Autoimmune polyendocrine syndrome type I - a rare disorder of childhood as a model for autoimmunity

Ziel

Autoimmune polyendocrine syndrome type I (APS I), a rare genetic disorder of childhood, has proven to be an invaluable tool in understanding autoimmune reactions. APS I (OMIM 240300), also known as APECED (autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy), is a severe autosomal recessive disorder caused by mutations in the Aire gene on chromosome 21. The disorder begins in early childhood and the patients gradually develop symptoms from autoimmune reactions in different endocrine and non-endocrine tissues and, in addition, mucocutaneous candidiasis, one of the hallmarks of the disease phenotype. APS I is characterized by autoantibodies against several defined autoantigens often identical to those found in more common autoimmune disorders such as type 1 diabetes mellitus and Addison's disease. The defective gene, Aire, has been identified and multiple mutations have been characterized. Aire-deficient mice, with the same genetic defect as the human disease, have been produced.

The aim is to capitalize on the collected strengths and expertise of European investigators and establish a pan-European patient database and biobank. A further aim is to use a genome-wide approach to define the signalling pathways affected by the defective Aire molecule. Another aim is to maximize the human-mouse cross talk in our efforts to identify tissue specific autoantigens and immunological peptides of importance in patients and mice, and to determine their role in the disease pathogenesis. Our genomics-based approach will facilitate the identification of genes modulating the intensity and/or the course of autoimmune reactions as well as the cause of Candida albicans infection in APS I patients. This information will not only help patients with this rare disorder but will also increase our understanding of the pathogenesis of autoimmune diseases in general and could potentially lead to novel therapeutic strategies for treating common autoimmune disorders.

Verwandte Informationen

Ergebnis in Kürze

Deciphering autoimmunity mechanisms

Berichtszusammenfassungen

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