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FMF in heterozygotes: are we able to accurately diagnose the disease in very young children?

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Background

Reports of heterozygous carriers of a *MEFV* mutation presenting a FMF phenotype are increasing, but no data is available on the outcome of FMF heterozygous children.

Aim

To assess the relevancy of clinical diagnosis of FMF in heterozygous children before the age of 6.

Methods

We performed a retrospective single-centre study of 29 patients diagnosed with FMF before the age of 6, who had only 1 mutation in the *MEFV* gene, compared to a group of 26 homozygous or compound heterozygous patients in whom the diagnosis of FMF was also made during early childhood.

Results

Presenting signs in heterozygous children did not differ from homozygous or compound heterozygous patients. Initial response to colchicine was identical in the two groups. During follow-up heterozygous patients were more likely to have a milder course of the disease. After puberty clinical signs of FMF totally disappeared in 6/11 heterozygous patients. In these 6 patients, colchicine could be withdrawn without recurrence of symptoms or rise of inflammatory markers. If applied after puberty, clinical diagnostic criteria sets were no longer positive in these 6 patients, whereas the same criteria applied retrospectively during early childhood concluded to FMF.

Conclusion

Our study suggests that the diagnosis of FMF in very young heterozygous children should be cautious. Heterozygous children can present with an FMF-like disease during early childhood that may disappear with age, while others will suffer lifelong from their disease. Only a careful follow-up of FMF heterozygotes allows an accurate diagnosis over time.

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