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CASE REPORT

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Case Report of Systemic Mastocytosis with Cutaneous Involvement

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ABSTRACT

Mastocytosis is a rare clonal disease of hematopoietic stem cells characterized by the accumulation of mast cells in the skin (cutaneous mastocytosis, CM) or in multiple organs such as the bone marrow, liver, spleen, and lymph nodes (systemic mastocytosis, SM). In this case report, we present a case of systemic mastocytosis with cutaneous involvement.

Keywords: Systemic mastocytosis, cutaneous mastocytosis, idiopathic anaphylaxis, tryptase

INTRODUCTION

Mastocytosis is a rare clonal disease of hematopoietic stem cells characterized by the accumulation of mast cells only in the skin (cutaneous mastocytosis, CM) or in multiple organs such as the bone marrow, liver, spleen, and lymph nodes (systemic mastocytosis, SM) (1). The clinical picture is quite heterogeneous and is usually limited to the skin with benign behavior in childhood, whereas adult patients present with systemic disease with or without skin lesions (2). Systemic mastocytosis is divided into different subtypes ranging from slowly progressing diseases with the risk of anaphylaxis to aggressive mast cell neoplasms (3,4).

In our case report, we tried to present a patient diagnosed with systemic mastocytosis with cutaneous involvement.

CASE REPORT

A 43-year-old male patient presented to our emergency department with complaints of sweating, vomiting, and syncope. His vital signs at admission were as follows; blood pressure: 100/60 mmHg, pulse rate: 66 beats per minute, temperature: 36.8°C, blood glucose: 99 mg/dl, together with loss of consciousness. Laboratory values, electrocardiogram, echocardiogram, lung and brain tomography, and cardiac, respiratory and neurologic evaluation revealed no abnormalities that could explain the hypotension and syncope. Since hypotension and syncope were present, he was followed up with a diagnosis of anaphylaxis. Adrenaline (0.5 mg) was administered intramuscularly, and pheniramine (45 mg) ampoule and methylprednisolone (60 mg) ampoule were administered intravenously 2 times with an interval of 15 minutes. After this treatment, blood pressure values returned to normal, symptoms of nausea and vomiting regressed, and the consciousness improved.

The patient had no history of any drug use, suspicious food intake, or bee/insect sting before the sudden onset of these symptoms and signs. In his medical history, it was learned that he had intermittent episodes of flushing, sweating with flushing, and a history of anaphylaxis 3 times after bee sting 10 years ago. Physical examination revealed multiple reddish brown macules on the back and upper extremities which had been present for about 10 years (Figure 1), and Darier's sign was positive (Figure 2). Skin biopsy could not be performed because the patient refused biopsy. Since the reddish brown maculopapular appearance, Darier's sign, tryptase elevation, and bone marrow biopsy were compatible with mastocytosis, the

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patient was diagnosed with cutaneous involvement of indolent systemic mastocytosis.

No significant abnormality was found in initial laboratory values. Laboratory results are shown in Table I. Abdominal ultrasonography revealed mild splenomegaly (130x72 mm).

Table I: Laboratory values of the patient

	Patient's values	Normal value
Wbc (10^3 /μL)	8.36	4.8-10.7
Platelets (10^3 /μL)	199	130-400
Haemoglobin (g/dL)	15.1	14-18
Eosinophil (10^3 /μL)	0.04	0-0.2
CRP (mg/L)	0.73	0-5
Sedimentation (mm/s)	5	0-20
Quantitative Ig E (IU/mL)	1.16	0-100
B 12 (pg/mL)	204	197-771
BUN / Creatinine (mg/dL)	11.4 / 0.8	6-20 / 0.5-1.20
ALT / AST (u/L)	13 / 14	0-41 / 0-40
Glucose (mg/dL)	99	74-106
Albumin / Protein (g/dL)	4.98 / 7.45	3.5-5.2 / 6.4-8.3
Troponin T (ng/L)	3.65	0-14
TSH (μIU/mL)	1.05	0.27-4.20
LDH (u/L)	156	135-250
D-Dimer (μg/L)	390	0-550

Due to the history and cutaneous involvement, the total serum tryptase level was analyzed on the 20th day after the attack with the suspicion of mastocytosis and was found to be very high (154 ug/L; Normal range: < 11.4 ug/L). Subsequent bone marrow (BM) biopsy and microscopic examination revealed 80% cellularity and diffuse mast cell infiltration, while histochemical staining revealed CD117+ / CD25+ / CD2 pale intensive positive / CD30 - / CD34- . The diagnosis of systemic mastocytosis was confirmed due to the clinical history, compatible skin lesions, positive Darrier's sign, tryptase elevation, and compatible BM biopsy.

