

Journal of Pediatric Sciences

Cutaneous Manifestations of GM 1 Gangliosidosis Type 1

Vidyadhar Sardesai, Trupti Agarwal

Journal of Pediatric Sciences 2013;5:e199

How to cite this article:

**Sardesai V, Agarwal T. Cutaneous Manifestations of GM 1 Gangliosidosis Type 1.
Journal of Pediatric Sciences. 2013;5:e199**

CASE REPORT

Cutaneous Manifestations of GM 1 Gangliosidosis Type 1

Vidyadhar Sardesai, Trupti Agarwal

Department of Dermatology, Venereology & Leprosy, Bharati Vidyapeeth University,
Medical College & Hospital, Pune 411043, India.

Abstract:

GM1 Gangliosidosis is a rare autosomal recessive disorder characterized by deficiency of lysosomal enzyme ganglioside β -galactosidase. We present a 9 month old male child with diffuse ecchymoses, mongolian spots with other clinical features and investigations suggestive of GM1 Gangliosidosis.

Keywords: GM1 Gangliosidosis, ecchymoses, mongolian spots

Corresponding author: V.R.Sardesai – 102, Alliance Nakshatra, 48 Tulshibagwale Colony, Sahakarnagar no. 2, Pune 411009, India

Telephone : +91-020-24227763

e-mail: nitin_sardesai@yahoo.com

Introduction

GM1 Gangliosidosis is a rare autosomal recessive disorder characterized by deficiency of lysosomal enzyme ganglioside β -galactosidase. It can be associated with dermatological manifestations.

Case report

A 9 month old male child was born to second degree consanguineous parents at full term by normal vaginal delivery with bluish colored macules. The child was brought to the hospital with complaints of fever and breathlessness since 10 days. He was immunized till date. He had recurrent lower respiratory tract infections, seizures and delayed milestones in the past 5 to 6 months. There was no history of birth trauma, asphyxia, or family history of similar complaints. General and systemic examination revealed pallor, respiratory rate 25/minute, generalized lethargy, hypotonia with brisk

reflexes suggestive of upper motor neuron involvement, hepatosplenomegaly, air entry decreased on right side with crepitations. On dermatological examination Mongolian spots (Figure 1) and diffuse ecchymosis (prominent over the placement of ECG chest leads) (Figure 2) were present on trunk and extremities. There was frontal bossing, depressed nasal bridge, long philtrum and low set ears (Figure 2). Occipito-frontal diameter was increased to 51 cm (normal 45 cm for a 9 month old infant). Ophthalmologic examination showed bilateral macular cherry red spots on funduscopy.

Haemoglobin was 7mg/dl with a total leukocyte count of 15,000/cmm. Chest radiograph revealed consolidation on right side. Ultrasound of the abdomen showed hepatosplenomegaly. Bone marrow examination revealed large foam cells and ballooned



Figure 1. Diffuse ecchymoses and Mongolian spots on dorsal surface of the trunk



Figure 2. Frontal bossing, depressed nasal bridge, long philtrum, low set ears and ecchymoses at site of ECG leads.

cells suggestive of lysosomal storage disorder. Computed Tomography Scan and Magnetic Resonance Imaging of Brain showed diffuse cortical atrophy and white matter demyelination. On Wood's lamp examination, accentuation of dermal pigmentation was seen. Enzyme assay in leukocytes confirmed deficiency of acid β -galactosidase. The normal value for the β -galactosidase ranges from 80 – 480 nanomol/mg protein/hour. This was decreased to 50 nanomol/mg protein/ hour. A skin biopsy revealed dermal dendritic melanocytes as seen in Mongolian spots. Liver and renal function tests were within normal limits. No abnormality was detected in bleeding time, platelet count, prothrombin time, partial thromboplastin time and fibrinogen level. There was no history of any bleeding tendency.

The baby was treated with antibiotics and anti-epileptic drugs, managed with nasogastric tube feeding, oxygen and physiotherapy. The condition of the patient improved temporarily. The family insisted on discharge against medical advice and the patient was lost for follow up.

Discussion

GM1-gangliosidosis is a rare autosomal recessive lysosomal storage disorder caused by a deficiency of β -galactosidase [1]. The estimated incidence is 1:100,000-200,000 live births [2].

The most common infantile form is evident at birth with coarse facial features, hepatosplenomegaly, generalized skeletal dysplasia, macular cherry-red spots, and developmental delay or arrest, as seen in our case. Death usually occurs during the second year of life because of infection and cardiopulmonary failure [3]. As many as 50% of affected infants have a macular cherry-red spot [1,2].

Dermatological findings are uncommon in GM1 gangliosidosis [4]. The first case of association between GM1 gangliosidosis type-1 and dermal melanocytosis was described by Weissbluth et al. [5]. Diffuse, extensive and unusual Mongolian spots have been reported in increasing number of cases of GM1 gangliosidosis type-1 in recent years [3,5]. Mongolian spots are benign and common having no known associations and histologically characterized by melanocyte proliferation in the mid dermis [3]. Hanson et al. 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 [6]. The most common lysosomal storage disease associated with generalized Mongolian spots is Hurler syndrome followed by GM1 gangliosidosis type-1. A literature review revealed 39 cases of lysosomal storage disease associated with dermal melanocytosis. 24 cases had Hurler disease and 11 patients were suffered from GM1 gangliosidosis type-1. An association with Niemann-pick disease, hunter and α mannosidosis was also reported. In these conditions hyperpigmentation is a long lasting symptom [6].

Some other dermatological manifestations of GM1 are angio-keratomas, forehead hypertrichosis, thick skin texture, facial and peripheral edema in early infancy [7].

Prenatal diagnosis can be done by estimation of Acid β -galactosidase level in cultured amniocytes and chorionic villi sampling.

There is no effective medical treatment.

The aim is to report a rare disease having characteristic cutaneous manifestations, which can be prevented by prenatal diagnosis and genetic counseling.

References

1. Suzuki Y, Oshima A, Namba E. β -Galactosidase deficiency (β -galactosidosis). GM1 gangliosidosis and Morquio B disease. In: Scriver CR, Beaudet AL, Sly WS, Valle D, Childs B, Kinzler KW, et al., eds. The metabolic and molecular bases of inherited disease. New York: McGraw-Hill, 2001: 3775–3809.
2. Brunetti-Pierri N, Scaglia F. GM1 gangliosidosis: review of clinical, molecular, and therapeutic aspects. *Mol Genet Metab*. 2008; 94:391-6.
3. Beattie RM, Harvey D. Extensive and unusual Mongolian blue spots in a child with GM 1 gangliosidosis type one. *J R Soc Med* 1992; 85:574-5.
4. Dweikat I, Libdeh BA, Murrar H, Khalil S, Maraqa N. Gm1 gangliosidosis associated with neonatal-onset of diffuse ecchymoses

- and mongolian spots. *Indian J Dermatol.* 2011; 56:98-100
5. Weissbluth M, Esterly NB, Caro WA. Report of an infant with GM1 gangliosidosis type I and extensive and unusual mongolian spots. *Br J Dermatol* 1981; 104: 195–200.
 6. Ashrafi MR, Shabani R, Mohammadi M, Kavusi S. Extensive Mongolian spots A clinical sign merits special attention *Pediatr Neurol.* 2006; 34:143-5.
 7. Hanson M, Lupski JR, Hicks J, Metry D. Association of dermal melanocytosis with Lysosomal storage disease. *Arch Dermatol* 2003; 139:916-20.