# WriteClick® Editor's Choice

**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.

1. Ayrignac X, Mouzat K, Magnin E, et al. Mystery Case: *CSF-1R* mutation is a cause of intracranial cerebral calcifications, cysts, and leukoencephalopathy. Neurology 2016; 87:e262–e263.
2. Jenkinson EM, Rodero MP, Kasher PR, et al. Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nat Genet 2016; 48:1185–1192.
3. Guerreiro R, Kara E, Le Ber I, et al. Genetic analysis of inherited leukodystrophies: genotype-phenotype correlations in the CSF1R gene. JAMA Neurol 2013;70:875–882.