PEDIATRIC RHEUMATOLOGY

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# A validation of diagnostic score for molecular analysis of hereditary autoinflammatory syndromes with periodic fever in Turkish children

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## Objective

In the present study, we validated a set of variables that predict the presence of a mutation in one of the 3 genes known to be associated with the common monogenic autoinflammatory diseases.

## Methods

A total of 93 consecutive patients with a clinical history of periodic fever were screened for mutations in the *MVK*, *TNFRSF1A*, and *MEFV* genes, and detailed clinical information was collected prospectively. For autosomal-recessive diseases (MKD and FMF), only homozygous or compound heterozygous patients were accepted as genetically positive. The developed diagnostic score was validated in an independent set of 93 patients.

## Results

93 patients whose suspected diagnosis was one of the autoinflammatory diseases were enrolled. The mean age at onset was 58,64+40,69 months (54 F, 39 M;). A total of 25 patients with periodic or recurrent fevers did not display any mutation in the 3 genes examined. Of these 68 patients with positive findings on genetic testing, 30 of them had a mutation only in one allele and 38 had mutations in two alleles for MEFV gene. In addition, we identified 4 low-penetrance R92Q mutations for TRAPS and 3 patients, who were compound heterozygotes for the MVK gene. The diagnostic score revealed moderate sensitivity (68%) and specificity (58%) for discriminating patients who were genetically positive from those who were negative.

## Conclusion

The suggested set of variables may be used to decide on genetic screening for patients with periodic fever syndromes. However, further studies are needed to improve the sensitivity of the suggested score.

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