Reviewer’s report

Title: Effect of a rare genetic variant of TM7SF4 gene on osteoclasts of patients with Paget's disease of bone

Version: 0 Date: 04 Sep 2017

Reviewer: Javier del Pino

Reviewer's report:

Thank you for letting me review this interesting study. It is a study of a rare genetic variant in the TM7SF4 gene, encoding DC-STAMP, observed in a PDB SQSTM1 negative cohort. Although he has been associated with PDB, this study explores the functional effects of this variant on osteoclast phenotype in PDB. The results increase the knowledge in genetic factors involved in the PDB’s pathogenesis. The main limitation of the study is the small sample included in the study. The authors mention that it is due to the rare frequency of the variant in their PDB cohort.

Minor concerning

Although in the method section is included the number of patients, it should also be included in the abstract.

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

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.

I am able to assess the statistics
Quality of written English
Please indicate the quality of language in the manuscript:

Acceptable

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