

Reviewer: 1

(1) Are there any prenatal signs of this rare syndrome?

(Response: Thanks for your comments. In this case, the mother of the child did not have clear indications for prenatal diagnosis during pregnancy, but for couples who have given birth to such children, genetic counseling and prenatal diagnosis are required for the next pregnancy to avoid the birth of such children. Please see the revision in line 207-211.)

(2) In the discussion part, I suggest providing a table summarising previously reported cases.

(Response: Thanks for your comments. We had provided a table summarising previously reported cases in the Discussion. Please see the revision in line 163-188 and the attachment Table 1.)

(3) As you are planning to follow up this patient, it would add more scientific value to your article if you provide a follow-up plan and possible treatment and rehabilitation measures that could be used in such cases. In addition, an algorithm would supplement your case report greatly.

(Response: Thanks for your comments. Because there is currently no effective treatment for the disease, the prognosis of the child is poor. In the follow-up, we will follow up the life of the child and plan to follow up every 1-2 years. In particular, we will pay attention to muscle and intellectual development with muscle and brain MRI examination, and carry out symptomatic treatment and intervention. Please see the revision in line 202-205. An algorithm can be found in our previous article. Please see the revision in line 131-133.)

Reviewer: 2

- (1) What instrument was used to measure muscle strength and developmental delay on this patient? It would be great if the the author(s) comparison with normal measurements for children of the same age as a comparison to this patient.

(Response: Thanks for your comments. The child has had an electromyography, Please see the revision in line 50-55.

Normal children raise their heads at 4 months of age, they can stand up independently, they will turn their heads and look for them when they hear the sound, they can be amused, and they will also make a first babble. However, the patient in our case had attention deficit disorder and delayed motor performance. Please see the revision in line 108-111.)

- (2) What is the prognosis of this patient?

(Response: Thanks for your comments. Because there is currently no effective treatment for the disease, the prognosis of the child is poor. Please see the revision in line 150.)

- (3) What is the follow-up plan? how will the author(s) plan to observe the progression of the disease?

(Response: Thanks for your comments. Because there is currently no effective treatment for the disease, the prognosis of the child was poor. In the follow-up, we will follow up the life of the child and plan to follow up every 1-2 years. In particular, we will pay attention to muscle and intellectual development with muscle and brain MRI examination, and carry out symptomatic treatment and intervention. Please see the revision in line 202-205.)

Thanks again for your consideration of this manuscript.

Best wishes!

Yours sincerely

Hua Jin