Transcriptomic profiling of asthma, atopic eczema and allergies and their combinations in Czech adult population

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Asthma, atopic eczema and allergies are widespread immune-mediated diseases that often occur together[1]. The multimorbidity is related with immunodeficiency and shares a common inflammatory pathophysiology[2,3,4]. The most common multimorbidity is known as atopic triad. The atopic triad frequently starts by atopic dermatitis at first years of life followed by allergies and with last stage of asthma occurrence. The manifestation of atopy is based on IgE-mediated response. On the other hand, contact dermatitis is other type of allergy. It is type of eczema caused by contact with a certain substance that dry and irritate the skin. The reaction is based on cellular response.

The molecular signatures within the diseases are heterogeneous and also vary across their co-occurrence. Importantly, the incidence of the atopic diseases still increases in population and investigation of underlying mechanism can positively impact the human health by improving mitigation strategies[4, 5].

The common profiles of blood gene expression in the immune-mediated diseases are still not well understood and only few studies performed the analysis using transcriptomics to fully understand the deregulation machinery.

Aims: **To investigate the transcriptomics changes in immune-mediated diseases:**
(1) to analyze transcriptomic profiles of i) IgE mediated diseases, i.e. allergies, eczema and asthma, and ii) contact dermatitis
(2) to identify differences in dermatitis and IgE-mediated allergies;
(3) to identify deregulated pathways in biological processes caused by altered gene expression
(4) to characterize potential novel gene biomarkers related to the diseases (future step)

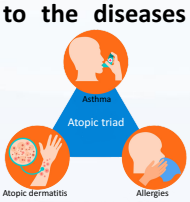

Fig.1 Atopic triad: atopic eczema, allergies and asthma

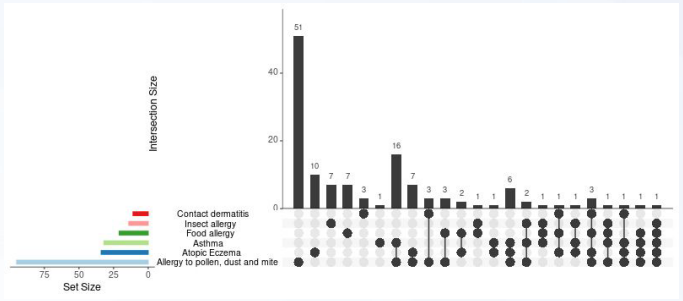

Fig.2 Description of population: distribution of studied diseases (their single occurence and combinations)

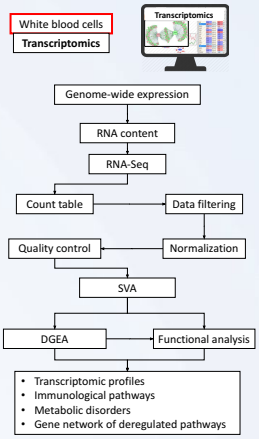

Fig.3 The workflow from the samples to data analyses for applied transcriptomics.

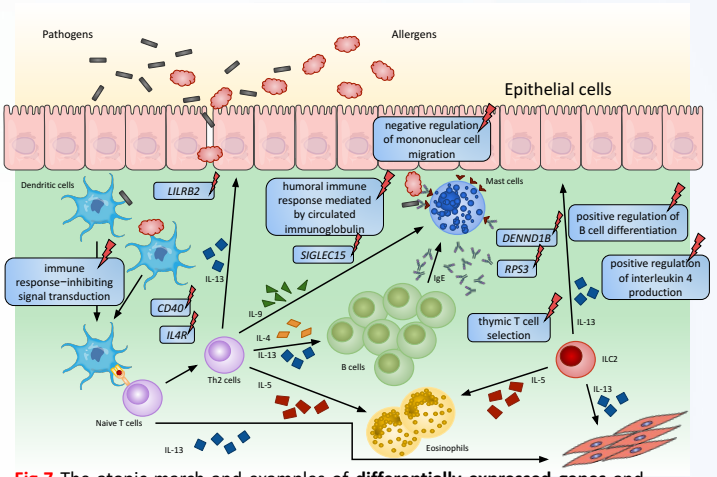

Fig.7 The atopic march and examples of **differentially expressed genes** and **altered biological processes** related with immune system in atopy.

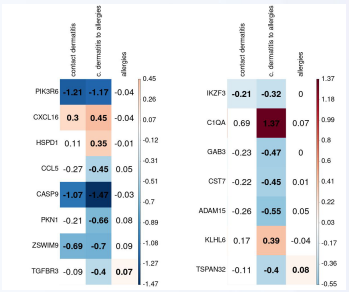

Fig.4 Deregulated immune-related genes in contact dermatitis compared to control group and atopic allergies. Individual cells show fold changes, significantly up-regulated genes are colored by red, significantly down-regulated genes are colored by blue (FDR <0.2).

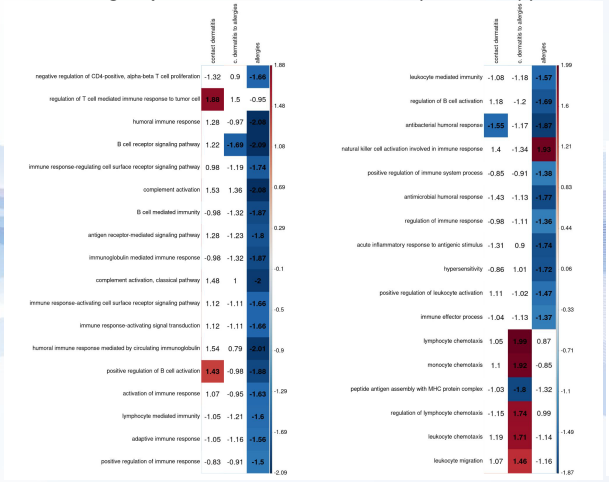

Fig.5 Example of deregulated immune pathways in allergies and contact dermatitis compared to control group and contact dermatitis compared to atopic allergies . Individual cells show normalized enrichment score, significantly deregulated pathways are colored (p value <0.005).

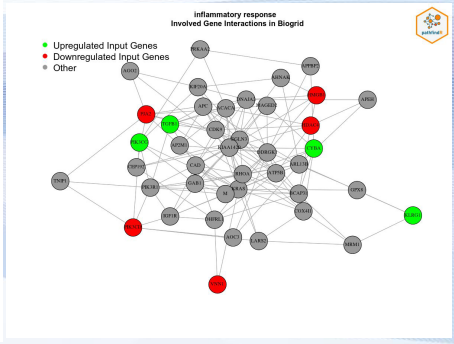

Fig.6 Example of altered genes involved in deregulated inflammatory response pathway in participants with any atopic disease analyzed by pathFindR

- About 150 genes were differentially expressed (FDR <0.2) in atopic triad, 20 of them are directly involved in immunological pathways
- Other 400 immune-related genes were found altered (p value <0.05) across all tested groups
- 15 immune-related genes showed differential expression (FDR <0.2) between contact dermatitis and IgE-mediated allergies
- Significant changes in gene networks and metabolic pathways associated with immune function were found in all disease groups in comparison with healthy individuals.

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