Mongolian Spots Are Not Always a Benign Sign

A female infant who had been born at term to healthy consanguineous parents was examined at the age of 9 months for delayed developmental milestones. She exhibited generalized hypotonia, hepatosplenomegaly, and widespread Mongolian spots extending over the back and upper and lower extremities (**Figure**, A). Fundoscopic examination revealed cherry red spot of the retina (**Figure**, B). β-Galactosidase activity was undetectable in peripheral leukocytes, consistent with the diagnosis of GM1 gangliosidosis type 1. The most common lysosomal storage disease associated with generalized Mongolian spots is Hurler syndrome, followed by GM1 gangliosidosis type 1.[1-3] The Mongolian spots result from entrapment of melanocytes in the dermis because of arrested transdermal migration from the neural crest into the epidermis. Through activation of receptors with tyrosine kinase properties, exogenous peptide growth factors regulate this migration. Some experts observed that accumulated metabolite such as GM1 and heparan sulfate bind to this tyrosine kinase–type receptor, which enhances nerve growth factor activity and leads to aberrant neural crest migration. Because melanocytes have chemotropic receptors for nerve growth factor, metabolite–tyrosine kinase–type receptor binding may lead to arrested melanocyte transdermal migration.[1] ■

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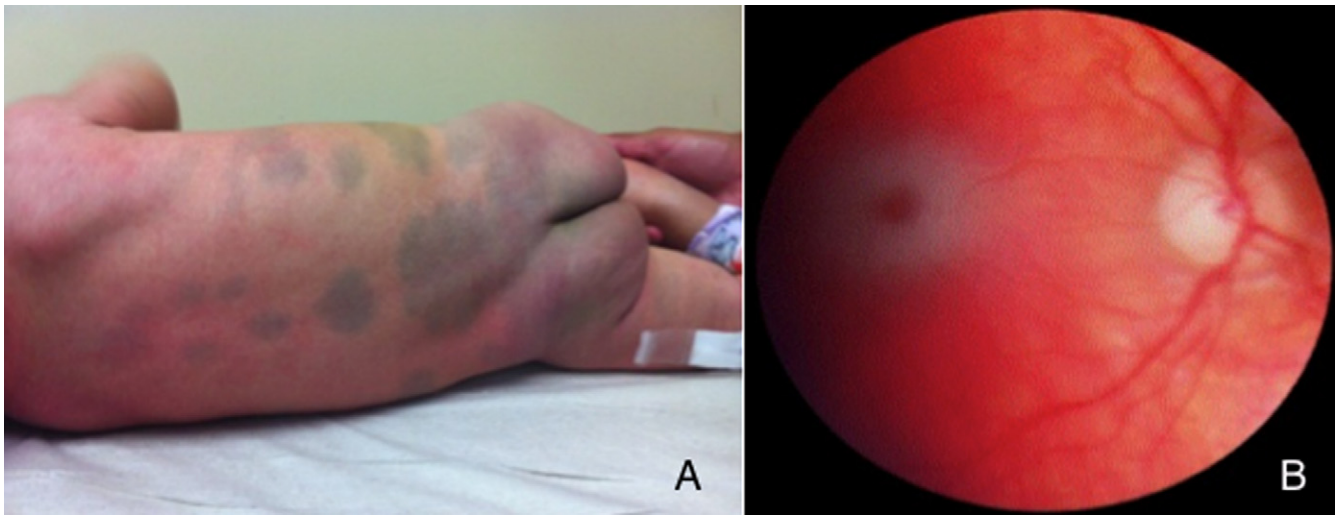

Figure. **A,** Extensive mongolian spots over the back, upper, and lower extremities. **B,** Cherry red spot of the retina.

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