



Urticaria Pigmentosa in a Child: A Case Report

Authors

Dr Sheethal B, Dr Sankar S, Dr Jayalekshmi P.L

Government Medical College, Kottayam, Kerala

Abstract

Mastocytosis is a clonal disease of hematopoietic stem cells characterized by local or diffuse increased growth and accumulation of mast cells in skin, bone marrow, liver, spleen and lymph nodes. Urticaria pigmentosa is the most common variant of cutaneous mastocytosis. We herein report of urticaria pigmentosa in a 9 year old child.

Keywords: Mastocytosis, Urticaria pigmentosa, Mast cells.

Introduction

Mastocytosis is a clonal disease of hematopoietic stem cells characterized by local or diffuse increased growth and accumulation of mast cells in skin, bone marrow, liver, spleen and lymph nodes.

It can be divided into cutaneous mastocytosis and systemic mastocytosis.

The most common type of cutaneous mastocytosis in children is urticaria pigmentosa with an approximate incidence of 1/1000-8000. The disease is clinically characterized by multiple erythematous and pigmented macules, papules and plaques and local blistering that may vary in size.

The clinical manifestations of mastocytosis are not directly due to mast cells but due to the functional effects of mast cell degranulation enzymes.

The presentation of pediatric mastocytosis is varied and differs clinically from adult onset. Pediatric mastocytosis has a benign nature and usually resolves spontaneously by puberty.

A typical case of this dermatosis is being presented.

Case Report

A 9 year old male child presented with generalized eruption of multiple brown maculopapular lesions on face, trunk and extremities of 5 year duration. There was also a history of generalized urticarial flushing. The general health, growth and development of the child was unaffected. There was no family history of similar disease or any other dermatological or autoimmune disease.

Physical examination was normal. Systemic examination also revealed no abnormality.

Dermatological examination of the child revealed multiple, sharply defined, red brown maculopapules on face, trunk and extremities. On stroking the individual lesions, there was formation of wheal and flare (Darier sign – positive). A complete blood count, liver and renal function tests and ultrasonography of the abdomen were normal.

A 3 mm punch biopsy specimen was sent for histopathological examination. It revealed diffuse infiltration of mast cells in upper one third of dermis.

The child is on treatment with antihistaminics and topical steroids for 4 weeks and his lesions are regressing. The child is under follow up.



Figure 1 and 2: Urticaria pigmentosa showing multiple well-defined hyperpigmented plaques on the trunk

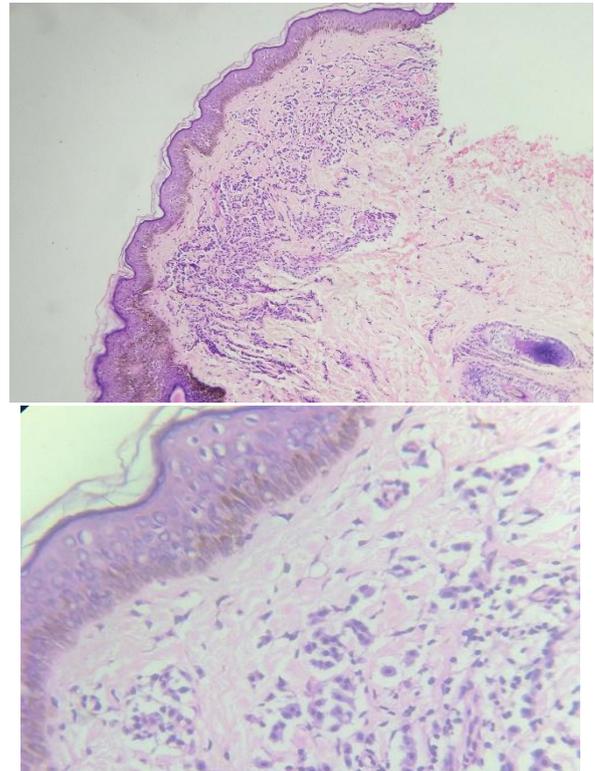


Figure 3: Epidermis showing increased basal layer pigmentation with perivascular lymphohistiocytic infiltrate in the dermis (H and E $\times 10$ and $\times 40$)

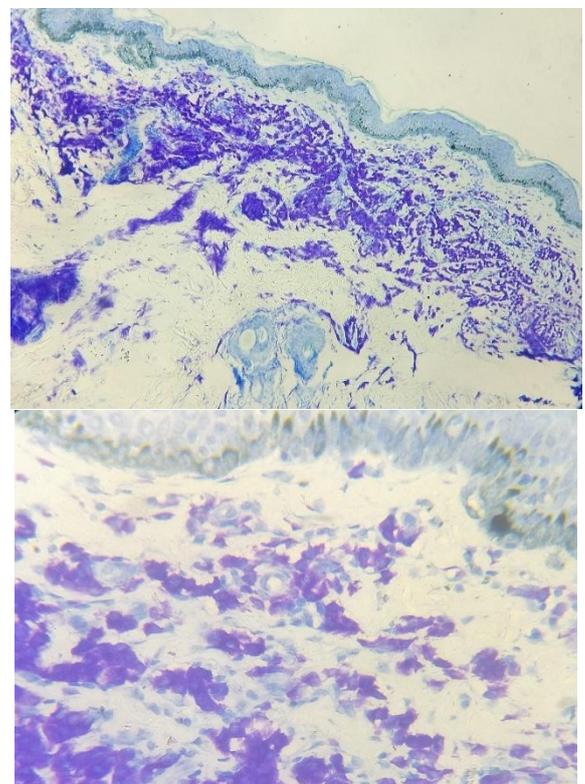


Figure 4: Toluidine blue stain showing multiple perivascular mast cells in the dermis (H and E $\times 10$ and $\times 40$)

