



Familial Mediterranean Fever

We read with interest Casey et al's report on a New Zealand case of Familial Mediterranean Fever (FMF) published recently in the New Zealand Medical Journal (NZMJ).¹ It is increasingly becoming evident that this disease, although only common in Middle East populations, should also be considered in every relevant clinical situation, everywhere in the world. Following cloning of the MEFV gene, it is expected that pyrin, the MEFV-coded protein that is mutated in FMF, will attract major interest in the years to come.²

Several independent groups have recently shown that the disease is considerably spread within many populations of the Mediterranean Basin—namely, Greeks, Cypriots, Italians, and Spanish.^{3–6} Some so-called phenotype II FMF cases (renal amyloidosis being the presenting symptom) are also increasingly reported from the same areas.^{3,7} The distribution of pyrin gene mutations is complex among highly affected populations (ie, Arabs-Jewish-Armenians-Turks); that is, a limited number of mutations (less than 5) account for the vast majority of cases—as opposed to populations non-highly affected by the disease where the 5 more common mutations cover less than 70–80% of FMF chromosomes.

In these last populations, many private mutations are encountered—some 50 have been recorded until now.⁸ Therefore, non-classically affected populations provide a tool for detecting more MEFV ('atypical' / 'private') gene mutations.⁹ The authors who published their case in the NZMJ are not very clear about the case's ethnic origin*, although according to their writing it is implied that she does not belong to the aborigine population group.¹ The case proved to be V726A homozygote and her sister carried the same mutation as well; mutation V726A is proportionally common in Cypriots,⁴ and also in other populations including Greeks.³

In terms of population genetics, it would be interesting if pyrin mutations exist among Oceanian Aborigines. To the best of our knowledge, no FMF cases from this population have been reported so far. Therefore, searching in this population group for MEFV mutations by NZ physicians is strongly encouraged (in clinically relevant cases). Given the pattern of mutations among populations, such testing may reveal more FMF associated mutants.⁹ In this context, screening Oceanian populations for MEFV mutations should be a part towards a proposed Human Genetic Diversity Project.¹⁰

***NZMJ note:** As outlined in Casey et al's paper, the case's ethnic origin is Ashkenazi Jew.

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