Reviewer's report

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

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Reviewer: S Reddy

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Laetitia colleagues examined the functional effects of TM7SF4 gene encoding DC-STAMP on osteoclast fusion in patients with Paget's disease (PDB) compared to healthy control subjects and non-carrier patients. The findings are interesting that DC-STAMP expression was elevated in patients with carrier of the variant than in non-carrier patients, which will imply in PDB prognosis and complications. The data is analyzed with statistical methods appropriately. The manuscript is thoroughly cited and conclusive. I have the following few minor suggestions.

1. Results - In abstract it is noted that DC-STAMP expression was elevated in patients with carrier of the variant than in non-carrier patients. Please clarify the sentence in Results section (p.8) that nuclei/multinucleated cell was higher in patients carrier of the variant than in healthy controls but not different from non-carrier patients.

2. Figure 2 show immunofluorescence of DC-STAMP expression in osteoclast cultures with similarity in healthy and carriers with PDB. I suggest removing the words (p.9; line 5) "but not at the plasma membrane" as DC-STAMP is known as a transmembrane protein.

3. I suggest to include a citation for OS-9 (p.9; line 45).

4. Figs.1&3-the authors may consider noting index (health controls; paget patients non mutated; paget patients mutated) once either in the figure or in the figure legend as they are same for all the graphs. Also, shorten (write in two lines than all in single line) the axis labeling in graphs.

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

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Please indicate the quality of language in the manuscript:

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