



## POSITION DESCRIPTION

Department of Medicine, Austin Health  
Melbourne Medical School  
Faculty of Medicine, Dentistry & Health Sciences

### Research Associate (Bioinformatics)

POSITION NO	0055902
CLASSIFICATION	Research Associate Grade 1 Level A
SALARY	\$75,289 – \$102,163 (pro rata)
SUPERANNUATION	Employer contribution of 17%
EMPLOYMENT TYPE	Full-Time (fixed term) position (1.0 FTE)
OTHER BENEFITS	<a href="http://hr.unimelb.edu.au/careers/working/benefits">http://hr.unimelb.edu.au/careers/working/benefits</a>
HOW TO APPLY	NA
CONTACT FOR ENQUIRIES ONLY	Professor Jeffrey Zajac Tel +61 3 9035 7030 Email <a href="mailto:j.zajac@unimelb.edu.au">j.zajac@unimelb.edu.au</a>  <i>Please do not send your application to this contact</i>

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

## ***Position Summary***

The Epilepsy Research Centre is based in the Department of Medicine at the Austin Hospital and Melbourne Brain Centre, and has long-standing collaborations with the Walter and Eliza Hall Institute (WEHI), and Murdoch Children's Research Institute (MCRI) / Royal Children's Hospital (RCH). Together, these institutions are running three research programs underpinning this new position: (i) 2020 Medical Research Future Fund (MRFF) - Genomics Health Futures Mission Stream 1 Targeted Call for Research APP2007707, 'Precision Diagnosis for the Remaining 50% of Unsolved Developmental and Epileptic Encephalopathies', (ii) 2020 Rare Cancers Rare Diseases and Unmet Need – General Clinical Trial APP2006631, 'Targeted Therapies for Vascular Malformations', and (iii) National Health and Medical Research Council (NHMRC) Ideas Grant APP2012287, 'Using Cerebrospinal Fluid Liquid Biopsy for Detection of Mosaic Brain Mutations'. These studies are being led by Associate Professor Michael Hildebrand, Professor Samuel Berkovic and Professor Ingrid Scheffer at University of Melbourne, Professor Melanie Bahlo and Dr Mark Bennett at WEHI, and Professor Tony Penington at MCRI/RCH.

These programs are broadly aimed at increasing diagnostic yield for patients with rare genetic epilepsies, vascular and brain malformations by detecting germline and somatic variants present in blood or lesional tissue or using alternative routes to diagnosis (e.g., CSF liquid biopsy). They rely on sensitive, high depth sequencing and high level genome mapping and variant calling analysis pipelines including application of the latest algorithms for somatic variant calling and classification.

We are seeking a highly motivated discovery scientist with a Masters or PhD in bioinformatics, molecular biology, mathematics or statistics, experience in bioinformatics, and preferably skills in molecular genetic analyses including next generation sequencing and rare variant calling. The full-time position will be based equally across (i) Translational Neurogenetics Laboratory in the Epilepsy Research Centre, Melbourne Brain Centre (Austin Campus), University of Melbourne, and (ii) the Population Health and Immunity Division Laboratory at WEHI. Associate Professor Michael Hildebrand (University of Melbourne), Professor Melanie Bahlo (WEHI) and Dr Mark Bennett (WEHI) will be primary supervisors, and Professor Samuel Berkovic (University of Melbourne), Professor Ingrid Scheffer (University of Melbourne) and Professor Tony Penington (MCRI/RCH) will be co-supervisors.

The successful candidate will complete a multidisciplinary project examining genetic contributions to rare genetic disorders using the latest genetics techniques and analysis approaches uniquely designed to leverage expertise across top genetics, bioinformatics and medical research departments or institutes. The appointee will undertake bioinformatics genomic and transcriptomic variant analyses using a range of established analysis pipelines and algorithms to achieve genetic diagnosis in unsolved patients with (i) developmental and epileptic encephalopathies for the MRFF GHFM project, (ii) vascular malformations for the MRFF Clinical Trial project, and (iii) brain malformations and lesional focal epilepsy for the Ideas Grant project. The appointee will be required to keep abreast of the scientific literature in the advancement of their work. They must keep adequate records of all completed works as well as being able to present this work when required. The appointee will also have some responsibility to assist undergraduate, Honours, Masters and PhD students working on these projects.

This position reports to A/Professor Michael Hildebrand, Professor Melanie Bahlo, Dr Mark Bennett, Professor Samuel Berkovic, Professor Ingrid Scheffer and Professor Tony Penington. Local and overseas applicants are encouraged to apply.

