Author’s response to reviews

Title: Idiopathic Inflammatory Myopathy Comorbid with Sturge-Weber Syndrome: A Case Report

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Re: NURL-D-18-00489R1

Dear Tim Hagenacker and reviewers,

Thank you and all the reviewers for your time and constructive comments and suggestions concerning our manuscript entitled “Polymyositis Comorbid with Sturge-Weber Syndrome: A Case Report”. We have studied their comments carefully and have made corrections accordingly. All the revised parts in the manuscript are highlighted by the red color. The point-to-point response to the reviewers’ comments is attached at the end of this letter.

We hope that the revised manuscript will satisfy the reviewers and meet for the standard of publication in your journal.

With best regards,

Yours sincerely,

Ping Fu,

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Reviewer reports:

Jana Zschüntzsch (Reviewer 1): The authors submitted a manuscript with the title Polymyositis Comorbid with Sturge-Weber Syndrome: A Case Report. In this case study, the author report of a Chinese female with a known Sturge-Weber-Syndrome (SWS) which presented with a steroid-responsiveness hyper-CKemia and muscle weakness.

The authors point out that although one rare disease was already diagnosed a thoughtful differential diagnosis should be performed upon new and untypical symptoms.

The clinical presentation, the EMG as well as the responsiveness to steroids might be in line with the PM, but the Ro52-antibody is associated with overlap myositis (OM) as well as with other connective tissue disorders such as Sjgren's syndrome, SLE, and systemic sclerosis.


Meanwhile, it is accepted that PM is over diagnosed and more cases can be reclassified as OM.

Answer: We studied the literatures the reviewer recommended carefully and some new content was added in the paper.

Question 2: Please clarify, which kind of muscle strength measurement you used (MRC ? ).

Answer: The muscle strength was evaluated by the UK Medical Research Council (MRC) criteria.

Question 3: The sentence: "In our study, it was revealed that the somatic GNAQ mutations may be associated with the pathways of PM" is not clear. Please explain.

Answer: GNAQ mutations were reported in SWS. And recently a case of SWS with rhabdomyolysis was reported to be associated with GNAQ mutation. Therefore, we hypothesized that GNAQ mutation may be involved in our case since this is a typical SWS with inflammatory myopathy.

Albert Selva-O'Callaghan (Reviewer 3): The authors report the case of a 41-year-old female patient diagnosed with Sturge-Weber syndrome who develop a full-blown myositis.
This reviewer raised several questions and comments that need to be clarified before publication.

1. Does hypothyroidism play a role? It is well known that hypothyroid myopathy can be misdiagnosed as an inflammatory myopathy. Please clarify.

Answer: Hypothyroid myopathy can be misdiagnosed as inflammatory myopathy and in this case, the patient was first diagnosed as hypothyroid myopathy. Levothyroxine was taken. However, the weakness was not improved. On admission to our department, thyroid function was within normal range. The treatment response and lab results did not support the diagnosis of hypothyroid myopathy.

2. Although the clinical picture resembles myositis (autoimmune origin) and the patients seems to respond to immunosuppressive therapy, a muscle biopsy would undoubtedly added more information to the reader and reinforce the diagnosis. We encourage the authors to avoid the term "polymyositis" (See Lancet Neurology 2018) given that in the myositis and scientific community nowadays is not more used because it is considered an exclusion criterion (hence the relevance of the muscle biopsy that may help to diagnose immune mediated necrotizing myopathy or sIBM at onset).

Answer: We studied the recent literature following the reviewer’s suggestions. Polymyositis is now considered as an exclusion criterion. In this case, no muscle biopsy was obtained. Therefore, we changed the term polymyositis to idiopathic inflammatory myopathy (IIM) in the context.

3. We suggest to apply the Classification Criteria recently published in Ann Rheum Dis / Arthritis Rheum 2017, Lundberg et al) for support the diagnosis in this case (webcalculator included in the article).

Answer: According to the webcalculator in the article, our case was definite IIM and the subgroup could be inclusion body myositis. The content was added in the paper according to the suggestion.