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Significance of I313V mutation of NLPR3 gene in two pediatric patients

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Background

The I313V mutation of the NLRP3 gene has been only anecdotally reported and described in association with the so-called Magic Syndrome (Infevers database). However, nor the clinical or pathophysiological significance of such mutation has been so far reported.

Aim

To describe the clinical picture of patients carrying the I313V mutation and its consequences in IL-1β secretion.

Methods

Two families carrying NLRP3 I313V mutation were evaluated. Monocytes were obtained from patients and their parents. Cells isolated from healthy donors (HD) (N=14) were used as negative control group. Pattern of secretion of IL-1β, IL-1Ra, IL-6 and IL-8 were then assessed by ELISA in the presence or absence exogenous LPS.

Results

Both case #1 (M.T) and #2 (V.C) displayed a mild clinical phenotype (episodes of urticarial rash and arthralgia associated with elevation of acute phase reactants), compatible with FCAS and Muckle-Wells syndrome, respectively. Both patients displayed good response to NSAID and/or steroid on demand. Compared to HD controls, patients displayed enhanced and delayed IL1β secretion. This was accompanied by higher levels of lL1Ra and IL-6 without any significant differences in IL-8. Interestingly, parents carrying the mutation also displayed higher levels of secreted IL-1β compared to HD control group.

Conclusion

The I313V mutation is associated with a mild CAPS phenotype and with an increased IL-1β secretion.

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