Acta Scientific Gastrointestinal Disorders

Volume 2 Issue 3 May 2019

Editorial

Paroxystic Abdominal Pain: Think Autoinflammatory!

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Received: March 26, 2019; **Published:** April 01, 2019

We received an 8 years old boy suffering from recurrent acute attacks of abdominal pain with irregular relapses during several months, Various radiological and laboratory investigations could not diagnose the cause of these episodes, until he was admitted in the ward where he presented a similar crisis with febrile abdominal pain, arthralgias and macular rash.

Such stereotyped clinical sequence, that spontaneously resolved within 48hours, is almost pathognomonic of Familial Mediterranean Fever, the most common auto-inflammatory syndrome.

The Algerian (Mediterranean) origin and parental consanguinity (associated to the dramatic response to Colchicine*) were major clues to the final diagnosis before confirmation by genetic tests.

Autoinflammatory syndromes are now considered – according to the last Primary Immunodeficiencies Classification- as a broad spectrum that depicts, with the autoimmune diseases, a continuum of immune deficiencies [1]. These peculiar disorders might be revealed by a heterogenous group of symptoms, including paroxystic abdominal pain that can even mimick an acute surgical condition [2,3].

Similar findings of hyperthermia, skin and joints involvements; along with concomitant severe sterile peritonitis, vomiting or mesenteric lymph nodes are also described for other monogenic autoinflammatory disorders: a myriad of rare diseases driven by cytokine-mediated extraordinary sterile inflammation resulting from the over-activation of innate immunity by interleukin 1 β (IL-1β), in contrast to autoimmune diseases where disorder involves adaptive immunity.

Gastrointestinal signs are at the forefront of the common cardinal signs of AIS, along with possible mouth sores and aphthosis, and are mainly due to an aseptic and transient peritoneal inflammation.

The other clinical hallmarks are the recurrence of crisis that involve the skin, the musculoskeletal system and the central nervous system [4].

As a cherry on the cake, microbiota may modulate the inflammasome and diet-associated changes in the gastrointestinal microbiota is an important factor regulating inflammasome-mediated maturation of IL-1β; and may invert, at least partially, the hyper-inflammatory status induced by the IL-1 activation in AIS [5]. At the opposite, the gut microbiome is regulated (or dys-regulated) by inflammasomes and their activation [6].

In sum, gastrointestinal symptoms may unmask rare genetic conditions, like an autoinflammatory syndrome.

A holistic approach of such recurrent episodes of severe abdominal pain and peritonitis, encompassing a well-described clinical evaluation of gastrointestinal and extra-digestive associated symptoms, as well as genetic studies, are mandatory to avoid a long odyssey before diagnosis.

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