SKIN MANIFESTATIONS OF INTERNAL DISEASE

MAST CELL ACTIVATION SYNDROME: AN UNDERRECOGNIZED CAUSE OF SKIN RASH AND SYSTEMIC SYMPTOMS

Angeliki Liakopoulou [1] - Georgios Bethimoutis [1] - Emanuele Verga [1] - Roberto Verdolini [1]

Princess Alexandra Hospital, Dermatology, Harlow, United Kingdom [1]

Background: The term Mast Cell Activation Disease (MCAD) is used to describe a vast spectrum of disorders characterized by accumulation of pathological mast cells (MC) in potentially every organ or tissue and/or aberrant or “unprovoked” release of mediators from hyperactive, over-responsive MCs. Mast Cell Activation Syndrome (MCAS) represents a commonly unrecognized spectrum of MCAD that fits the complex clinical picture of MC mediators-induced symptoms but fails to fulfil the criteria for Systemic Mastocytosis (SM).

Observation: A 61-year old female presented with intermittent severe erythema of the face and anterior chest. The patient had an uneventful familiar and personal past medical history and was physically and mentally well until the onset of her symptoms. The course of her condition was dramatic: within 5-6 weeks since the onset of facial erythema, the patient started having palpitations and malaise, followed by severe hypotensive episodes, vomiting, diarrheas, anaphylactic/syncope episodes and weight loss. She experienced a dramatic decrease in bone mineral density and her eyesight acuity dropped. She also suffered chronic sinusitis, recurrent conjunctivitis and dental decay. Quite rapidly she became lethargic. Blood tests, skin and bone marrow biopsies were normal. The patient was diagnosed with MCAS. Treatment with antihistamines, steroids and Montelukast was ineffective and the patient eventually responded to treatment with Imatinib.

Key message: Since a quite large number of mutations in MC regulatory elements has been detected in MCAS patients with no clear genotype-phenotype patterns, it is important to remember that such a large mutational heterogeneity is likely to drive the heterogeneity of aberrant MC expression, in turn causing the extreme heterogeneity of clinical manifestations and symptoms found in MCAS. The diagnosis often is just clinical and should be suspected when a quite broad spectrum of symptoms appears in a relatively short time, with laboratory tests often be negative.