November 13, 2018

Dennis A Bloomfield & Sandro Vento
Editors-in-Chief
World Journal of Clinical Cases

Dear Editor:

Thank you for all comments from reviewers on our case report titled "First Report of Variant Late Infantile Ceroid Lipofuscinosis in Jordan" for submission in the *World Journal of Clinical Cases*.

We have incorporated your feedback and believe it has significantly improved the quality of the article.

Response to reviewers' comments

Reviewer 1

Answer:

1- As used here, "novel" actually refers to the fact that this is the first report or record of such a case in Jordan. Hence, we have changed the title accordingly to "First report of Variant Late Infantile Ceroid Lipofuscinosis in Jordan".
2- The differential diagnoses included:

a-Leukodystrophy: The MRI results were not suggestive of this condition

b-Subacute sclerosing panencephalitis: The EEG did not reveal any periodic patterns indicating the presence of this condition.

d-Tuberous sclerosis: No skin manifestations suggestive of this disorder were present.

e-Childhood disintegrative disorder (CDD)

The first three differential diagnoses were excluded based on history, examination, and investigations; hence, a diagnosis of childhood disintegrative disorder was established. When we were given the opportunity to perform whole exome sequencing on a group of patients from our hospital, we selected these two cases since we did not yet have a final diagnosis for them. This allowed us to deliver a definitive diagnosis and increase our awareness of the value and potential of whole exome sequencing for similar cases.

3- MRI reports were found for the two cases, but the films were not available.

In addition to CBC, liver and kidney function tests and serum electrolyte and blood sugar levels were assessed and no abnormalities were observed. Other test results, such as serum amino acid profile, B12

levels, thyroid function tests, and a brain CT scan were all normal. Subsequent brain magnetic resonance imaging (MRI) revealed mild fronto-parietal brain atrophy.

Reviewer 2

Answer:

Thank you for your positive comments.

Reviewer 3

Answer:

Hydrogen 1 (1H) magnetic resonance (MR) spectroscopy enables noninvasive *in vivo* quantification of metabolite concentrations in the brain. Currently, metabolite concentrations are most often presented as ratios (e.g., relative to creatine) rather than as absolute concentrations. A recent study (*Magnetic resonance spectroscopy of the frontal region in patients with metabolic syndrome*)* concluded that the NAA/Cr and Cho/Cr ratios of the frontal region can differentiate patients with metabolic syndrome from healthy volunteers.

**Reference*:

El-mewafy Z, Abdel Razek AAAK, El-Eshmawy M, Abo El-Eneen N, EL-Biaomy A. MR spectroscopy of the frontal region in patients with metabolic syndrome: Correlation with anthropometric measurement. *Polish J Radiol* 2018; 83: e215-e219.

Thank you for your consideration of the new version of our manuscript with appropriate changes and edits incorporated. We look forward to hearing from you.

Sincerely,
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