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Título	A negative screening of rare genetic variants in the ADIPOQ and STATH genes in cystic fibrosis
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Resumo	Background: The phenotypic variability in cystic fibrosis (CF) is widely recognized and modulated by environmental and genetic factors, including CFTR pathogenic variants and modifier genes genetic variants. In this context, determining the presence of variants in genes involved in immune response may allow a better understanding of CF variability, mainly in lung disease. Thus, ADIPOQ and STATH genes were selected and the analysis of exons and exon/intron junctions was performed for the determination of variations in its sequence, to determine the possible genetic modulation. Methods: A total of 49 patients with CF, diagnosed for showing abnormal [chloride] levels in the sweat test, and identification of two pathogenic variants in CFTR categorized as class I and II were included. Genetic sequencing was performed for the identification of variants in the modifier genes. Results: In our analysis, there was absence of rare genetic variants in STATH and ADIPOQ genes associated with the clinical variability. Thus, we are not able to establish an association between the disease severity and rare genetic variants in STATH and ADIPOQ genes, considering exons and exon/intron junctions. Conclusions: Considering the negative screening for rare genetic variants in ADIPOQ and STATH genes, it may be concluded that these genes are not associated with phenotypic modulation of CF in our population. To understand the modifier genes and its action at CF variability it is essential to promote a

	better overview of the disease. Also, negative reports can help to direct new studies without the use of unnecessary financial support.
Fomento	