Event title	WORKSHOP: Variant calling in humans, animals and plants with Galaxy
Event type	Workshop
Date of event	25/05/2021
Time of event	1 - 5pm AEST
Topic description	Variant calling in polyploid organisms, including humans, plants and animals, can help determine single or multi-variant contributors to a phenotype. Further, sexual reproduction (as compared to asexual) combines variants in a novel manner; this can be used to determine previously unknown variant - phenotype combinations but also to track lineage and lineage associated traits (GWAS studies), that all rely on highly accurate variant calling. The ability to confidently call variants in polyploid organisms is highly dependent on the balance between the frequency of variant observations against the background of non-variant observations, and even further compounded when one considers multi-variant positions within the genome. These are some of the challenges that will be explored in the workshop. In this online workshop we focused on the tools and workflows available for variant calling in polyploid organisms in Galaxy Australia. The workshop provided opportunities for hands-on experience using Freebayes for variant calling and SnpEff and GEMINI for variant annotation. The workshop made use of data from a case study on diagnosing a genetic disease however the tools and workflows are equally applicable to other polyploid organisms and biological questions. Access to all of the tools covered in this workshop was via Galaxy Australia, an online platform for biological research that allows people to use computational data analysis tools and workflows without the need for programming experience. The Galaxy tutorial used in the workshop is freely available via the Galaxy Training Network. Wolfgang Maier, Bérénice Batut, Torsten Houwaart, Anika Erxleben, Björn Grüning, 2021 Exome sequencing data analysis for diagnosing a genetic disease (Galaxy Training Materials). https://training.galaxyproject.org/training-material/topics/variant-analysis/tutorials/exome-seq/tutorial.html Online; accessed 25 May 2021
Format description	Workshop, online via Zoom as described in https://zenodo.org/record/4158583 Dr Gareth Price lead the training by introducing

	introducing key concepts and demonstrating the steps involved in the variant calling process. Participants then moved into breakout rooms where they had the chance to apply these skills in breakout sessions where they were supported by facilitators. Participation was free but subject to application with selection. Applications were reviewed by the organising committee.
Identifier(s)/URL	https://www.biocommons.org.au/events/variants-polyploid
Licence	Materials are shared under a Creative Commons Attribution 4.0 International agreement unless otherwise stated on the materials
Keywords	Variant calling http://edamontology.org/operation_3227 Genetic variation analysis http://edamontology.org/operation_3197 SNP annotation http://edamontology.org/operation_3661
Contact	Melissa Burke melissa@biocommons.org.au
Audience	This workshop was for Australian researchers who have or will work on variant calling in polyploid organisms as part of their projects. Participants were required to be associated with an Australian organisation and provide an appropriate organisational email address for their application to be considered.
Prerequisites	• Familiarity with the concepts of variant calling. • Some experience with the basics of using Galaxy Australia such as setting up a history, uploading data and running tools. • No programming experience is required.
Technical requirements	• A Galaxy Australia account • This workshop was run using Galaxy version 21.01 and made use of access to Galaxy Training Infrastructure as a Service (TIaaS). • Slack was used to facilitate discussions. • Access to the internet, speakers, a webcam, microphone and Zoom.
Learning outcomes	• Identify genetic variants in samples based on exome sequencing data

	• Identify causative variants associated with a given phenotype • Apply Freebayes for variant calling • Apply SnpEff and GEMINI for variant annotation
Lead Trainer	Dr Gareth Price, QCIF Bioinformatics
Facilitators	Khalid Mahmood, Melbourne Bioinformatics Adele Barugahare, Monash University
Related work	Wolfgang Maier, Bérénice Batut, Torsten Houwaart, Anika Erxleben, Björn Grüning, 2021 Exome sequencing data analysis for diagnosing a genetic disease (Galaxy Training Materials). https://training.galaxyproject.org/training-material/topics/variant-analysis/tutorials/exome-seq/tutorial.html Online; accessed 25 May 2021 Batut et al., 2018 Community-Driven Data Analysis Training for Biology Cell Systems 10.1016/j.cels.2018.05.012