Diffuse neonatal mastocytosis with possible Autosomal Dominant inheritance

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Abstract

Diffuse cutaneous/neonatal mastocytosis (DCM) is a very rare form of cutaneous mastocytosis with marked pruritus, blistering after minor trauma or scratching and thickened skin with a peau d' orange appearance. DCM almost exclusively occurs in neonates and infants. It may persist into adult life. Most cases are sporadic. However autosomal dominant inheritance with familial occurrence has been recognized.

Diagnosis is mainly clinical and can be confirmed with histology. Treatment is unsatisfactory and directed mainly to alleviate the symptoms. Here we report a case of diffuse neonatal mastocytosis with possible autosomal dominant inheritance.

A seven months old child, a product of non consanguineous marriage presented with spontaneous blistering associated with intense pruritus. Examination revealed indurated leathery skin mainly on lower back, multiple erosions on face and scalp and few vesicles on trunk. His father also had been affected similarly in his early childhood. Diagnosis was made clinically and confirmed by skin biopsy.

Case history

A seven months old baby, a product of a non consanguineous marriage presented with blistering since the age of two months. Blisters were both spontaneous and trauma induced. There was generalized blistering with marked involvement of scalp and associated intense pruritus. He had no evidence suggestive of systemic involvement. Examination revealed leathery indurated skin on lower back (Figure 1), fresh erosions and few scattered pigmented macules on face and trunk and prominent truncal creases (Figure 2 & 3).

His father gave a similar history starting in infancy. Examination of the father revealed leathery indurated skin with peau d' orange appearance, most apparent on trunk and neck (Figure 4).

The clinical diagnosis of DCM was made and confirmed with skin biopsy which showed profuse mast cell infiltration in the papillary dermis (Figure 5). He was started on chlorpheniramine to alleviate pruritus. Skin lesions were managed with topical steroids and antibiotics when necessary. Parents were educated to carry the diagnosis card with detailed list of possible triggers of the disease.

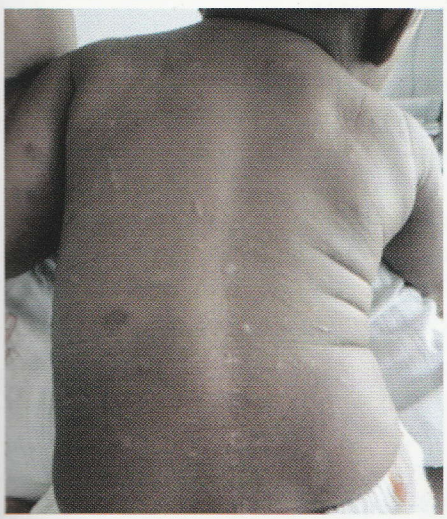

Figure 1.

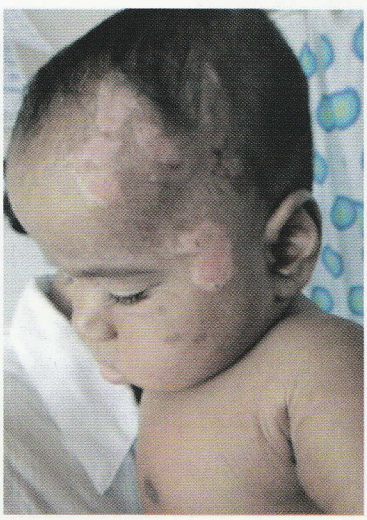

Figure 2.

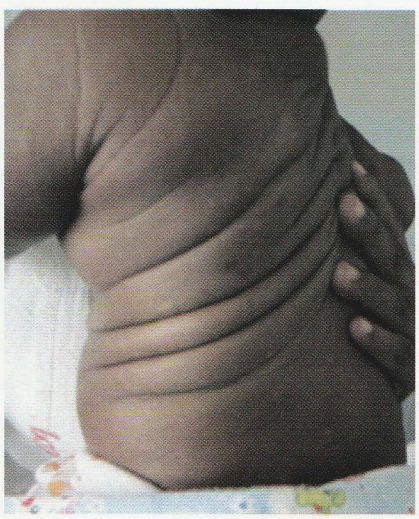

Figure 3.

