Response to Letter Regarding Article, “The Cerebral Autosomal-Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy Scale. A Screening Tool to Select Patients for NOTCH3 Gene Analysis”

Response:

The cerebral autosomal-dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) scale, recently described in this journal,1 is a screening tool predictive of the genetic diagnosis of CADASIL. The scale was developed to help clinicians select patients to be subjected to NOTCH3 gene analysis with a high probability to be affected by CADASIL. We found that this scale is accurate with optimal sensitivity and specificity values (96.7% and 74.2%, respectively); however, our results need to be confirmed and further validated. Benbir et al applied the scale to their patients in whom NOTCH3 gene analysis was performed in the suspicion of CADASIL. With the limitation of the small sample size, this report supports the capacity of this tool in distinguishing CADASIL from NOTCH3-negative patients.

We believe that the use of the scale in the clinical setting might allow to improve the recognition of patients with CADASIL in centers with less expertise in this disease without increasing the number of performed tests. Moreover, this selection might lead to characterize a more homogeneous group of NOTCH3-negative patients that could be more appropriately considered CADASIL-like (ie, patients without mutations on the NOTCH3 gene but with a CADASIL score ≥15) and in whom further genetic investigations on the NOTCH3 gene or in other genes involved in causing CADASIL-like hereditary cerebral small vessel disease could be conducted.2,3

The application of the CADASIL scale on further and larger samples is desirable to verify the accuracy of this tool in series different from the one in which it was created. An important aspect to consider with this regard is that only clinical, family history, and neuroimaging data available at the time of disease suspicion, that is when genetic testing is performed, should be used.

Disclosures

None.

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