Immunology Summit-2013

OMICS Group Conferences
Accelerating Scientific Discovery

2nd International Conference on
Clinical & Cellular Immunology

October 15-17, 2013 Hampton Inn Tropicana, Las Vegas, USA

Activating killer-cell immunoglobulin-like receptor (KIR) genes in autism

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Abstract

Autism is a term used for a complex group of neurodevelopmental disorders characterized by deficits in communication, social skills and the presences of repetitive stereotyped behaviors. Although there has been extensive research into the etiology of autism, little is known. Genome wide association studies have only identified about 5-10% of the genetic risk after studying thousands of autism subjects.

Natural killer-cells (NK-cells) are a subset of lymphocytes with the inborn ability to produce cytokines and kill target cells without prior sensitization. They recognize the lack of HLA proteins on the surface of virally infected and transformed cells ("missing self"). Among the surface receptors on NK-cells are killer-cell immunoglobulin-like inhibitory or activating proteinreceptors (KIR) that are encoded by genes in the leukocyte receptor complex (Chromosome 19q13/4). The ligands that bind to these receptors are cell-surface HLA class I proteins encoded by genes in the class I region (Chromosome 6).

KIR genes and their cognate HLA ligands have been associated with several autoimmune diseases and there is evidence that NK-cells activity is associated with autism. In this presentation, I will present data that suggests a strong association with certain activating KIR genes and their cognate HLA genes in the Caucasian autism population (Torres, Brain, Behavior and Immunity 26 (2012) 1122-1127.

Biography

Torres received his MD degree at the University of Utah and completed his postdoctoral training as a Research Associate at the National Institutes of Health and as a resident at Yale University. He has worked in the biotechnology industry and is currently the Director of the Biomedical Laboratory at the Center for Persons with Disabilities, Utah State University. He has published in the field of protein chemistry, molecular biology, and the genetics of autism.