Response to Letter Regarding Belgian Fabry Study: Prevalence of Fabry Disease in a Cohort of 1000 Young Patients With Cerebrovascular Disease

Response:

We thank Drs Lidove, Joly, and Touzé¹ for their interest in our publications with regard to screening for Fabry disease in stroke patients, and for their analysis of currently available epidemiological data.²-⁶

Several issues, however, preclude drawing solid conclusions from their meta-analysis. First, relevant heterogeneity in the study designs exists, especially with regard to the stroke subtype, the stroke etiology, and the demographic data, and even the screening methodology should be taken into account.²,⁴,⁵ Second, limiting screening for Fabry disease to cryptogenic stroke patients may result in selection bias because the condition is known to be associated with cerebral microangiopathy and macroangiopathy,⁷-⁸ cardioembolic phenomena,⁹ and coagulopathy.¹⁰ Finally, exclusion of patients with the D313Y mutation is debatable because this mutation has been identified in classically affected males.¹¹

Obviously, additional data are mandatory to substantiate decision-making about screening for this treatable condition in stroke patients. For instance, a standardized assessment of first-degree relatives of those with newly diagnosed index cases may importantly enhance the diagnostic yield and may improve the prognosis of presently unrecognized Fabry patients. So far, this aspect of screening in high-risk populations has received little attention, but it is the focus of the current Belgian Fabry Study II (BeFaS II), which is still ongoing but already has resulted in diagnosis of 4 additional cases of pathogenic mutations. In addition, studies with larger cohorts of stroke patients are in progress and may provide more definite answers on the prevalence of Fabry disease in high-risk populations.

It is our opinion that the conclusion drawn by Lidove et al is premature and that it may not be justified to restrict screening for Fabry disease to stroke patients with a compatible family history only. Moreover, results of ongoing studies will be available in the near future and will provide additional information on the usefulness of screening for Fabry disease in stroke patients.

Disclosures

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