Correspondence

Response to Simsek et al.

We thank Simsek and Pay, coauthors of the manuscript titled ‘Fabry’s disease mimicking familial Mediterranean fever’, for their interest in our article ‘Misdiagnosis of familial Mediterranean fever in patients with Anderson–Fabry disease’.

All the patients enrolled in our study are from Southern Italy, part of the Mediterranean Basin, where the frequency of familial Mediterranean fever (FMF) is high. For this reason, in cases where the patient presents not only some typical signs and symptoms of FMF but also characteristic of other systemic diseases, including Fabry disease (FD), clinicians are led to consider FMF as the primary diagnostic hypothesis. We studied 42 patients with clinical diagnosis of FMF, without genetic confirmation and some of whom did not respond to colchicine. In such subjects we analyzed the GLA gene, in order to search mutations that could be responsible for FD; these were detected in three patients, leading to the diagnosis of FD. Our data are reported in the above-mentioned paper.

Currently, we are working on a research project that includes the study of the GLA gene in 800 patients with clinical diagnosis of FMF. This project has been ‘suggested’ by the results obtained in the published study. With regard to clinical manifestations, according to our experience, it is difficult to indicate specific symptoms suggestive of FD in these subjects.

Considering only the classic form of FD, with angiokeratoma, cornea verticillata, renal involvement, hypo-anhidrosis, etc., specific symptoms could be indicative or discriminant. However, in recent years it is increasingly evident that there are atypical variants with mild symptomatology, often unclear, and with a late-onset, in a significant number of patients. This set of symptoms makes the diagnosis complicated and usually time consuming. Till today, many papers confirm this difficulty, and we consider emblematic the article of Hoffmann and Mayatepek titled ‘Fabry disease: often seen, rarely diagnosed’ (1).

Therefore it is not possible to discriminate FD from FMF just on the basis of clinical manifestations, so the genetic study is essential. In particular, we think that the study of the GLA gene should be performed in all patients with FMF that manifest unusual clinical features and/or in cases in which not all the manifestations of FMF can be explained with this diagnosis.

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