Bone densitometry for additional evaluation revealed mild osteodegenerative changes in the femoral neck. Genetic testing for myeloproliferative malignancies was negative. Since the patient had no additional system involvement, indolent systemic mastocytosis with cutaneous involvement was accepted. Mast cell stabilizer (ketotifen) and adrenaline autoinjector for possible anaphylactic shock were prescribed. A consent form was obtained from the patient for this case report.

DISCUSSION

Mastocytosis is a heterogeneous disease characterized by the accumulation of morphologically and immunophenotypically abnormal mast cells in one or more organs (5,6). The clinical presentation of mastocytosis is quite heterogeneous and is limited to the skin (cutaneous

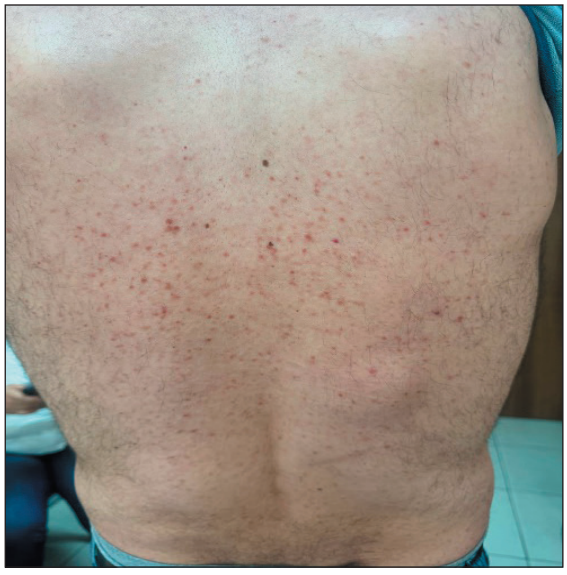

Figure 1. The lesions on the patient's back (Picture is presented with the permisison of the patient).

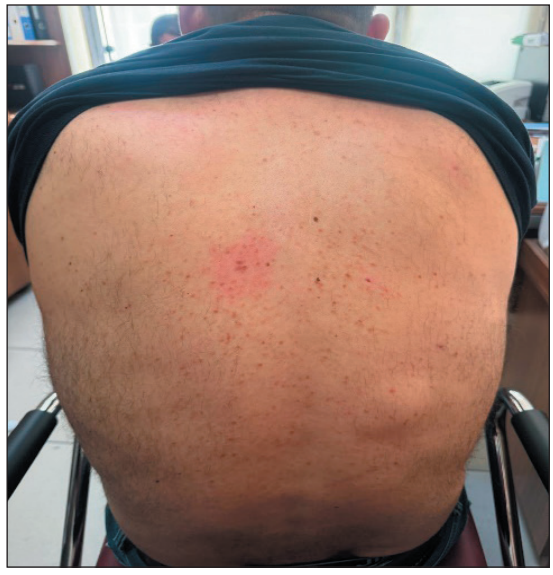

Figure 2. Darier's sign (Picture is presented with the permisison of the patient).

mastocytosis, CM) with onset in the first 2 years of life in childhood and spontaneous regression of skin lesions usually in adolescence (6). Most adult patients with cutaneous lesions suffer from systemic mastocytosis. Therefore, investigation for CM is always recommended in adult patients (4).

In adult patients, the clinic extends to a more aggressive variant with extracutaneous involvement. Patients may present with various symptoms including skin itching, flushing, nausea, diarrhea, chronic abdominal cramps, syncope, palpitations, depression, headache, recurrent unexplained anaphylaxis, osteopenia, osteoporosis, weakness, fatigue, and arthralgia as mast cell mediator release symptoms (6, 7). In addition, the rate of Hymenoptera venom allergy is higher in these patients compared to the general population (8). The fact that our patient had frequent episodes of hot flushes, pruritus, and a history of anaphylaxis with bee venom is compatible with what has been reported in the literature.

In conclusion, as stated in the WHO 2023 mastocytosis guidelines, the diagnosis of SM with cutaneous involvement has now taken its place in the literature. When cutaneous mastocytosis is diagnosed in an adult patient, additional investigations for systemic involvement should be initiated. Serum tryptase can be used as a screening test for systemic mastocytosis and persistent elevations should encourage further investigations such as bone marrow studies.

Conflict of Interest

The authors have no relevant conflict of interest concerning this work.

Author Contributions

Concept: **Elif Acar, Murat Turk, Insu Yilmaz,** Design: **Elif Acar, Murat Turk, Insu Yilmaz,** Data collection or processing: **Elif Acar, Murat Turk, Insu Yilmaz,** Analysis or Interpretation: **Elif Acar, Murat Turk, Insu Yilmaz,** Literature search: **Elif Acar, Murat Turk, Insu Yilmaz,** Writing: **Elif Acar, Murat Turk, Insu Yilmaz,** Approval: **Elif Acar, Murat Turk, Insu Yilmaz.**

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