Discussion

Mastocytosis was first described by Nettleship and Tay in 1869 and later due to the appearance like urticaria, the term urticaria pigmentosa (UP) was coined by Sangster in 1878. Their patient was a female child of 2 years and their paper was entitled "Chronic Urticaria Leaving Brown Stains of Nearly Two Years Duration". Ellis in 1949 while doing an autopsy in a child with fatal UP documented the presence of mast cell infiltration in skin, liver, spleen, lymph nodes, and bone marrow.

Mastocytosis occurs at any age. Cutaneous mastocytosis can present during the neonatal period, in infancy or childhood. There is no sex predominance and is seen in all races. Familial cases have been reported, and mastocytosis in identical twins and triplets have been described. However systemic mastocytosis frequently occurs in adults and tend to persist permanently.

The clinical features are multiple oval or round hyper pigmented macules, papules or patches, brown red and 2-4 mm in diameter. These lesions urticate by manipulation (e.g. rubbing) or spontaneously. This reaction is Darier sign. Darier's sign is not always demonstrable, especially in those with a long history of this disorder and is not 100% specific for mastocytosis since it has been described rarely in Juvenile xanthogranuloma and ALL of neonate. The sites of predilection are chest and dorsal areas of body. Systemic manifestations include flushing, vomiting, diarrhea, tachycardia, headache, weight loss and wheezing. Histologically there is mild to moderate perivascular infiltrate with dendritic mast cells in the papillary dermis sometimes extending to the subcutis, especially with special stains like toluidine blue and chloroacetate esterase.

Mastocytosis represents a spectrum of clinical disorders with clinical features determined by infiltration of various organs and skin with mast cells.

In 2001, four distinct clinical variants of cutaneous mastocytosis were published by WHO. These were

1. Urticaria pigmentosa
2. Isolated mastocytoma
3. Diffuse cutaneous mastocytoma
4. Telangiectasia Macularis Eruptiva Perstans (TMEP).

In 2008, WHO updated the classification of cutaneous mastocytosis as

1. Maculopapular cutaneous mastocytosis (Urticaria pigmentosa)
2. Diffuse cutaneous mastocytosis
3. Solitary mastocytosis

Mast cells are connective tissue cells that play a role in immunologic and inflammatory reactions. They originate from pluripotent progenitor cells in the bone marrow that express CD34, CD117 (kit), and CD13. These cells migrate in the blood and invade the tissues where they proliferate and differentiate into mature mast cells. The growth and development of mast cells is influenced by Stem cell factor (SCF)/Mast cell growth factor /kit ligand which binds to the human kit protein, the receptor for SCF on the surface of human mast cells.

There is increased expression of soluble SCF in lesions of cutaneous mastocytosis. Dysregulation of mast cell apoptosis is another pathogenesis occurring in mastocytosis and this is seen in systemic mastocytosis.

The clinical symptoms of mastocytosis are due to the release of mast cell mediators. They produce mediators histamine, heparin, tryptase, proteases, chymase, carboxypeptidase A, chondroitin sulfate, glycosaminoglycans, leukotrienes, prostaglandin D₂, vascular endothelial growth factor, platelet activating factor, cytokines (tumor necrosis factor and interleukins) and chemokines. These mediators related symptoms are flushing, itching, blistering, diarrhea, abdominal pain, vomiting, hypotension, headache, and bone pain.

Histopathological examination is the gold standard for diagnosis. All types of mastocytosis show increased number of mast cells in the dermis. In UP and mastocytoma, there is diffuse infiltration of mast cells in upper one-third of dermis along with perivascular aggregates. There is increased melanin in the basal layer and melanophages in the upper dermis. It is important to choose a biopsy site that has not been traumatized as there will be degranulation of mast cells and histopathology may not be evident of mastocytosis.

Special stains are required to demonstrate mast cells. Giemsa stain, toluidine blue or Leder's stain are special stains that identify metachromatic cytoplasmic granules in mast cells. Giemsa and toluidine blue stain metachromatic granules purple and Leder's stain used stain red colour.

The diagnosis of mastocytosis is mainly clinical. A high index of suspicion is necessary to recognize the skin lesions of mastocytosis and this should be corroborated with other features like positive Darier's sign and investigations. Serum tryptase levels correlate with the numbers in the skin, and increased levels are seen in severe disease. There is a positive correlation between the extent of skin involved, physical appearance of the lesions, associated symptoms such as pruritus, flushing, positive Darier's sign, bone pain, diarrhea, and tryptase levels.

Treatment of UP is aimed at three aspects: Counseling, avoidance of triggers known to stimulate mast cell degranulation and suppressing skin and mast cell mediator related symptoms.

Cutaneous mastocytosis should be followed once in 6 months to 1 year, and investigations must be repeated.

References

1. Srinivas S, Dhar S, Parikh D. Mastocytosis in children. *Indian Journal of Paediatric Dermatology*. 2015;16(2):57.
2. Keen M. Urticaria pigmentosa: A case report. *Nepal Journal of Dermatology*,

Venereology & Leprology. 2016;13(1):57-60.