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NLRP12- associated autoinflammatory disorder: case report

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Introduction

During latest years, the identification of genes involved in the control of inflammation, apoptosis processes and a better comprehension of the mechanisms connected to an anomalous activation of the inflammasome, have made possible to delineate a new group of illness called "Monogenic autoinflammatory syndromes".

Objectives

Report a case of recurring fever due to NLRP12 mutation.

Methods

E.M., 4 years old, comes to our observation because of the presence of recurrent episodes of fever lasting 4-5 days,since about one year, every 15 days, associated with diarrhea, urticaria-like rush decreasing spontaneously, mainly on the back and lower limbs.

From November 2012 to April, the patient presented daily fever (Tmax 40°C) associated with worsening rush, arthralgia and arthritis of knees and ankles , edema of eyelids and lips and palmar angioedema during 7 days. From April 2013, every 15 days, the child had again febrile episodes (Tmax 40°C) lasting about seven days with the associated symptoms already described. Each episode has been exclusively cured with Paracetamol. Blood tests revealed just a small increase of the inflammatory indexes. After excluding other common causes of persistent and recurrent fever in the pediatric age, we suspected an auto-inflammatory disease. Then , we directed the patient to a third level structure, where analysis of the CIAS1 gene have been carried out. Suspecting strongly an auto-inflammatory disease, the NLRP12 gene has been examined too. Analysis of the CIAS1 gene didn't reveal pathological mutations, while the analysis of NLRP12 gene resulted positive.

Results

Considering the clinical feature associated with increase of inflammatory indexes and the genetic analysis outcomes, the diagnosis of disease was NLRP-12 associated autoinflammatory disorder (NLRP12AD).

Conclusion

The NLRP12AD is an autosomic dominant disease , due to mutation of NLRP12 gene which encode for the NLRP12 protein or "MONARCH-1", playing a crucial role in the immune mechanisms against pathogens agents. As for Cryopyrinopathies, symptoms can be inducted by the exposure to the cold and are characterized by recurring fever episodes lasting 5-10 days, associated with rash, headache, lymphadenopathies, oral ulcers and abdominal pain. Therapy depends on the seriousness of the symptoms: in less serious cases , treatement is based on antihistamines, NSAIDs and corticosteroids; in more serious ones, the administration of Anakinra can be useful. In the refractory cases, further therapeutic strategies are based on Anti-TNF and ANTI-IL-6 agents.

Disclosure of interest

None declared.

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