



POSTER PRESENTATION

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# NOD2/CARD 15 gene mutations in patients with Familial Mediterranean Fever

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## Background

Familial Mediterranean fever (FMF) and Crohn's disease (CD) are autoinflammatory disorders, associated with genes (MEFV and NOD2/CARD15), encoding for regulatory proteins, important in innate immunity, apoptosis, cytokine processing and inflammation. While mutations in the MEFV gene were shown to modify CD, the role of NOD2/CARD 15 gene mutations in the FMF disease phenotype was never studied before.

## Methods

The cohort consisted of 103 consecutive children with FMF, followed in a single referral center. NOD2/CARD15 genotypes were analyzed in all patients and 299 ethnically matched unaffected controls. Demographic data, clinical characteristics and disease course of FMF patients with and without NOD2/CARD 15 mutation were compared.

## Results

A single NOD2/CARD 15 mutation was detected in 10 (9.7%) FMF patients and 26 (8.7%) of controls. No homozygotes or compound heterozygotes were discovered in the 2 groups. FMF patients carrying a NOD2/CARD 15 mutation had higher rate of erysipelas-like erythema and acute scrotum attacks and a trend for higher rate of colchicine resistance and a more severe disease as compared to patients without mutations.

## Conclusion

NOD2/CARD 15 mutations are not associated with a susceptibility to develop FMF, yet the presence of these

mutations in FMF patients appears to be associated with a trend to a more severe disease.

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