PPSNV:A novel predictor for pathogenicity of SNV based on ensemble learning

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1. Introduction

With the next-generation sequencing (NGS) technologies developing and personalized medicine applied, it is practical for doctors to conduct genetic testing in clinical diagnosis. Among the numerous output of genome sequencing, nonsynonymous single nucleotide variant(SNV) is a common type of genetic alteration which probably cause disease. However, the actual consequence of a genetic variant veils behind complex biological mechanisms and identifying whether a SNV is pathogenic accurately remains challenging.

Good or Bad, it is a question. To answer the question, many tools are developed to predict the pathogenicity of genetic variants.

1. methods

The next subsections provide instructions on how to insert figures, tables, and equations in your document.

* 1. Datasets

We use the dataset provided of a previous work, containing 5 subsets and 45081 variants which are collected from publicly available and commonly used benchmark datasets. All variants in the dataset are labeled and the variants overlapping with CADD training data are removed. After removing overlapping variants between 5 subsets, *HumVar, ExoVar, VariBenchSelected, predictSNPSelected* are merged into a new dataset which we use to train our framework. As for *SwissVarSelected,* we leave it as an independent test dataset based on which we compare our proposed framework with other methods.

The details of our datasets are listed below.

* 1. Training
  2. Features

1. RESULT
   1. Characterization of Features
   2. Performance of PPSNV compared with other methods
2. DISCUSSION
3. Citing Related Work

This section cites a variety of journal [5, 15], conference [1, 6, 8, 12, 13], and magazine [3] articles to illustrate how they appear in the references section. It also cites books [9, 10], a technical report [7], a PhD dissertation [4], an online reference [14], a software artifact [11], and a dataset [2].

ACKNOWLEDGMENTS

Acknowledgments are placed before the references. Add information about grants, awards, or other types of funding that you have received to support your research. Author can capture the **grant sponsor information**, by selecting the grant sponsor text and apply style ‘GrantSponsor’. After this, select grant no and apply ‘GrantNumber’ from style panel. Example of Grant sponsor: Competitive Research Programme and example of Grant no: CRP 10-2012-03.

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A  APPENDICES

In the appendix section, three levels of Appendix headings are available.

A.1 General Guidelines (AppendixH2)

1. Save as you go and backup your file regularly.
2. Do not work on files that are saved in a cloud directory. To avoid problems such as MS Word crashing, please only work on files that are saved locally on your machine.
3. Equations should be created with the built-in Microsoft® Equation Editor included with your version of Word. (Please check the compatibility at <http://tinyurl.com/lzny753> for using MathType.)
4. Please save all files in DOCX format, as the DOC format is only supported for the Mac 2011 version.
5. Tables should be created with Word’s “Insert Table” tool and placed within your document. (Tables created with spaces or tabs will have problems being properly typeset. To ensure your table is published correctly, Word’s table tool must be used.)
6. Do not copy-and-paste elements into the submission document from Excel such as charts and tables.
7. Footnotes should be inserted using Word’s “Insert Footnote” feature.
8. Do not use Word’s “Insert Shape” function to create diagrams, etc.
9. Do not have references appear in a table/cells format as it will produce an error during the layout generation process.
10. MS Word does not consistently allow the original formatting to be modified in the text. In these cases, it is best to copy all the document’s text from the specific file and paste into a new MS Word document and then save it.
11. At times there are font problems such as “odd” stuff/junk characters that appear in the text, usually in the references. This can be caused by a variety of reasons such as copying-and-pasting from another file, file transfers, etc. Please review your text prior to submission to make sure it reads correctly.

A.1.1 Preparing Graphics (AppendixH3)

1. Accepted image file formats: TIFF (.tif), JPEG (.jpg).
2. Scalable vector formats (i.e., SVG, EPS and PS) are greatly preferred.
3. Application files (e.g., Corel Draw, MS Word, MS Excel, PPT, etc.) are NOT recommended.
4. Images created in Microsoft Word using text-box, shapes, clip-art are NOT recommended.
5. IMPORTANT: All fonts must be embedded in your figure files.
6. Set the correct orientation for each graphics file.

A.2 Placeholder Text

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