tricular areas. Although it is not typical for patients with adult MLD, we think that the diagnosis of MLD cannot be excluded, considering the heterogeneity of imaging manifestations in patients with MLD. Other clinical data should be considered together for final diagnosis.

Second, residual ARSA activity is highly variable between patients with MLD with similar presentations and for individual patients at repeated testing because of the limitations of assays and the contributions of other genetic or environmental factors. The levels of ARSA activity in patients with MLD from East Asia seem much higher than that of patients with MLD from other areas. In a review that included 11 patients with MLD from East Asia, the levels of ARSA activity ranges from 14.50% to 30.75% of the normal controls. In addition, Lorioli et al3 have described a presymptomatic patient with MLD with an ARSA activity equal to 50% of normal values, which is similar to a carrier in this family. Therefore, we still advised the patient in our case report1 (a Chinese patient) to undergo an ARSA gene test although his ARSA activity was half of the normal lower limit.

Considering the presence of ARSA pseudodeficiency and the variety of the ARSA activity among patients with MLD, the diagnosis or exclusion of MLD should be based not only on the ARSA activity but also on other clinical data, such as genetic analyses. Moreover, other causes of leukoencephalopathies, including cobalamin C disease, should be considered even though lower ARSA enzyme activity was found.

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