

Genetic Factors of the Disease Course After Sepsis: Rare Deleterious Variants Are Predictive.

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Abstract

Sepsis is a life-threatening organ dysfunction caused by dysregulated host response to infection. For its clinical course, host genetic factors are important and rare genomic variants are suspected to contribute. We sequenced the exomes of 59 Greek and 15 German patients with bacterial sepsis divided into two groups with extremely different disease courses. Variant analysis was focusing on rare deleterious single nucleotide variants (SNVs). We identified significant differences in the number of rare deleterious SNVs per patient between the ethnic groups. Classification experiments based on the data of the Greek patients allowed discrimination between the disease courses with estimated sensitivity and specificity >75%. By application of the trained model to the German patients we observed comparable discriminatory properties despite lower population-specific rare SNV load. Furthermore, rare SNVs in genes of cell signaling and innate immunity related pathways were identified as classifiers discriminating between the sepsis courses. Sepsis

patients with favorable disease course after sepsis, even in the case of unfavorable preconditions, seem to be affected more often by rare deleterious SNVs in cell signaling and innate immunity related pathways, suggesting a protective role of impairments in these processes against a poor disease course.

Involved units

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