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## The NOD-like receptor (NLRP3) gene variability in patients with recurrent aphthous stomatitis

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**Statement of the Problem:** Recurrent aphthous stomatitis (RAS) is a multifactorial disease with an unclear etiopathogenesis, resulting from the interplay between genetic and environmental factors. As the dysregulation of the immune system can play a role in the RAS development, single nucleotide polymorphisms (SNPs) in the genes for immune and inflammatory molecules were studied. The NOD-like receptor (*NLRP3*) gene, encoding the component of the inflammasome, has been proposed as one of the candidate genes for RAS. The aim of our study was to investigate three SNPs (rs4612666, rs10754558, rs3806265) in *NLRP3* gene in patients with RAS and healthy controls in the Caucasian population.

**Methodology:** A total of 200 Czech subjects were enrolled in this case-control study. 143 healthy controls, 57 patients with RAS were genotyped by method based on polymerase chain reaction using 5' nuclease TaqMan<sup>\*</sup> assays. Clinical parameters such as complete blood count, levels of immunoglobulins including allergen-specific immunoglobulin E or presence of antibodies against cytomegalovirus, Epstein-Barr virus were determined in RAS patients.

**Findings:** Although no significant differences in the *NLRP3* (rs10754558, rs3806265) allele and genotype frequencies between patients with RAS and controls were observed, statistically significant differences in *NLRP3* rs4612666 genotype frequencies between subjects with RAS and controls were found. Carriers of *NLRP3* rs4612666 TT genotype had a higher risk of developing RAS in comparison to subjects with CT + CC genotypes (OR=16.71, 95% CI=1.96-142.14, P=0.0024). No association between *NLRP3* haplotypes and RAS was detected.

**Conclusion & Significance:** In contrast to the previous study, associations between *NLRP3* (rs10754558, rs3806265) polymorphisms and RAS were not confirmed. However, we suggest that *NLRP3* rs4612666 polymorphism can strongly influence the risk of developing RAS in the Czech population.

## Biography

Simona Valova studied Molecular Biology and Genetics at Faculty of Science, Masaryk University, Brno, Czech Republic. She is currently in her third year of PhD in Physiology and Pathological Physiology at Faculty of Medicine, Masaryk University. She works in the team of Professor Lydie Izakovicova Holla that focuses on variability in candidate genes for multifactorial diseases, including periodontitis, recurrent aphthous stomatitis, diabetes mellitus or gastroesophageal reflux disease.

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