological findings reveal diffuse epidermal hyperpigmentation in the basal layer without melanocytic proliferation or pigmentary incontinence.[1-5]

Linear and whorled nevoid hypermelanosis has been related to the pigmentation variability in cellular mosaicisms leading to distinct population of melanocytes with different potentials of pigment production. Frequently, it has been associated with chromosomopathies including trisomy 7, 14, 18, 20, inversion of chromosome 9 and X-chromosomal mosaicism. However, its underlying pathogenesis is yet unknown. In the literature, rare cases associated with extracutaneous manifestations have been reported, mostly with cardiac defects, developmental and growth retardation or body asymmetries, among others.[2-5]

The differential diagnoses include pigmentary stage of incontinentia pigmenti, hypomelanosis of Ito, early-stage linear epidermal nevus and Goltz syndrome. The clinical absence of preceding vesicular or verrucous lesions, and microscopic findings lacking pigmentary incontinence, papillomatosis, rete ridge elongation or dermal hypoplasia differentiate the linear and whorled nevoid hypermelanosis from these diseases.[3,4] In addition, it should be remembered that early stages of incontinentia pigmenti could take place in-utero, recommending a histological examination to rule out this diagnosis.

In conclusion, we present a linear and whorled nevoid hypermelanosis in association with a mosaic ring 18 chromosome. We were unable to find any previous reports of this syndrome associated with cutaneous hypermelanosis.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understands that name and initials will not be published and due efforts will be made to conceal identity but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Eduardo Varas-Meis, Sonsoles Delgado-Vicente, Paula Fernández-Canga, Manuel Ángel Rodríguez Prieto

Department of Dermatology, Complejo Asistencial Universitario de León, León, Spain

Correspondence: Dr. Eduardo Varas-Meis, Department of Dermatology, Complejo Asistencial Universitario de León, Altos de Navas S/N, 24080 León, Spain.
E-mail: edu.varas.meis@gmail.com

References

1. Kalter DC, Griffiths WA, Atherton DJ. Linear and whorled nevoid hypermelanosis. J Am Acad Dermatol 1988;19:1037-44.
2. Di Lernia V. Linear and whorled hypermelanosis. Pediatr Dermatol 2007;24:205-10.
3. Ito T, Yoshida Y, Goto H, Furue M, Yamamoto O. Linear and whorled nevoid hypermelanosis with tetralogy of fallot. Indian J Dermatol 2015;60:325.
4. Hong SP, Ahn SY, Lee WS. Linear and whorled nevoid

hypermelanosis: Unique clinical presentations and their possible association with chromosomal abnormality inv (9). Arch Dermatol 2008;144:415-6.
5. Komine M, Hino M, Shiina M, Kanazawa I, Soma Y, Tamaki K. Linear and whorled naevoid hypermelanosis: A case with systemic involvement and trisomy 18 mosaicism. Br J Dermatol 2002;146:500-2.
6. Lo-Castro A, El-Malhany N, Galasso C, Verrotti A, Nardone AM, Postorivo D, *et al*. De novo mosaic ring chromosome 18 in a child with mental retardation, epilepsy and immunological problems. Eur J Med Genet 2011;54:329-32.
7. Heydari S, Hassanzadeh F, Hassanzadeh Nazarabadi M. Ring chromosome 18: A case report. Int J Mol Cell Med 2014;3:287-9.

Access this article online

Quick Response Code:

Website: www.ijdvl.com

DOI: 10.4103/ijdvl.IJDVL_282_18

How to cite this article: Varas-Meis E, Delgado-Vicente S, Fernández-Canga P, Rodríguez Prieto MÁ. Blaschkoid hypermelanosis in a patient with ring 18 chromosome. Indian J Dermatol Venereol Leprol 2020;86:316-8.

Received: May, 2018. **Accepted:** December, 2019.