Variant Curator/Research Scientist

**POSITION NO** 0061632

**CLASSIFICATION** Level A

**SALARY** $80,258 - $108,906 p.a.

**SUPERANNUATION** Employer contribution of 17%

**WORKING HOURS** Full time (1.0 FTE)

**BASIS OF EMPLOYMENT** Fixed term until 31 December 2025

**OTHER BENEFITS** [https://about.unimelb.edu.au/careers/staff-benefits](https://about.unimelb.edu.au/careers/staff-benefits)

**HOW TO APPLY** Online applications are preferred. Go to [http://about.unimelb.edu.au/careers](http://about.unimelb.edu.au/careers), select the relevant option ('Current Opportunities' or 'Jobs available to current staff'), then find the position by title or number.

**CONTACT FOR ENQUIRIES ONLY**
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*Please do not send your application to this contact*

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For information about working for the University of Melbourne, visit our website: [about.unimelb.edu.au/careers](http://about.unimelb.edu.au/careers)
Acknowledgement of Country

The University of Melbourne acknowledges the Traditional Owners of the unceded land on which we work, learn and live: the Wurundjeri Woi Wurrung and Bunurong peoples (Burnley, Fishermans Bend, Parkville, Southbank and Werribee campuses), the Yorta Yorta Nation (Dookie and Shepparton campuses), and the Dja Dja Wurrung people (Creswick campus).

The University also acknowledges and is grateful to the Traditional Owners, Elders and Knowledge Holders of all Indigenous nations and clans who have been instrumental in our reconciliation journey.

We recognise the unique place held by Aboriginal and Torres Strait Islander peoples as the original owners and custodians of the lands and waterways across the Australian continent, with histories of continuous connection dating back more than 60,000 years. We also acknowledge their enduring cultural practices of caring for Country.

We pay respect to Elders past, present and future, and acknowledge the importance of Indigenous knowledge in the Academy. As a community of researchers, teachers, professional staff and students we are privileged to work and learn every day with Indigenous colleagues and partners.

Position Summary

The variant curator/research scientist for The Advanced Genomics Consortium (TAGC) Ocular Genotyping Hub will undertake duties relating to assessing and interpreting genomic variants from associated with inherited retinal diseases (IRDs). This project aims to uncover target genes for innovative diagnostics and treatments for IRDs and establish a local genome sequencing pipeline for diagnosing genetic eye diseases.

This position is located within the Vision Optimisation Unit within the Departments of Optometry and Vision Sciences, and Surgery (Ophthalmology) at the University of Melbourne. The Principal Investigator of this unit is Associate Professor Lauren Ayton. The Unit’s main research focus is supporting the development of vision restoration treatments and therapies for people with rare inherited retinal diseases (IRDs). Our team has strong collaborations within the University and the Centre for Eye Research Australia, as well as interstate and internationally (including Harvard and Oxford Universities).

Our team’s Flagship project is the Victorian Evolution of Inherited Retinal Diseases Natural History Registry (VENTURE) study, which is recruiting people for research studies and to be notified of future clinical trial opportunities, such as retinal gene therapy. More information on the VENTURE study can be found here: https://www.cera.org.au/research/venture-study.

The appointee will support interpretation of genomic data generated in a diagnostic context for rare ocular diseases. The position will involve review of genomic findings to assist in the curation of the biomedical literature and use of basic bioinformatics skills and resources. They will work with the project lead, Dr Ceecee Britten-Jones, to support project development and data management, and closely with our clinical research teams at both the University of Melbourne and the Centre for Eye Research Australia.

This new role will contribute to the University of Melbourne's partnership with international genomics company Illumina. The University has entered a long-term strategic partnership with
Illumina, a global leader in genomics technology, to jointly foster the development of world-leading genomics research and innovation activities and to accelerate the translation of these innovations through to adoption in the healthcare system. Together, Illumina and the University have established The Advanced Genomics Collaboration (TAGC), a Victorian Government funded initiative which aims to support a pipeline of commercially focused genomics innovation projects to deliver a cluster of new high growth, high-tech start-ups within the Melbourne Biomedical Precinct.

This is a flexible role, with the possibility of working from the University of Melbourne Parkville campus and/or working from home.

1. Key Responsibilities

1.1 DATA INTERPRETATION AND REPORTING

- Analyse and interpret genomic data (next generation sequencing), including the assessment, interpretation, and classification of DNA variants in individuals with rare eye diseases, and developing research genomic reports.
- Perform periodic variant re-analysis and assessment for re-classification.
- Manage communications and interpretive discussions with clinical genetics, genetic counsellors, and other relevant third party services.
- Ensure provision of high-quality data generation within the project in assessing results from genomic sequencing
- Assist Dr Britten-Jones, A/Prof Ayton and team with requirements for grant proposal and publications.

1.2 DATA AND RESEARCH PROJECT MANAGEMENT

- Oversee the collection, storage, and management of research data, including genomic data, and maintain study databases and workflows.
- Support excellent data integrity and data management processes within the project and the wider research team.
- Assist in the education of the clinical team in basic genomic concepts, and work as a liaison with external partners in this field
- Contribute to troubleshooting and developing various aspects of the project.

