

TITLE: *AIM2* (rs1103577) gene polymorphism in pulmonary tuberculosis patients

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ABSTRACT:

Tuberculosis (TB) is an infectious disease considered a serious public health problem, resulting in high mortality caused by *Mycobacterium tuberculosis*. In 2018, Brazil notified around 72,788 new cases of TB and Amazon state had the highest incidence of TB cases in the country. These high rates have been associated with social, environmental and genetic factors. Innate immune response is the first line of defense against mycobacteria infection, activating intracellular protein complexes known as inflammasomes. Inflammasomes are essential for controlling bacterial growth through activation of caspase 1 which cleaves and activates pro-inflammatory cytokines. Thus, the studies of polymorphisms of genes involved in response by inflammasomes is essential and may contribute to elucidate the host immune responses to TB infection. For this reason, the present study evaluated single-nucleotide polymorphisms (SNPs) of *AIM2* (rs1103577) in patients with TB. In this study, were included 416 TB patients and 532 controls recruited from Reference Center for Sanitary Pneumology “Policlínica Cardoso Fontes”, Manaus-AM, Brazil. The genotyping of the samples was performed by the qPCR (Quantitative Real Time PCR) through statistical analysis program R. In the present study, the frequency of the minor allele (MAF) of *AIM2* in the control group (T allele) was 0.47% and in the TB patients (T allele) was 0.44%. In relation to association analysis of SNPs for the genes, one significant genotypic and allelic differences were observed between the groups studied with *AIM2* gene. *AIM2* rs1103577 T/T genotype was significantly associated with protection in male patients with pulmonary TB ($p=0,027$ e $OR=0,69$; $p_{adj}=0,051$ e $OR_{adj}=0,68$). The results of this study increasingly reinforce the importance of inflammasome in TB, contributing to better understanding of the mechanisms involved in the immune response of the host of TB.

Keywords: Tuberculosis, inflammasome, *AIM2* and polymorphisms

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