Reviewer’s report

Title: Constitutional mutation in CDKN2A is associated with long term survivorship in multiple myeloma

Version: 0 Date: 14 Jul 2017

Reviewer: Sabarinath Venniyil Radhakrishnan

Reviewer's report:

In this case report "Constitutional mutation in CDKN2A is associated with long term survivorship in multiple myeloma: a case report" Shah et al has described a patient who developed myeloma at a young age and subsequently also had multiple melanomas and other tumors and was found to have a germline mutation in CDKN2A. The case presentation is interesting but the work up is incomplete in my opinion. Please see my comments below

1) The authors have not described how the genetic testing was done and what the allele frequency was?

2) I understand that the bone marrow for MM was done 30 years back and so would not be possible to have genetic studies done on the plasma cells but she has recent lung cancer and also multiple melanomas but no mention of genetic abnormalities in these tumors. It would be very interesting to know if there is a loss of heterozygosity of these this locus in these tumors. Furthermore there is no family history of melanoma or pancreatic cancer as described in the report and so it would be speculative to assume this mutation is associated with myeloma.

3) Please provide evidence of the pathogenic nature of this missense mutation as reported on line 54. The previous report of germline mutation in CDKN2A associated with MM is not a missense mutation.
Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

No

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

No

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

No

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

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Please indicate the quality of language in the manuscript:

Acceptable

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