



Dermal melanocytosis

Mongolian spot refers to a macular blue-gray pigmentation usually on the sacral area of healthy infants. Mongolian spot is usually present at birth or appears within the first weeks of life. Mongolian spot typically disappears spontaneously within 4 years but can persist for life.

Pathophysiology

Mongolian spot is a congenital, developmental condition exclusively involving the skin. Mongolian spot results from entrapment of melanocytes in the dermis during their migration from the neural crest into the epidermis. This migration is regulated by exogenous peptide

growth factors that work by the activation of tyrosine kinase receptors. It is postulated that accumulated metabolites such as GM1 and heparan sulfate bind to this tyrosine kinase receptor and lead to severe neurologic manifestations and aberrant neural crest migration.

History

In Mongolian spot, an asymptomatic bluish discoloration overlying the sacrococcygeal area is present at birth.

Physical

- Mongolian spots consist of blue-gray macular pigmentation. The distinctive skin discoloration is due to the deep placement of the pigment in the dermis, which imparts a bluish tone to the skin from the Tyndall effect of scattered light

- Typically, it is a few centimeters in diameter, although much larger lesions also can occur. Lesions may be solitary or numerous.

- Most commonly, Mongolian spot involves the lumbosacral area, but the buttocks, flanks, and shoulders may be affected in extensive lesions.

- Generalized Mongolian spots involving large areas covering the entire posterior or anterior trunk and the extremities have been reported.

- Several variants exist, as follows:

- Persistent Mongolian spots are larger, have sharper margins, and persist for many years.²

- Aberrant Mongolian spots involve unusual sites such as the face or extremities.^{3,4}

- Persistent aberrant Mongolian spots also are referred to as macular-type blue nevi.

- Superimposed Mongolian spots, in which a darker Mongolian spot overlies a lighter one,⁵ have been described.

- Mongolian spots have been associated with cleft lip,⁶ spinal meningeal tumor, melanoma,⁷

phakomatosis pigmentovascularis types 2 and 5,

^{8,9,10,11,12,13}

and Sjögren-Larsson syndrome (one case report).

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A few cases of extensive Mongolian spots have been reported with inborn errors of metabolism, the most common being Hurler syndrome,

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followed by gangliosidosis type 1, Niemann-Pick disease, Hunter syndrome,

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and mannosidosis.

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In such cases, the Mongolian spots are likely to persist rather than resolve.

Causes

Mongolian spot is a hereditary developmental condition caused by entrapment of melanocytes in the dermis during their migration from the neural crest into the epidermis.

Usually, laboratory studies are not indicated except when extensive Mongolian spots are present. In these circumstances, it is advisable to evaluate the patient for inborn errors of metabolism in order to avoid irreversible organ damage.¹⁹

Imaging Studies

In extensive Mongolian spots involving the back, radiographic studies are needed to rule out a spinal meningeal tumor or anomaly.

Histologic Findings

Dendritic melanocytes with variably pigmented melanosomes typically are located in the deep reticular dermis. The melanocytes usually are oriented parallel to the epidermis. In contrast, the melanocytes in blue nevi are denser in number and more focally aggregated.

Medical Care

Opaque cosmetics may be used as camouflage for Mongolian spots.

Surgical Care

The value of lasers in Mongolian spot is uncertain. However, in a retrospective study of 26 Japanese patients, the Q-switched alexandrite laser showed better outcomes in extrasacral lesions treated at a younger age

Mongolian spots usually fade in the first year of life, but, at times, they may persist indefinitely.