Extensive Mongolian spots in a healthy child

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Mongolian spots (MS) are solitary or numerous, irregularly shaped non-blanching macules to patches with varying shades of blue-green, gray, or blue-black discoloration [1, 2]. MS are usually transient and arise from the physiological developmental processes in the skin [3]. They are either present at birth or appear within the first few weeks of life. The most common site is the sacral-gluteal region [4]. They measure few millimeters to less than 10 cm in size. Although they are usually benign in character, they can cause significant anxiety for both parents and doctors due to their unusual appearance, unexpected location and number. They usually fade during first few years of life. MS with persistence beyond their physiological duration, increased number, atypical pigmentation, larger size, and their presence in the extra-sacral region have been found to be associated with fatal disorders of inherited inborn errors of metabolism (IEM) [1–5]. Herein, we are presenting the case of extensive MS in a 15-month-old female child.

A 15-month-old female child born of parents of South Asian ethnicity, was brought to the skin outpatient clinic with extensive bluish-black discoloration over chest, back and both thighs since birth. On cutaneous examination, the child had diffuse, irregularly bordered flat patches of grayish-blue discoloration over the abdomen but sparing the peri-umbilical area (fig. 1). Similar pigmentation was present over the back except the scapular regions (fig. 2). The pigmentation was persistent and not showing any change with age. The head, neck, eyes, external ear, and genitals did not show any abnormal looking skin pigmentation. Examination of the mucosae revealed no abnormalities. Overall, 60% of the body area was covered with the lesions. Based on morphology and clinical characteristics, the extensive MSwere considered as the most probable diagnosis. A complete clinical evaluation including laboratory tests was carried out in view of its association with IEM. Vital parameters were within normal limits. She had normal length and weight for her age. The child was well alert to visual, vocal, and sensory stimuli. Developmental milestones were reached according to the age. Parents reported normal feeding and according to patterns. Shape/symmetry of the chest and abdomen was normal. There was no abnormal finding on palpation of the abdomen. A complete hematological, biochemical and radiological skeletal survey, ultrasound of the abdomen and renal system was also normal. None of the family members had any history of similar skin manifestations. Findings of these evaluations strongly suggested possible absence of any concealed systemic disorder in the child.

MS are the most common type of dermal melanocytosis [6]. They are also the most common transient neonatal skin conditions which present as solitary or numerous, irregularly shaped, non-blanching macules to patches with varying shades of blue-green, gray, or blue-black, discoloration [1, 2]. They can be present at birth or appear later during the first few weeks of life [1–4]. The pigmentary appearance of MS is due to the scattering of colors of shorter wavelength to the epidermis. The amount of melanin, the number of dermal melanocytes and their depth in the dermis are important determinants of color of MS [7]. Both genders are affected equally. The prevalence is affected by ethnicity, with the highest prevalence (81–100%) occurring in the Asian population [8].

MS presenting at birth tend to regress spontaneously in the first 2 years of life and other types regress by early childhood. Various mechanisms have been postulated to describe spontaneous regression, clearance by dermal macrophages and gradual loss of protective extracellular fibrous sheath of dermal melanocytes is widely accepted. This sheath is seen to be preserved in persistent MS [1, 6]. MS are most commonly classified as sacral and extra-sacral. Based on the speed of regression, they have been classified into three further types: the common type (regresses by early childhood), extensive type (regresses very slowly), and persistent type (may persist into adulthood). Historically, MS have been thought to be benign but in recent time evidence of their association, especially of extensive and persistent MS, with numerous dis-

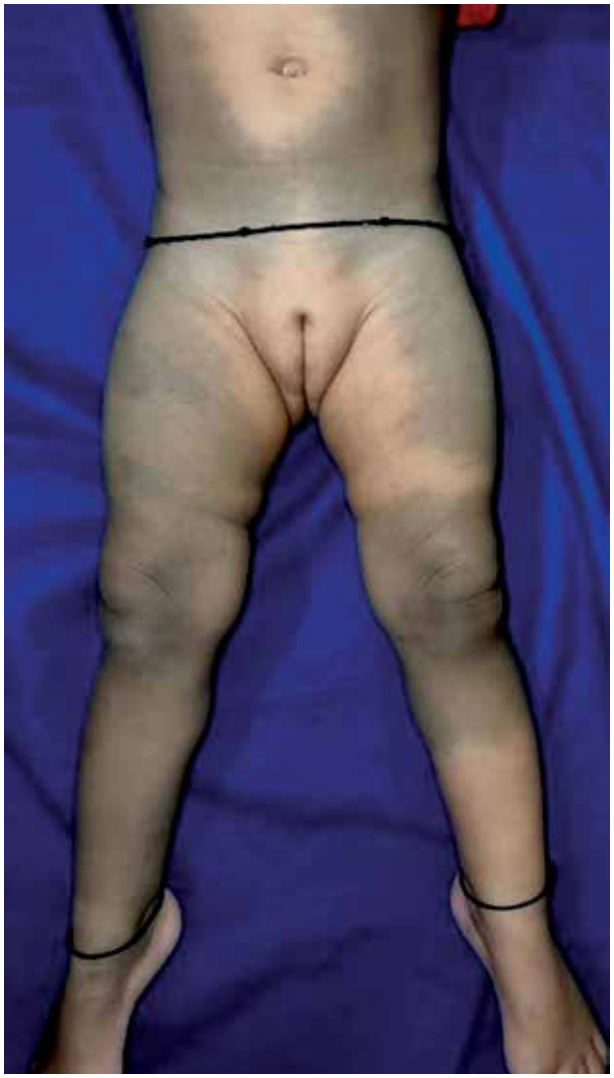

Figure 1. Diffuse, irregularly bordered flat patches of grayish-blue discoloration over the abdomen and anterior aspect of both legs sparing the peri-umbilical area and genitals

Figure 2. Diffuse greyish-blue patches over the lower two-third of the back, buttocks and lower limbs

Table 1. Common associations of Mongolian spots

Inborn error of metabolisms (IEMs)	Lysosomal storage disorders Mucopolysaccharidoses I/Hurler's disease – most common Mucopolysaccharidoses II/Hurler's syndrome GM1 gangliosidosis Mucolipidosis Niemann-Pick disease Mannosidosis Sjögren-Larsson syndrome
Neurocristopathies and vascular nevi	Phakomatosis pigmentovascularis Noninvoluting congenital hemangioma Sturge-Weber syndrome Klippel-Trenaunay syndrome Cutis marmorata telangiectatica congenita
Others	Nevus of Ota Down syndrome Leptomeningeal melanocytoma Spinal dysraphism

orders of IEM is increasing (table 1). MS in IEM show a generalized distribution involving the dorsal and ventral trunk in addition to the sacral region and extremities. These lesions are persistent and may also progress over time. The pigmentation is deeper as compared to the common MS [1, 3]. MS may mimic

a hematoma due to blunt force and may even be seen as a proof of recurrent trauma in the presence of "additional" differently colored hematoma [9].

The systemic association of MS forms the main rationale for an extensive evaluation of neonates with atypical MS as IEM are fatal and produce irreversible organ damage and subsequently a shorter life span. Some of these disorders respond well to stem cell transplantation or enzyme replacement therapy if instituted at an early stage, before irreversible organ damage occurs. Early palliative care decisions can be made. It also helps in identification of at-risk families and prevention of complications [1]. In our case, we carried out extensive physical and biochemical evaluation to rule out any IEM. Fortunately, none was detected. Parents have been advised to be on regular follow-up.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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