A new perspectives in a detection of markers of human diseases in Slovakia

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I. INTRODUCTION

Nowadays is very popular individualized therapy. This advance is characterized by application of genetic (genome) and molecular and biochemical (RNA, proteins, metabolites) data for exploration disease etiology and pathophysiology. A big challenge is to define persons with a predisposition to any disease and phenotype of a disease as well. Analysis of DNA and single nucleotide polymorphisms is a modern strategy for these aims. Common variable immunodeficiency (CVID) is the most common symptomatic primary immunodeficiency. Symptoms of common variable immunodeficiency are heterogeneous and unspecific therefore its diagnosis and management is challenging. This immune deficiency is considered to be a collection of genetic immune defects with complex inheritance patterns but exact gene modifications are still unknown. Our aim is to find an early marker, to optimize therapy and to increase efficacy of treatment.

II. METHODS

We decided to use DNA chip array (Genome-Wide Human SNP Array 6.0 – Affymetrix) and we analyzed DNA from patients with common variable immunodeficiency and DNA of healthy persons. We analyzed 906 600 single nucleotide polymorphisms and 946 000 probes for the detection of copy number variation.

III. RESULTS

We found several significant changes and it is possible that some single nucleotide polymorphisms can be responsible for protection against common variable immunodeficiency or can participate in origin and development of common variable immunodeficiency. We identified 28 221 single nucleotide polymorphisms with p less than 0.05 and 27 single nucleotide polymorphisms with p less than 10^{-5} in our small population of patients with common variable immunodeficiency.

IV. CONCLUSIONS

Common variable immunodeficiency represents a clinical and immunological syndrome that merges various diseases with different genetic roots. It is still problem to diagnose common variable immunodeficiency during childhood and very often patients are without diagnose and an adequate treatment. Using DNA chip array or whole exome sequencing is needed to illuminate the causes of this disease. We found several potential positive and negative markers of common variable immunodeficiency. Additional deeper analysis of determined single nucleotide polymorphisms is necessary.

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