PEDIATRIC RHEUMATOLOGY

MEETING ABSTRACT Open Access

P01-008 – FMF genotype-phenotype correlations in Germany

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Introduction

Familial Mediterranean fever (FMF) is one the most common autoinflammatory disease (AID). Pathogenomic relevant mutations in the MEFV gene show autosomal recessive inheritance, but co-dominant mutations have been described.

Objectives

We aimed to evaluate correlations between ethnic origin, phenotype and genotype for FMF patients in the German AID-Net-registry.

Methods

We used two common scoring systems modified for children (Mor et al., Pras et al.) to assess disease severity in 243 FMF patients of the AID-Net-registry. For the four most frequent mutations, we tested for a correlation of the genotype with the phenotype, mean CRP and ethnic origin, respectively. Furthermore, we evaluated the applicability of the two severity scores for children.

Results

Among the 243 patients, we detected a total of 433 pyrin mutations and 22 different sequence variants, including one new mutation (p.Gly488Asp). The four most frequent alterations were p.Met694Val (55%, n=238), p.Met680Ile (12%, n=52), p.Val726Ala (10%, n=44) and p.Glu148Gln (8%, n=34). Ethnic origin could be determined in 224 cases; the prevailing ancestry was Turkish (83%, n=185), 8% (n=18) were Lebanese. P.Met694Val in homozygous form (n=74; 30.5%) was correlated with a more severe disease activity, based on the score by Mor, as well as with a higher mean CRP (74 mg/l, n=60, 31 mg/l, n=59) compared to patients without this mutation (p=0.01 and p<0.01, respectively). The score suggested by Pras did not yield a significant genotype-phenotype correlation; indeed, the two scoring systems were inconsistent with each other (κ<0.07). Although a typical distribution of mutations in different ethnic populations was obvious, this trend was not statistically significant, probably due to the divergent number of cases.

Conclusion

The homozygous p.Met694Val substitution was associated with a more severe disease activity. There was no origin-genotype correlation in this FMF population. The well-known severity scores for children (Mor, Pras) are inconsistent.

Disclosure of interest

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