2. Selection Criteria

2.1 ESSENTIAL

- A strong scientific background in biology, molecular mechanisms, functional studies of genes and proteins, or training in human genetics
- Experience with variant assessment, curation, classification, and assignment of pathogenicity
- Has a good understanding of interpretation of germline next generation sequencing data, including the use of relevant population and clinical variant databases
Ability to critically read, understand, and interpret data from scientific publications

- Broad experience with utilising established databases
- Experience with coordinating research and/or project management
- Demonstrated ability to work both independently and collaboratively within a multidisciplinary team
- Strong interpersonal skills, including both written and verbal communication skills
- Demonstrated ability to learn new techniques and procedures
- High level of organisation and excellent attention to detail

### 2.2 DESIRABLE

- A tertiary qualification in life sciences (Molecular Biology/Genetics), genomics, clinical bioinformatics, or related area
- Experience in genomic research and handling genomics and multi-omics data
- Familiarity with clinical research, clinical trials, and research guidelines and regulations
- Knowledge and experience with standard bioinformatic languages including Python, R, Linux, or related software
- Experience with data analysis, manuscript preparation, and scientific presentations

### 2.3 OTHER JOB RELATED INFORMATION

- To be eligible for this position you must be an Australian or New Zealand citizen, permanent resident or hold a valid work permit or visa
- Occasional work out of ordinary hours, travel, etc
- You will be required to consent to a police check (as the physical location of this role is within a public hospital). Please note that people with criminal records are not automatically prevented from applying for the position and each application will be considered on its merits.

### 3. Equal Opportunity, Diversity and Inclusion

The University is an equal opportunity employer and is committed to providing a workplace free from all forms of unlawful discrimination, harassment, bullying, vilification and victimisation. The University makes decisions on employment, promotion, and reward on the basis of merit.

The University is committed to all aspects of equal opportunity, diversity and inclusion in the workplace and to providing all staff, students, contractors, honorary appointees, volunteers and visitors with a safe, respectful and rewarding environment free from all forms of unlawful discrimination, harassment, vilification and victimisation. This commitment is set out in the Advancing Melbourne strategy that addresses diversity and inclusion, equal employment opportunity, discrimination, sexual harassment, bullying and appropriate workplace behaviour. All staff are required to comply with all University policies.
The University values diversity because we recognise that the differences in our people’s age, race, ethnicity, culture, gender, nationality, sexual orientation, physical ability and background bring richness to our work environment. Consequently, the People Strategy sets out the strategic aim to drive diversity and inclusion across the University to create an environment where the compounding benefits of a diverse workforce are recognised as vital in our continuous desire to strive for excellence and reach the targets of Advancing Melbourne.

4. Occupational Health and Safety (OHS)

All staff are required to take reasonable care for their own health and safety and that of other personnel who may be affected by their conduct.

OHS responsibilities applicable to positions are published at:

https://safety.unimelb.edu.au/people/community/responsibilities-of-personnel

These include general staff responsibilities and those additional responsibilities that apply for Managers and Supervisors and other Personnel.

5. Other Information

5.1 ORGANISATION UNIT


The Department of Optometry and Vision Sciences educates future optometrists, performs internationally recognised vision science research, and contributes widely to the advancement of optometry as an essential health care discipline.

5.2 BUDGET DIVISION

https://mdhs.unimelb.edu.au/#about

The Faculty of Medicine, Dentistry and Health Sciences is an extremely diverse faculty comprised of six schools, and a number of departments and centres. Our Faculty is renowned for global leadership in health research, policy and practice.

5.3 THE UNIVERSITY OF MELBOURNE

Established in 1853, the University of Melbourne is a leading international university with a tradition of excellence in teaching and research. The main campus in Parkville is recognised as the hub of Australia’s premier knowledge precinct comprising eight hospitals, many leading research institutes and a wide-range of knowledge-based industries. With outstanding performance in international rankings, the University is at the forefront of higher education in the Asia-Pacific region and the world.

The University employs people of outstanding calibre and offers a unique environment where staff are valued and rewarded.

Further information about working at The University of Melbourne is available at http://about.unimelb.edu.au/careers
5.4 ADVANCING MELBOURNE

The University’s strategic direction is grounded in its purpose. While its expression may change, our purpose is enduring: to benefit society through the transformative impact of education and research. Together, the vision and purpose inform the focus and scale of our aspirations for the coming decade.

Advancing Melbourne reflects the University’s commitment to its people, its place, and its partners. Our aspiration for 2030 is to be known as a world-leading and globally connected Australian university, with our students at the heart of everything we do.

- We will offer students a distinctive and outstanding education and experience, preparing them for success as leaders, change agents and global citizens.
- We will be recognised locally and globally for our leadership on matters of national and global importance, through outstanding research and scholarship and a commitment to collaboration.
- We will be empowered by our sense of place and connections with communities. We will take opportunities to advance both the University and the City of Melbourne in close collaboration and synergy.
- We will deliver this through building a brilliant, diverse and vibrant University community, with strong connections to those we serve.

The means for achieving these goals include the development of the University of Melbourne’s academic and professional staff and the capabilities needed to support a modern, world-class university. Those means require a commitment to ongoing financial sustainability and an ambitious infrastructure program which will reshape the campus and our contribution to the communities we engage with. This strategy, and the priorities proposed, is centred around five intersecting themes; place, community, education, discovery and global.

5.5 GOVERNANCE

The Vice Chancellor is the Chief Executive Officer of the University and responsible to Council for the good management of the University.

Comprehensive information about the University of Melbourne and its governance structure is available at https://about.unimelb.edu.au/strategy/governance