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Case Report

Large Mongolian spots in GM1 gangliosidosis

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ABSTRACT

A 9-month old male child with GM1 gangliosidosis type 1 presented with Mongolian spots. The cutaneous lesions were present since birth before the appearance of the other features of the disease. Our patient, whose clinical course and physical signs were in keeping with GM1 gangliosidosis, had extensive Mongolian blue spots and this adds to the evidence supporting such an association.

Keywords: Mongolian blue spots, GM1 gangliosidosis

INTRODUCTION

GM1 gangliosidosis is an autosomal recessive lysosomal storage disease caused by deficiency of the lysosomal hydrolase, acid beta-galactosidase. The infantile form (type 1) is characterized by progressive organomegaly, dysostosis multiplex, facial coarsening and progressive neurologic deterioration within the first year of life. A variety of cutaneous signs have been described in children with GM1 gangliosidosis. We describe an infant with GM1 gangliosidosis after taking consent from his parents associated with Mongolian spots on the trunk and extremities with ventral and dorsal distribution.

CASE REPORT

A nine month old male child born by normal delivery to non-consanguineous parents, presented with developmental delay, coarse facial features and seizures. Coarse facial features included low set ears, broad nasal bridge, a long philtrum and frontal bossing, wide open fontanel and congenital hydrocele. He had hepatosplenomegaly, hypotonic with brisk tendon reflexes and global developmental delay. In addition, large hyper pigmented, well demarcated macules resembling Mongolian blue spots were scattered all over the body since birth. Ophthalmology examination showed cherry red spot but no corneal clouding.

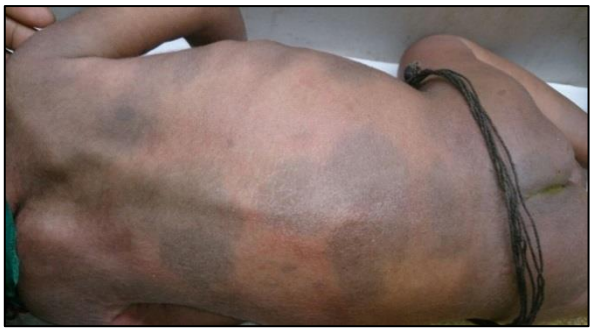

Figure 1: Mongolian spots on back of child.

Radiography showed rounding of vertebral body and proximal pointing of metacarpals and bullet shaped phalanges, iliac flare. MRI Brain showed bilaterally symmetrical confluent hyper intensities on T2/FLAIR involving periventricular; sub cortical white matter with involvement of sub cortical u fibre with mild brain atrophy and also involving bilateral internal and external capsule and bilateral cerebellar white matter. There is

evidence of mild T1hyperintensities in bilateral medial globuspallidus. Beta galactosidase assay was done in blood by flurometry and it showed deficient enzyme activity.

Figure 2: Coarse facial features and congenital hydrocele in child.

DISCUSSION

Clinical and biochemical evidence supported the diagnosis of GM1 gangliosidosis type 1 in our patient. He had numerous and diffusely distributed hyper pigmented lesions which is an unusual presentation for typical Mongolian spots. Mongolian blue spots or dermalmelanocytosis are birth marks with wavy borders and irregular shapes and are seen in up to 90% of Asian babies, and less commonly in other races. They are benign and have no known associations. Histologically they are characterized by melanocyte proliferation in the mid dermis. They are usually lumbo-sacral and can be single or multiple. They can increase in both size and density but usually disappear by the age of 5 years. Extensive Mongolian blue spots do certainly occur in up to 5% of cases although their extent and natural history have not yet been studied. The Mongolian blue spots seen in our child were both very extensive (including the ventral surface of the trunk and extremities) and unusual in that they were rapidly increasing at the age of 6 months. Dermatologic findings are not commonly described in GM1 gangliosidosis. Eczematoid facial rash, truncal macular rash, angiokeratomas and generalized telangiectasia, in patients with this illness, have been sporadically described in the literature.[1-4] However, diffuse, extensive and unusual Mongolian spots have been reported in increasing number of cases of GM1 gangliosidosis type 1 in recent years.[5-7]

Table 1 summarizes the reported cutaneous findings in these patients. This association has also been described with other lysosomal storage diseases such as Hurler's and Hunter's syndromes.[11,12]

Table 1: Cutaneous findings reported in children with GM1 gangliosidosis.

Source	Year	Gender	Ethnicity	Age*	Lesion
Landing et al.[1]	1964	NA	NA	NA	Rash
Hoof et al.[2]	1969	NA	NA	NA	Telangiectasia
Ginsburg et al.[3]	1977	NA	NA	NA	Telangiectasia
Beratis et al.[4]	1989	M	Greek	3+	Mongolian spot and angiokeratoma
Weissbluth et al.[5]	1981	F	AA	5	Mongolian spot
Selsor et al[6]	1989	M	AA	10	Mongolian spot
Beattie et al.[7]	1992	F	Pakistani	5	Mongolian spot
Tang et al.[8]	1993	F	AA	13	Mongolian spot
Silengo et al.[9]	1999	F	Caucasian	2	Mongolian spot
Hanson et al.[10]	2003	M	NA	NA	Mongolian spot
Ashrafi et al.	2006	M	Iranian	12	Mongolian spot and ecchymosis

NA = Not available; M = Male; F = Female; AA = African American
*Age in months at the time of documented presentation
+Angiokeratoma as appeared between age 3 and 10 months

Weissbluth et al. reported the first case of possible chance association between GM1 gangliosidosis type 1 and extensive Mongolian spots in a 5-month-old female.[5] Selsor et al. reported a 10-month-old male with GM1 gangliosidosis type 1 who also had hyperpigmented macules and patches which were most probably Mongolian spots.[6] Beattie et al. described a 5-month-old female with GM1 gangliosidosis who had unusual Mongolian blue spots on her dorsal and central trunk.[7] Tang et al. presented a 13-month old child with GM1 gangliosidosis who had multiple Mongolian spots and further demonstrated swelling of the endothelial cells of the dermal capillaries with narrowing of the vascular lumen. The authors postulated that this may lead to weakening and dilatation of the vascular walls resulting in angiokeratoma and telangiectasia in these patients.[8] Hanson et al. described two infants with extensive dermal

melanocytosis in association with GM1 gangliosidosis type 1 in one and with Hurler's syndrome in the other. They hypothesized that the accumulating metabolites in these illnesses may contribute indirectly to the arrest of the transdermal migration of melanocytes within the dermis leading to the appearance of these cutaneous findings. This may occur through interference with neural growth factor and tyrosine kinase-type receptor interactions.[11] Ochiai et al. described seven Japanese boys with Hunter's syndrome and reported extensive Mongolian spots in all of them; the authors suggested that the extent and persistence of the hyperpigmentation could allow earlier diagnosis and possible intervention before irreversible nervous system impairment develops.[12] These children therefore suggest that there might be an association of GM1 gangliosidosis type1with extensive and unusual Mongolian blue spot.

CONCLUSION

May be Mongolian spots are just association of GM1gangliosidosis but we think our patient adds to the evidence that patients with this disorder may manifest abnormal dermal pigmentary lesions, which may be present since birth thus helping in early diagnosis. Small, light blue-green coloured spots confined to lumbosacral area can be ignored but extra sacral, extensive, persistent and dark coloured spots should be looked upon with suspicion, especially in the presence of a consanguineous marriage or a strong family history of storage disorders. Future research should focus on further quantifying and validating parameters like size, percentage of Total body surface area, location and colour of Mongolian spot, as markers for IEMs and their place in screening and diagnosis of these syndromes.

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