Definition and characterization of type I interferonopathies

From 2013-03-01 to 2018-10-31, ongoing project

Objective

Although the concept of grouping Mendelian disorders associated with an up-regulation of type I interferon (IFN) has not been previously recognised in the medical literature, my past and current work argues that this concept has scientific validity and clinical utility. I believe that such conditions, which include Aicardi-Goutières syndrome (AGS), spondyloenchondrodysplasia, and some cases of systemic lupus erythematosus (SLE), can usefully be considered to represent a novel set of inborn errors of immunity, and that the recognition of diseases as type I interferonopathies will have significance for the development of targeted therapies, as well as informing our understanding of viral and retroelement biology, and the pathogenesis of some forms of autoimmunity.

This proposal will combine my expert phenotyping skills with revolutionary sequencing technologies, cutting edge systems for the interrogation of retroelements, and contemporary immunological assays. Deliberately focusing on human studies, I will identify new disease-related genes for AGS and lupus, and define currently unrecognised phenotypes arising from a dysregulation of type I IFN metabolism. I hypothesise that, to a currently undefined extent, SLE represents a heterogeneous collection of individually rare Mendelian subtypes, and suggest that the definition of new genetic subtypes of lupus will deliver a step change in our knowledge of disease pathogenesis through the delineation of pathways relevant to larger groups of patients. Since gene identification frequently represents a first step in understanding relevant cell biology, the work described herein has the potential to transform the clinical management of type I interferonopathies through the development of directed therapeutic approaches. Furthermore, I believe that our proposed experiments to study retroelement biology in the AGS-context are likely to reveal truly exciting insights into the activity and control of ‘junk’ DNA.

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