## **1. Key Responsibilities**

### **1.1 RESEARCH**

- The conduct of gene discovery research into epilepsy, vascular and brain malformations
- Analysis of data using next-generation sequencing analysis tools
- Development and maintenance of data analysis pipelines
- Data analysis of results using computer based statistical and graphics programs
- Understanding and interpretation of techniques and results
- Production of publications arising from scholarship and research

### **1.2 SERVICE AND LEADERSHIP**

- Involvement in professional activity, including preparation and presentation of data and findings to local seminars and meetings, and participation in conferences and meetings of professional societies (subject to availability of funds)
- To actively participate in research group laboratory meetings and departmental meetings and seminars.
- Occupational Health and Safety (OH&S) and Environmental Health and Safety (EH&S) responsibilities as outlined in section 5

## **2. Selection Criteria**

### **2.1 ESSENTIAL**

- Masters or PhD in bioinformatics, computational or molecular biology, mathematics, statistics, computer science or a similar quantitative field
- BSc or equivalent in biological sciences, biomedical sciences, mathematics, statistics, computer science or a similar quantitative field
- Expertise in computer programming with R, Python, Perl, or similar, and post analysis interpretation skills, including generation of reports such as in R Markdown including experience or willingness to work on high performance supercomputing platforms
- Training in analysis of next-generation sequencing and genetic data
- A demonstrated record of achievement in research, evidenced by coursework or publications arising from PhD, Masters, Honours or other research positions
- Well-developed interpersonal skills including demonstrated capacity to work as both a team member and independently, and excellence in written and oral communication
- Excellent organisational skills and the ability to undertake a range of tasks concurrently

- Excellent communication skills and a demonstrated ability to present oral summaries of research to various audiences

## 2.2 DESIRABLE

- BSc (Hons) or equivalent in biological sciences, biomedical sciences, mathematics or statistics
- Vocation, study, interest or background in genetics or molecular biology, particularly genetic diseases

## 3. *Special Requirements*

- You need to be comfortable with, and have the ability to work on data from human subjects

## 4. *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 University's People Strategy 2015-2020 and policies that address 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 Growing Esteem.

## 5. *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:

<http://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.

## 6. Other Information

### 6.1 DEPARTMENT OF MEDICINE, AUSTIN HEALTH

<http://www.austinmedicine.unimelb.edu.au/>

The Department of Medicine, Austin Health is a large department in the School of Medicine and is committed to high standards of teaching, research and clinical care. The Department has major programs in basic and applied research, has clinical responsibilities at Austin Hospital and Heidelberg Repatriation and contributes significantly to the undergraduate teaching program for medical students. The research base is broad with significant funding from NH&MRC and other competitive grant schemes. There are 70 academic, technical and administrative staff, and 55 students who are enrolled to pursue higher degrees from BSc (Hons), MSc, PhD and MD.

### 6.2 EPILEPSY RESEARCH CENTRE

<http://www.austinmedicine.unimelb.edu.au/research/epilepsy/index.html>

<http://www.epilepsyresearch.org.au/>

The Epilepsy Research Centre is a large cohesive research group within the Department of Medicine with a world-leading international profile. The Centre is currently comprised of more than 20 individuals including neurologists, research fellows, PhD students and research assistants. Our important Australia-wide and international study investigating the genetic causes of epilepsy has been ongoing for more than 20 years. The molecular genetic work is performed primarily in a laboratory located at the Melbourne Brain Centre, and by collaborating laboratories in Australia and internationally. The studies have been responsible for the identification of several epilepsy syndromes and the identification of many epilepsy genes. The research group is situated at the new Melbourne Brain Centre building at the Austin campus of Austin Health, Heidelberg.

### 6.3 MELBOURNE MEDICAL SCHOOL

<http://www.medicine.unimelb.edu.au/>

The Melbourne Medical School is organised into nine Academic Centres comprising more than 20 departments and five general clinical schools. It also supports the Medical Education Unit.

The School is closely associated with major teaching and other hospitals, (<http://mdhs.unimelb.edu.au/our-health-network>) general practices and community health centres in metropolitan, regional and rural Victoria which provides clinical education for medical students

Through its Academic Centres and Departments the School also maintains a close relationship with major research institutes. (<http://mdhs.unimelb.edu.au/research-institutes>).

### 6.4 FACULTY OF MEDICINE DENTISTRY & HEALTH SCIENCES

The Faculty of Medicine, Dentistry & Health Sciences has an enviable research record and is the University of Melbourne's largest faculty in terms of management of financial resources, employment of academic and professional staff, teaching of undergraduate and postgraduate (including research higher degree) students and the conduct of basic

and applied research. The Faculty's 2013 gross revenue was in excess of \$550M. Approximately 40% of this income relates to research activities.

The Faculty has a student teaching load in excess of 7,500 equivalent full-time students including more than 1,000 research higher degree students. The Faculty has approximately 2,200 staff comprising 700 professional staff and 1,500 research and teaching staff.

