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POSTER PRESENTATION Open Access

Evidence based recommendations for genetic diagnosis of Familial Mediterranean Fever

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Introduction

Familial Mediterranean Fever (FMF) is a disease that starts in childhood and can lead to significant morbidity. In 2012, a European initiative called SHARE (Single Hub and Access point for pediatric Rheumatology in Europe) has been launched to optimize and disseminate diagnostic and management regimens in Europe for children and young adults with rheumatic diseases. For FMF, attention was focused on genetics.

Objectives

The aim of the SHARE recommendations in FMF is to provide a diagnostic tool for inexperienced pediatric rheumatologists to cope with FMF in their clinical practice. This is possible through a correct interpretation of the diagnostic value of *MEFV* gene mutations in predicting FMF phenotype.

Methods

Evidence-based recommendations were developed using the European League Against Rheumatism (EULAR) standard operating procedure. An expert committee was instituted, consisting of pediatric rheumatologists, and search terms for the systematic literature review were defined. Two independent experts scored articles for validity and level of evidence. Recommendations derived from the literature were evaluated by an online survey. Those with less than 80% agreement during the online survey were reformulated. Subsequently, all recommendations were discussed at a consensus meeting using the nominal group technique. Recommendations were accepted if more than 80% agreement was reached.

Results

The literature search yielded 3386 articles, of which 240 about genetics and, among them, 25 considered relevant and therefore scored for validity and level of evidence. 17 articles were scored valid and used in the formulation of the recommendations. 9 recommendations for diagnosis were suggested in the online survey and 8 were finally accepted with 100% agreement after the consensus meeting. Topics covered for diagnosis were: clinical versus genetic diagnosis of FMF; genotype – phenotype correlation; genotype – age at onset correlation; silent carriers and risk for amyloidosis; role of the specialist in FMF diagnosis.

Conclusion

The SHARE initiative provides recommendations for the diagnosis of FMF and thereby facilitates improvement and uniformity of care throughout Europe.

Disclosure of interest

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