Reviewer's report

Title: COPA syndrome in an Icelandic family caused by a recurrent missense mutation in COPA

Version: 0 Date: 14 Jun 2017

Reviewer: Anthony Shum

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The manuscript by Jensson et al. reports an Icelandic family with the COPA syndrome that through whole genome sequencing identified a p.Glu241Lys missense mutation previously reported in 2015. This is the second family with this mutation to be reported in the literature and importantly provides further confirmation of both the pathogenic nature of the mutation and the clinical features of the disease. The genetic analysis is outlined thoroughly and is convincing. If space limitations allow, further discussion of the autoimmune and rheumatologic features would be helpful, including which medications were used to treat the patients. It would also be of use to the medical community to note whether joint erosions were present and also indicate the autoantibody titers, if known.

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

Yes

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Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:

Acceptable
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