TWO CASES OF MAST CELL ACTIVATION DISORDER

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BACKGROUND:

Anaphylaxis is a potentially fatal allergic reaction with systemic clinical manifestations and usually associated to a transitory increase in tryptase levels. Syncope is an uncommon manifestation of anaphylaxis. The finding of persistent high levels of tryptase is very suggestive of mast cell activation disorders (MCAD).

PATIENTS:

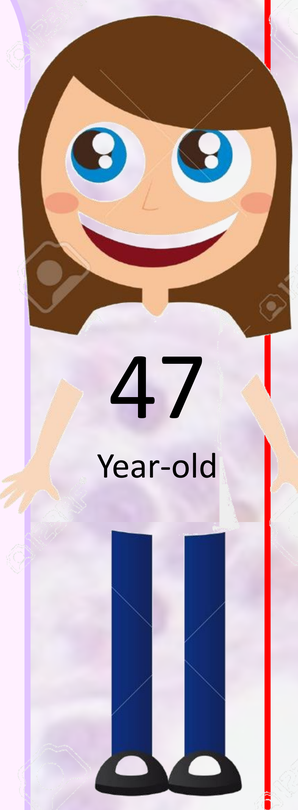
42 Year-old

Sudden episode of sweating, dyspnoea, wheezing and stridor, cyanosis, blurred vision and loss of consciousness, hypotension, tachycardia and urinary incontinence <u>while working outdoors</u>.

He reported REPEATED SIMILAR EPISODES for the last **5 years**, with complete recovery. For the last few months he suffered persistent palm and sole pruritus, without skin lesions.

Sudden episode of asphyxiation, palpitations, pruritus, facial and cervical erythema, vomiting, faecal incontinence, dizziness and hypotension, <u>immediately after dinner.</u>

She REPORTED SIMILAR EPISODES for the **last year**.



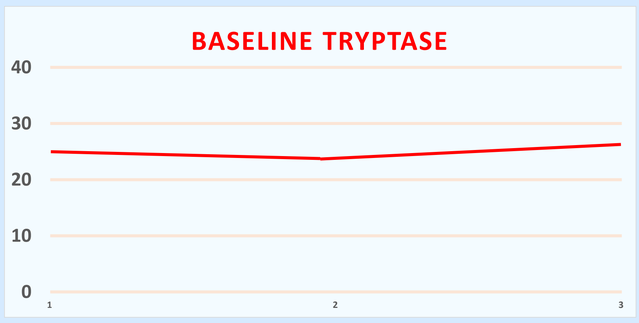
47 Year-old

NEUROLOGIC, CARDIOLOGIC AND RESPIRATORY CONDITIONS WERE RULED OUT.

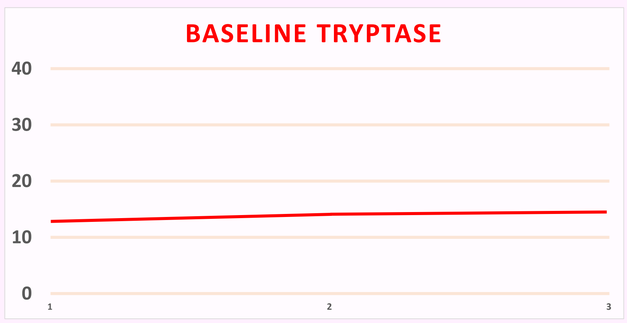
BOTH PATIENTS were diagnosed with recurrent anaphylaxis: submitted firstly for allergy evaluation (skin tests with airborne and food allergens, total and specific IgE and baseline tryptase levels) and secondly for bone marrow analysis (cytomorphology, cytometry and cytogenetics).

RESULTS:

Pollen sensitization with normal total IgE.



Negative allergy evaluation.



<u>BONE MARROW:</u>
High percentage of basophils (13%) with atypical morphology in 11% (spindle-shaped).

<u>BONE MARROW:</u> Hypercellular
Normal percentage of basophils (1%), with 100% morphologically atypical mast cells (spindle-shaped).

BOTH PATIENTS showed CD25+/CD2+ mast-cells with negative c-kit mutation in exon 8 and 17 and were diagnosed with mast cell activation disorder.

They remain asymptomatic following instructions to prevent mast-cell degranulation and are provided with epinephrine self-injectors.

CONCLUSIONS:

We present two cases of MCAD in patients with recurrent episodes of syncope, without skin lesions. The differential diagnosis of syncope, should include MCAD. A persistent tryptase level > 20 µg/L in the first case, and repeated episodes in the second case, sustained the indication for bone marrow biopsy.

The authors declare no conflict of interests.