

COMMERCIALLY AVAILABLE INDUSTRY-RELEVANT SOFTWARE IN THE EDUCATION OF GENOME VARIANT CURATION

Callum J. Vidor*, Hendrika M. Duivenvoorden*, Desirée du Sart, Robert J. Bryson-Richardson, and Thomas J. Hiscox

Presenting Author: Dr Callum J Vidor (callum.vidor@monash.edu)
School of Biological Sciences, Monash University, Clayton, VIC 3800, Australia
*Co-primary investigators

KEYWORDS: work readiness, genetics, genomics, Masters, software

BACKGROUND

Genome analytics is a drastically expanding field, and there is an increasing demand for individuals with the necessary skillset to analyse the genome data that is being generated. A new Masters by coursework was developed to train individuals in genome analytics. Institutions analysing genomes commonly utilise in-house analysis tools, but increasingly commercial software packages that integrate AI are being considered in the research and diagnostic space (De La Vega et al., 2021).

AIMS

1. Evaluate the effectiveness of commercially available software as a tool for teaching variant analysis, classification and curation, enabling the analysis of real-world case examples for the teaching, training and assessment of students in the field of diagnostic genome analysis.
2. Assess students' perspectives on work readiness after using commercially available tools in the educational environment.

DESIGN AND METHODS

Students were exposed to the use and limitations of a commercial software package for Human genome curation during two core units of the course. This software was used as part of both in-class training and in their assessment case studies. Students were invited to voluntarily complete an online survey including qualitative and quantitative components featuring Likert scale questions, both pre and post exposure to the software. Paired data from 23 individuals (73% response rate), most aged between 18 and 25, were recorded and anonymised prior to analysis. Qualitative data were thematically coded blind by two individuals independently using emergent coding (Charmaz, 2008).

RESULTS AND CONCLUSIONS

This project indicates that after the completion of the units that integrated commercially available industry software, we measured increased student confidence (increase in percentage reporting fairly confident or higher) in joining the genetic analysis workforce (significant change from 37% to 70%) and in completing job-specific tasks (significant increase in 7 out of 9 tasks of between 28% to 39%). The aspects of their studies the students valued in relation to these changes and their perception of the usefulness of integration of the commercial software were elucidated from qualitative theming, and can inform others looking to integrate commercially available software within their tertiary degree.

REFERENCES

- Charmaz, K. (2008). Grounded theory as an emergent method. In S. N. Hesse-Biber & P. Leavy (Eds.), *Handbook of emergent methods*. (pp. 155-170). The Guilford Press.
- De La Vega, F.M., Chowdhury, S., Moore, B., Frise, E., McCarthy, J., Hernandez, E.J., Wong, T., James, K., Guidugli, L., Agrawal, P.B., Genetti, C.A., Brownstein, C.A., Beggs, A.H., Löscher, B.S., Franke, A., Boone, B., Levy, S.E., Öunap, K., Pajusalu, S., ... Kingsmore, S.F. (2021). Artificial intelligence enables comprehensive genome interpretation and nomination of candidate diagnoses for rare genetic diseases. *Genome Med*, 13(1), 153. <https://doi.org/10.1186/s13073-021-00965-0>

Proceedings of the Australian Conference on Science and Mathematics Education, The University of Tasmania, 30 August – 1 September 2023, page 72, ISSN 2653-0481.