



POSTER PRESENTATION

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B cells characterization in ADA2 Deficiency patients

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Introduction

ADA2 deficiency, a recently described disease, is characterized by systemic vasculopathy and episodes of strokes. The defect is due to a loss of function mutation of *CECR1* gene, codifying for Adenosine Deaminase 2 protein. This protein regulates the catabolism of extracellular adenosine, which we have recently shown is an important regulator of Class Switch Recombination in B lymphocytes. Accordingly DADA2 patients can present hypogammaglobulinemia.

Objectives

Therefore we decided to characterize peripheral B and T lymphocytes of DADA patients to directly address if ADA2 mutation affects B-cell function and in particular we focused on B cell- T cell interaction.

Patients and methods

3 patients carrying mutations in *CECR1* were examined. They showed clinical history with livedo reticularis, fever, vasculitis and neurological symptoms. Two patients presented hypogammaglobulinemia requiring intravenous immunoglobulin replacement therapy. We analyzed peripheral B and T cell phenotype by flow cytometry, *in vitro* B-cell proliferation and differentiation to Immunoglobulin secreting cells in response to CpG and T cell help.

Results

Flow cytometer analysis showed a reduction of total B cells compared with age matched controls. Intriguingly a decrease in the percentage of memory B cell compartment (CD19+CD27+) was observed. Moreover we noted that the rate of B cells proliferation and differentiation to Immunoglobulin Secreting Cells of DADA2 patients with autologous T cell help are impaired. In fact *in vitro* IgM, IgG and IgA secretion is significantly reduced with

respect to HD B lymphocytes in presence of mutated CD4 helper T cells.

Conclusions

Our findings suggest that ADA2 defect could lead to a defect in B cell function and to a reduced T cell dependent B cell response.

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