Abstract Title: GeneTree: an innovative solution to build simultaneously a pedigree, downloadable in BOADICEA and CanRisk files, and the clinical history of a family
Control Number: 1031
Topic: 18. Bioinformatics, Machine Learning and Statistical Methods
Presentation Preference: Oral Presentation
Authors:

Jean-Marie Ravel*[1]

[1]CHRU de Nancy, Laboratoire de génétique médicale, Service de génétique clinique, Nancy, France

Background/Objectives:

To date, no free solution allows the generation of a family pedigree that can be quickly modified and exported with the corresponding text.

Methods:

I build a web application host by GitHub servers.

Results:

This application is an integrated tool to help doctors and genetic counsellors in their genetic counselling. The application is available without priori installation in French and English :
https://jimouse.github.io/GeneTree/

It has a specific mode for oncogenetic consultation and a mode using HPO phenotypes. The family can be loaded (JSON or BOADICEA files) or created from a standard or custom structure, then completed via a table or a graphical interface, both interconnected.

The pedigree can be exported in several file formats and modified by a vector editor (PDF, SVG) or can be printed directly.

Finally, this application allows the automatic generation of text based on the content of the table.

Moreover, a second interface, specifically designed for patients, allow them to fill their familial information prior to the consultation.

Conclusion:

This tool is extremely time-saver and has been particularly optimised for oncogenetic consultations to avoid triple entry (text-tree-boadicea risk score). This is the first application allowing text-generation based on a pedigree.

References:

CanRisk Tool—A Web Interface for the Prediction of Breast and Ovarian Cancer Risk and the Likelihood of Carrying Genetic Pathogenic Variants. Carver, T. et al. Cancer Epidemiol Biomarkers Prev (2020). doi:10.1093/bioinformatics/btx705

pedigreejs: a web-based graphical pedigree editor. Carver T, Cunningham AP, Babb de Villiers C, Lee A, Hartley S, Tischkowitz M, et al. Bioinformatics doi:10.1093/bioinformatics/btx705

Grants: