Mongolian Spots as a Clue to Mucopolysaccharidoses

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Mongolian spots are irregular blue patches of skin pigmentation. Most commonly they are seen over the sacral area and buttocks, however, rest of the trunk and extremities may also be affected. They usually disappear by 6 months of age.

It is generally said that Mongolian spots have no clinical significance. However, it is a less known fact that unusually excessive Mongolian spots, especially at unusual sites as well as their unusual persistence (beyond 6 months of age) may be a clue to mucopolysaccharidoses, especially Hurler’s and Hunter’s syndrome.

The photo shows excessive Mongolian spots in a 1 year old female, who was diagnosed to be a Hurler’s syndrome.