Editorial

Autoinflammatory syndromes: rare diseases with important implications in quality of life

Síndromes autoinflamatórias: doenças raras com comprometimento importante da qualidade de vida

The systemic autoinflammatory syndromes often present with recurrent fevers and high inflammatory markers. In the last decades, autoinflammatory syndromes have been increasingly recognized and the identification of genetic mutations has helped not only the diagnosis but also to understand the role of inflammasome and of the innate immune system. Diagnosis relies on clinical suspicion followed by genetic testing.

In this edition, the members of the Brazilian Pediatric Rheumatology Committee have elaborated three publications reviewing consensus on diagnosis and treatment of three important systemic autoinflammatory syndromes: Familial Mediterranean fever (FMF), Periodic Fever, Aphthous Stomatitis, Pharyngitis, and Adenopathy Syndrome (PFAPA) and cryopyrinopathies.1–3 Although extremely rare, they are important to be recognized, as many can now be completely controlled by long-term drug therapies.

The consensus emphasizes the importance of clinical suspicions in patients with recurrent or continuous signs of systemic inflammation in the absence of infections and malignancies.1–3 Duration of fever and concomitant clinical manifestations are important features for diagnosis. Laboratory investigation is usually unspecific, revealing signs of systemic inflammation. Although genetic testing is costly and not widely available, it is often the key to establish definitive diagnosis in FMF and cryopyrinopathies.5,3 However, up to date more than 150 mutations have been described in each of the disease and new mutations are constantly being published.

Treatment has shown to be effective and reduce long-term complications such as amyloidosis. First line treatment is colchicine in FMF and prednisone in PFAPA.1,3 IL1 inhibitors have been usefully used in cryopyrinopathies, and have been described in case series and case reports in other systemic autoinflammatory diseases.1,2

REFERENCES


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