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## Genetics of inflammasome: From monogenic rare syndromes to complex diseases

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Inflammasomes are cytoplasmic complexes capable of triggering the inflammatory cascade through the activation of caspase-1, and the consequent production of inflammatory cytokines IL-1 $\beta$  and IL-18. Different inflammasome receptors (NLRP-1, NLRP-3, NLRC-4, AIM-2, IFI-16 and pyrin) are activated by distinct pathogens and/or damage molecular patterns, contributing to innate immune response in a stimulus- and cell/tissue-dependent way. Mouse genetic models have revealed the mechanistic details of inflammasome activation and its role in physiologic immune response, however little is known about the action of inflammasome in humans. Genetic studies within the human population uncover mutations and polymorphic variants in inflammasome components resulting in gain-of-function of caspase-1, and the consequent development of rare monogenic inflammatory diseases (auto-inflammatory syndromes) as well as of complex disorders which common in general population. Our group demonstrated the association of inflammasome genes in different multifactorial diseases. Candidate gene analysis have shown a significant association of inflammasome genes with a wide range of disorders, including infections (HIV-1, HPV and mycobacteria), autoimmune diseases (type-1 diabetes, SLE, celiac disease and multiple sclerosis), neurodegenerative disorders (Alzheimer and Parkinson disease), cancer (melanoma and cervical cancer) and metabolic syndromes (renal failure and obesity), revealing the contribution of one or another inflammasome in the pathogenesis of specific diseases. Deeper investigations are still going on to further depict the involvement of inflammasome dysregulation in the pathogenesis of such diseases.

### Biography

Alessandra Pontillo has a PhD in Experimental and Clinical Pathology (University of Trieste; 2003), and Master's degree in Genetic Medicine (University of Trieste; 2008). Since 2013, she is an Assistant Professor at the University of Sao Paulo/USP (Sao Paulo, Brazil). She is the PI of the Laboratory of Immunogenetics in the Department of Immunology. She has expertise in innate immunity and inflammation, inflammasome, auto-inflammatory diseases, immunogenetics and population genetics. Her research projects focuses on human genetics of innate immunity and inflammation with particular interest in inflammasome complex.

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