Reviewer's report

Title: Infantile-onset inflammatory bowel disease in a patient with Hermansky-Pudlak syndrome: a case report

Version: 1  Date: 01 Nov 2018

Reviewer: Bernadette Gochuico

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Ishihara, et al. report a case of an infant with Hermansky-Pudlak syndrome (HPS) and inflammatory bowel disease, and genotyping identified previously reported compound heterozygous variants in HPS1. The authors are commended for their efforts to report this case with a rare disease. The patient's very young age at diagnosis and clinical response to therapy are of interest.

Comments:

1. In the abstract (line 43), the authors state that the patient "was diagnosed with Crohn's-like inflammatory bowel disease at 11 months." Information should be added to the abstract to describe the basis for establishing this diagnosis.

2. The authors describe the endoscopic biopsy findings (lines 87-88). The report would be improved by adding this description to the abstract and showing the histopathologic images in a figure.

3. The authors state in the Discussion (lines 111-112), "IBD is more common in HPS-1, -4, and -6;" but do not indicate what it is more common than. Perhaps the authors should consider revising the statement to "IBD is reported in patients with HPS-1, -4, and -6; ..."

4. Genetic analysis is a useful diagnostic modality for many inherited disorders, as exemplified by this case. In this case, clinical features (e.g., oculocutaneous albinism with nystagmus) are consistent with an underlying diagnosis of HPS. This report would be strengthened by the addition of a short discussion of the differential diagnosis of infantile-onset IBD, and clinical features that aid in diagnosing specific etiologies of infantile-onset IBD.

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.

Unable to assess

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

Yes

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript

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

Acceptable

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