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I read with interest the article by Sener (1), in which he demonstrates a peak at 3.50 parts per million in proton MR spectroscopy of the brain in a patient purported to have "van der Knaap syndrome." The peak is interpreted as representing glycine. The author relates this finding to the observation of elevated CSF glycine in patients with "van der Knaap syndrome" (2).

Regrettably, the author has confused two different disorders of great interest to members of our department: megalencephalic leukoencephalopathy with subcortical cysts (MLC) and vanishing white matter (VWM). The patient described by Sener has MLC, as evident from the clinical course and MR imaging findings (1), whereas elevated CSF glycine has been found in patients with VWM (2). Glycine elevation in the CSF of patients with VWM is, however, far below the level of detection for in vivo MR spectroscopy (2).

Confusion arises from attaching names of authors to diseases, particularly when the same author has contributed to the detection and description of multiple diseases. A good example is Jean Aicardi, whose name is associated with a particular type of neonatal epilepsy (3), a syndrome in girls characterized by agenesis of the corpus callosum, cortical dysplasia, and retinal abnormalities (4), and the white matter disorder Aicardi-Goutières syndrome (5). In addition, many people have difficulty pronouncing "Van der Knaap," the name is frequently misspelled as "van der Knapp," and attaching it to a disease leads to disturbing statements, such as "My doctor and I think that my son has your disease." I suggest that the names MLC and VWM (alternatively called CACH, for childhood ataxia with central hypomyelination) be used to indicate the respective disorders.

Finally, what about the observation of possibly elevated glycine in proton MR spectroscopy of the brain in an MLC patient? We have performed proton MR spectroscopy in several MLC patients and have not observed any elevation in glycine. We have also analyzed the CSF of several MLC patients and have not found elevated levels of glycine. The quality of the spectrum published is suspect and suggests that we could be looking at noise or at an artifact related to water-suppression problems.

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References

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Reply:

As mentioned in my previous letter, I understand and accept that the disease has two genetically different forms, MLC and VWM. I agree that this distinction should be clarified. If I were writing the article today, I would stress the existence of these two forms. At the end of 2000, when I wrote the article, I did not pay attention to the two forms because I was focused on describing a new peak, glycine. Presently, an international journal has accepted a manuscript dealing with a variety of conditions, including ischemia, tumors, leukoencephalopathies (including a new patient with a disease referred to as "MLC," without reference to a person's name), and some other lesions, all of which have distinct glycine peaks. In this article, too, my main concern was describing glycine without detailing the related disease processes.

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