RESEARCH PROJECT:

The p2x7-inflammasome connection: a novel pathway in autoinflammatory diseases

THE AIM OF THE RESEARCH

Autoinflammatory diseases are a group of genetic diseases characterized by recurrent fever with inflammation localized to skin, pleura, peritoneum, joints, and also eyes, liver, spleen or the nervous system. Sometimes, liver or kidney involvement may lead to death. The most relevant pathogenetic factor in autoinflammatory diseases is no doubt the excessive production of inflammatory mediators known as cytokines, notably Interleukin-1B (IL-1B). In general, with the exception of a disease known as Familial Mediterranean Fever, which is relatively common in populations of middle-eastern origin and in Southern Italy, they are rare. These diseases are of increasing interest by physicians and biomedical investigators as there is no current therapy. Our Research Unit has identified in recent years one of the most potent receptors (named P2X7) responsible for IL-1B maturation and release. Furthermore, we have unveiled the mechanism by which this receptor stimulates the intracellular pathways that lead to processing and release of this cytokine, and how it is released by leukocytes, and have developed potent pharmacological blockers. In this project we will 1) investigate the role of the P2X7 receptor; 2) verify if its deletion (in in vitro cell system or in mice genetically-modified) lowers IL-1B release; 3) check the use of pharmacological blockers; 4) study the presence of mutations or polymorphisms.

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Orphanet Database.
http://www.orpha.net/consor/cgi-bin/ResearchTrials.php?lng=EN