RESPONSE TO LETTER TO THE EDITOR

Diagnosis of Wilson's disease

We appreciate the comments made by Dr Brewer.[1,2] He argues that the diagnosis of Wilson's disease (WD) should have been made using quantitative copper assay of liver specimen, and suggests that this is the 'gold standard'. Unfortunately, contrary to his statement, there is no gold standard for diagnosis in WD. If this were to be true, there would be no need for diagnostic criteria as have been developed for this disease. Indeed, as we have experienced, the diagnosis of WD in an individual with chronic hepatic disease is not straightforward. We concur that liver copper measurement is a valuable diagnostic test, but the diagnostic yield has been subject to debate and as Dr Brewer agrees, (a)typical WD cases with normal copper quantification have been reported.[3]

Indeed, in hindsight with all the diagnostic clues available as laid out in the paper, it is fairly straightforward to make the diagnosis in this case. Unfortunately, as we have learned, clinical reality is sometimes different and diagnosing an atypical case of WD may be a challenging task. Then why publish such a case? The prime reason for publication of our case was to post the readers of this journal on the diagnostic pitfalls in WD. As duly noted there are multiple confounders in WD such as atypical age at presentation, non-characteristic laboratory parameters and unusual clinical features.[4] In addition, we hope to draw attention to the possibility of mutational analysis of ATP7B to make the right diagnosis. Dissemination of this information helps other clinicians to make better decisions for their patients in the future, and that is central to this case report.

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