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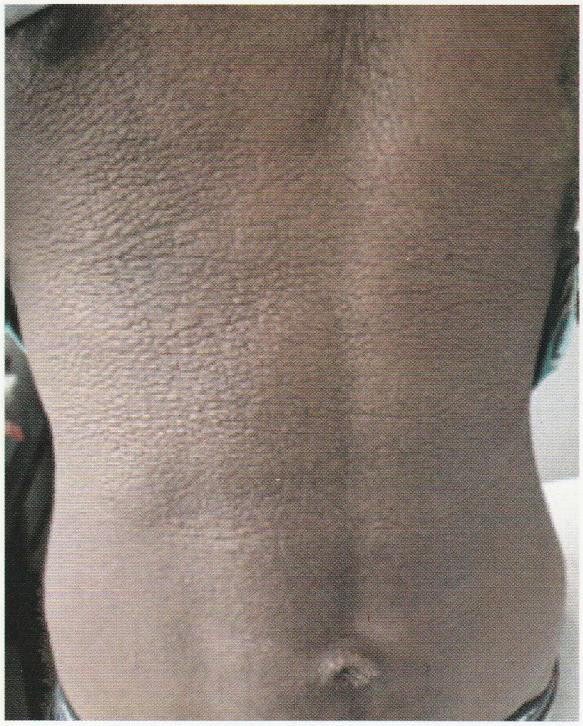

Figure 4.

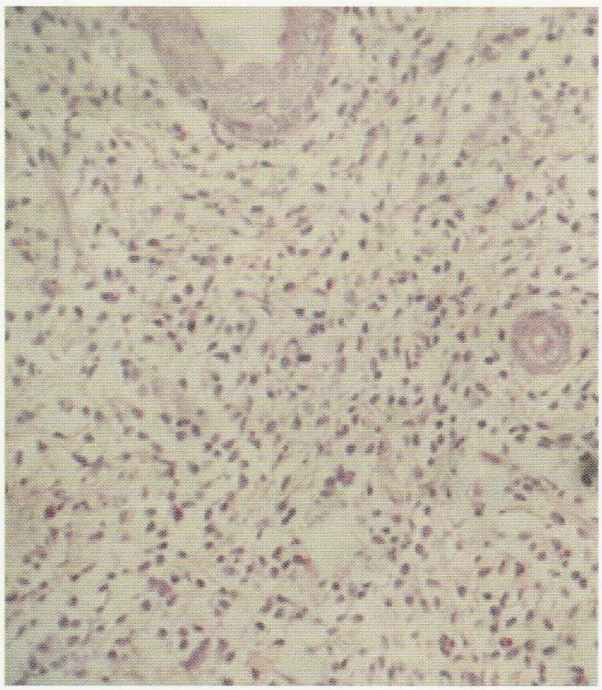

Figure 5.

Discussion

The term mastocytosis was first described by Nettleship and Tay in 1869. Mastocytosis is of two types, cutaneous and systemic. DCM is a very rare form accounting for 1-2% of all cases of cutaneous mastocytosis. DCM almost exclusively presents during neonatal period and infancy. It may persist into adult life. The disease is usually sporadic but few familial cases have been reported with autosomal dominant inheritance.

Two clinical types have been identified: bullous DCM and pseudoxanthomatous[6]. DCM usually presents with spontaneous or trauma induced blistering and indurated leathery skin with peau d' orange appearance. Pigmented lesions are usually absent. Other cutaneous manifestation includes urticaria, dermographism and positive Darier's sign. Systemic complications (flushing, headache, palpitation and abdominal pain) are more likely to occur with extensive cutaneous involvement. Most serious complications are gastrointestinal haemorrhage and shock.

Diagnosis is mainly clinical, confirmed by histological examination using mast cell stains (Giemsa and Toluidine blue) or immunohistochemical staining for tryptase or kit. Full thickness infiltration of skin or band like infiltration of papillary dermis with mast cells is observed.

DCM should be considered in the differential diagnosis of epidremolysis bullosa and poikiloderma of Kindler.

No effective treatment is available. Principles of managing these patients are avoidance of triggers for mast cell degranulation, symptomatic relief and early detection of systemic complications. Antihistamines and mast cell stabilizers and potent topical steroids are commonly used in the management of DCM. Phototherapy with PUVA or NB UVB alleviates pruritus through reduction of mast cell density in skin. This has a place in treatment of advanced DCM.

References

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