Editors’ Note: In WriteClick this week, Drs. Lynch and Houlden point out an error in the Mystery Case Responses section of “Mystery Case: CSF-1R mutation is a cause of intracranial cerebral calcifications, cysts, and leukoencephalopathy,” in which CSF-1R-related leukoencephalopathy is mistaken to be the same as Labrune syndrome. Drs. Lynch and Houlden and author Ayrignac discuss the differences between the 2 disorders. A correction appears on page 1979.

—Megan Alcauskas, MD, and Robert C. Griggs, MD

LETTER RE: MYSTERY CASE: CSF-1R MUTATION IS A CAUSE OF INTRACRANIAL CEREBRAL CALCIFICATIONS, CYSTS, AND LEUKOENCEPHALOPATHY

David S. Lynch, Henry Houlden, London: Ayrignac et al.1 presented an interesting case that highlighted the importance of CSF-1R in adult-onset leukoencephalopathies. However, we are concerned that the discussion by Dr. Ganesh in the Mystery Case Responses section confused 2 different diseases as the same.1 This case clearly described a typical presentation of CSF-1R-related disease with apparent autosomal dominant inheritance. However, Dr. Ganesh described this as a case of Labrune syndrome (leukoencephalopathy with calcifications and cysts), an autosomal recessive disorder caused by mutations in SNORD118.2 While calcifications occur in both syndromes, they are far more widespread and severe in Labrune syndrome and the imaging appearance of both conditions is distinctive. Dr. Ganesh incorrectly attributed 11% of adult-onset leukoencephalopathy to Labrune syndrome by referencing Guerreiro et al.,3 who clearly referred to CSF-1R-related disease.

In our experience of adult-onset leukoencephalopathy, the most common causes are classic leukodystrophies, CSF-1R, CADASIL, and AARS2 mutations. Therefore, it is not surprising that so few residents chose Labrune syndrome or Coats-plus syndrome as likely diagnoses.


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