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Towards molecular medicine approaches in hereditary autoinflammation and the inflammasome

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Abstract

Hereditary autoinflammatory diseases are heterogenous group of disorders characterized by seemingly unprovoked fever and localized recurrent episodes of excess systemic inflammation without any pathogens stimuli. Unlike autoimmune disorders, autoinflammatory disorders lack the production of high-titer autoantibodies or antigen-specific T cells. The most important determinants of autoinflammatory diseases which are named also as 'hereditary periodic fevers' are abdominal pain and fever characterized with acute and usually short attacks of serosal, synovial and cutaneous inflammation.

These disorders are caused by primary dysfunction of the innate immune system, without evidence of adaptive immune dysregulation. The protein products of the genes which are associated with these disorders are known to control innate immunity and apoptosis. Role of pyrin (PyD) and/or caspase recruitment domain (CARD) containing regulator and adaptor proteins in inflammation, apoptosis, and innate immunity has been previously outlined. The inflammasome is composed of intracellular receptor NALP and ASC adapter which stimulates caspase-1 activation. Mutations in the NALP3 inflammasome and NLR cause autoinflammatory syndromes by increased inflammasome activity responsible for uncontrolled IL-1β production.

In this communication, we have documented molecular analysis data of the studied patient populations with autoinflammatory disorders, and our research activities conducted on autoinflammatory disease genes. Moreover, current work also underlined the critical significance of molecular diagnosis which refers to detailed mutation screening of autoinflammatory disease genes in particular for mutation negative or asymptomatic individuals among at-risk populations. Since there is still way in understanding and exploring the undefined periodic syndromes, further studies are required to discover novel inflammatory pathways.

Biography

Sinem Nalbantoglu has completed her Ph.D at the age of 27 years from Ege University and postdoctoral studies from Ege University School of Medicine. She is now working as assistant professor in Nisantasi University. She has been publishing papers in reputed medical journals and serving as referee in many peer-reviewed journals.