The Faculty has appointed Australia's first Associate Dean (Indigenous Development) to lead the development and implementation of the Faculty's Reconciliation Action Plan (RAP), which will be aligned with the broader University – wide plan. To enable the Faculty to improve its Indigenous expertise knowledge base, the Faculty's RAP will address Indigenous employment, Indigenous student recruitment and retention, Indigenous cultural recognition and building partnerships with the Indigenous community as key areas of development.

Further information about the Faculty is available at <http://www.mdhs.unimelb.edu.au/>

## 6.5 THE UNIVERSITY OF MELBOURNE

The University of Melbourne is a leading international university with a tradition of excellence in teaching and research. With outstanding performance in international rankings, Melbourne is at the forefront of higher education in the Asia-Pacific region and the world. The University of Melbourne is consistently ranked among the world's top universities. Further information about our reputation and global ranking is available at [www.futurestudents.unimelb.edu.au/explore/about/reputation-rankings](http://www.futurestudents.unimelb.edu.au/explore/about/reputation-rankings)

Established in 1853, shortly after the founding of Melbourne, the University is located just a few minutes from the centre of this global city. The main Parkville campus 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.

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 [hr.unimelb.edu.au/careers](http://hr.unimelb.edu.au/careers).

## 6.6 GROWING ESTEEM, THE MELBOURNE CURRICULUM AND RESEARCH AT MELBOURNE: ENSURING EXCELLENCE AND IMPACT TO 2025

- ▶ Growing Esteem describes Melbourne's strategy to achieve its aspiration to be a public-spirited and internationally-engaged institution, highly regarded for making distinctive contributions to society in research and research training, learning and teaching, and engagement. [www.growingesteem.unimelb.edu.au](http://www.growingesteem.unimelb.edu.au)
- ▶ The University is at the forefront of Australia's changing higher education system and offers a distinctive model of education known collectively as the Melbourne Curriculum. The new educational model, designed for an outstanding experience for all students, is based on six broad undergraduate programs followed by a graduate professional degree, research higher degree or entry directly into employment. The emphasis on academic breadth as well as disciplinary depth in the new degrees ensures that graduates will have the capacity to succeed in a world where knowledge boundaries are shifting and reforming to create new frontiers and challenges. In moving to the new model, the

University is also aligning itself with the best of emerging European and Asian practice and well-established North American traditions.

- ▶ The University's global aspirations seek to make significant contributions to major social, economic and environmental challenges. Accordingly, the University's research strategy *Research at Melbourne: Ensuring Excellence and Impact to 2025* aspires to a significant advancement in the excellence and impact of its research outputs.

<http://www.unimelb.edu.au/research/research-strategy.html>

The strategy recognises that as a public-spirited, research-intensive institution of the future, the University must strive to make a tangible impact in Australia and the world, working across disciplinary and sectoral boundaries and building deeper and more substantive engagement with industry, collaborators and partners. While cultivating the fundamental enabling disciplines through investigator-driven research, the University has adopted three grand challenges aspiring to solve some of the most difficult problems facing our world in the next century. These Grand Challenges include:

**Understanding our place and purpose** – The place and purpose grand challenge centres on understanding all aspects of our national identity, with a focus on Australia's 'place' in the Asia-Pacific region and the world, and on our 'purpose' or mission to improve all dimensions of the human condition through our research.

**Fostering health and wellbeing** – The health and wellbeing grand challenge focuses on building the scale and breadth of our capabilities in population and global health; on harnessing our contribution to the 'convergence revolution' of biomedical and health research, bringing together the life sciences, engineering and the physical sciences; and on addressing the physical, mental and social aspects of wellbeing by looking beyond the traditional boundaries of biomedicine.

**Supporting sustainability and resilience** – The sustainability and resilience grand challenge addresses the critical issues of climate change, water and food security, sustainable energy and designing resilient cities and regions. In addition to the technical aspects, this grand challenge considers the physical and social functioning of cities, connecting physical phenomena with lessons from our past, and the implications of the technical solutions for economies, living patterns and behaviours.

Essential to tackling these challenges, an outstanding faculty, high performing students, wide collaboration including internationally and deep partnerships with external parties form central components of *Research at Melbourne: Ensuring Excellence and Impact to 2025*.

## 6.7 EQUITY AND DIVERSITY

Another key priority for the University is access and equity. The University of Melbourne is strongly committed to an admissions policy that takes the best students, regardless of financial and other disadvantage. An Access, Equity and Diversity Policy Statement, included in the University Plan, reflects this priority.

The University is committed to equal opportunity in education, employment and welfare for staff and students. Students are selected on merit and staff are selected and promoted on merit.

## 6.8 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 [www.unimelb.edu.au](http://www.unimelb.edu.au).