AUTOIMMUNE CONNECTIVE TISSUE DISEASES

ADA2 MUTATION: PARADIGM SHIFT IN VASCULAR DAMAGE? – A REPORT OF A PRELIMINARY STUDY ON 4 CASES OF LIVEDOID VASCULOPATHY

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Background: Adenosine deaminases (ADA) play an important role in the human body, regulating the level of adenosine, a signaling molecule that controls various cellular responses by binding to and activating adenosine receptors, which are expressed by most cells. It has been proposed that mutation of ADA2 result in weak endothelium of small and medium sized blood vessels due to the loss of the catalytic and immune growth factor activity of ADA2.

Objective: We aim to investigate if there is evidence that correlates between ADA2 mutation and condition such as livedoid vasculopathy, lymphocytic thrombophilic arteritis, cutaneous polyarteritis nodosa, Degos' disease and Buerger's disease. In this preliminary study we report a study on four cases of livedoid vasculopathy.

Materials and Methods: Patients who fulfil the diagnostic criteria for livedoid vasculopathy were recruited for the study. Genomic DNA was isolated from blood samples. Each of the 17 ADA2 exons and its exon-intron boundaries was PCR amplified and then genetic analysis performed.

Results: DNA samples from all four patients and a Chinese healthy control individual were screened for mutation in the coding region of ADA2 gene. Except some silent mutations and polymorphism, there was no mutation detected in all DNA samples.

Conclusions: This preliminary study on 4 patients showed that there is no correlation between livedoid vasculopathy and ADA2 mutation.