



creating possible

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## Position Description

<b>Position Title:</b>	Bioinformatician
<b>Salary Range:</b>	MCRI Research Salaries - Level A Step 6 to Level B Step 2
<b>Reporting Manager:</b>	Dr Simon Sadedin
<b>Direct Reports:</b>	None
<b>Home Group:</b>	Bioinformatic Methods

### Who are we?

The Murdoch Children's Research Institute (MCRI) is home to significant scientific discoveries. We believe there is an answer, a cure or a better treatment for every childhood condition – and we're determined to find it.

We are a diverse team of world-leading researchers, doctors, engineers, and hardworking professionals in corporate and scientific services from all corners of the world with one shared goal – to transform child health worldwide.

Our strength lies in our partnership and co-location with The Royal Children's Hospital and the University of Melbourne – the Melbourne Children's Campus. This rare model amplifies opportunities to quickly translate research into clinical care.

At MCRI, you'll also find our subsidiary organisation, the Victorian Clinical Genetics Services (VCGS), a specialist childhood, prenatal and adult genetics service. VCGS provides an integrated genetic consultation, counselling, testing and diagnostic support service to children, adults, families and prospective parents.

Together, we share a powerful vision: re-imagine the future of child health.

### What is it like to work for us?

We are committed to ensuring a positive working environment that values all backgrounds and experiences. We cultivate an inclusive culture that is underpinned by equal opportunity for all and a culture based on respect, consideration and dignity. We are also committed to developing our people and fostering an environment where learning and development is central to our staff reaching their full potential.

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## About the Bioinformatics Methods Team

The Bioinformatics Methods team at MCRI is a group of software engineers, bioinformaticians, data scientists and genomics experts who work together to make state of the art methods for genomics analysis accessible to MCRI researchers. Our goal is to enable researchers to extract the maximum possible value from their genomic data. We specialise in working at the forefront of genomics technology and data science to ensure that the most advanced methods are made available and usable to researchers at MCRI. We collaborate with research groups within Australia and around the world to drive the frontiers of what is possible.

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The Bioinformatics Methods team is directly linked to the Victorian Clinical Genetics Services (VCGS), one of Australia's leading advanced genetic testing laboratories. Sitting at the nexus of research translation within the Royal Children's Hospital campus, the team offers a unique opportunity to put data science to work in helping people suffering from debilitating genetic disorders.

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### Position Overview

The Bioinformatician will work with the Group Head and the Interpretation Lead to develop and maintain state of the art solutions to enable MCRI researchers to analyse and interpret genomic data.

Positioned within MCRI's "genomics informatics hub", the activities of the bioinformatician will include evaluation, development and customisation of bioinformatic tools and methods, connecting these into workflows and pipeline scripts, and integration of the pipelines into MCRI's platform for genomic data storage and analysis. Key projects that the bioinformatician will participate in will include MCRI's RDNOW initiative to find answers for undiagnosed rare disease patients and engaging with the Centre for Population Genomics to enabling population scale genomics. The bioinformatician will also participate in developing and refining the Bioinformatics Methods group's flagship pipelines and applications, enabling interpretation of mitochondrial variants and whole genome and exome copy number variants.

Your role will be to help identify and create these tools by evaluating the landscape of available options, reviewing best practices both within and external to MCRI and where appropriate, leverage your own skills and expertise to develop and improve upon these to create optimal solutions for MCRI research use. As part of this work, you will be responsible for integrating the tools identified into MCRI's automated pipeline infrastructure to enable high quality automated analyses through our genomic data platform.

As part of your role, you will have opportunity to participate in many of MCRI's grant funded projects, including RDNOW (solving undiagnosed rare disease cases), the National Reanalysis Infrastructure Project (building automated analysis pipelines using machine learning to reprocess undiagnosed patients), and engaging with MCRI's population scale initiatives through the Centre for Population Genomics led by Daniel MacArthur and Generation Victoria (GenV).

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### Key Accountabilities

- Identification and evaluation of bioinformatic tools and methods for genomic analysis.
  - Maintenance and development of pipelines for genomic analysis.
  - Integration of pipelines into genomic data platforms.
  - Reviewing research articles and educating team and other institute staff on genomic data analysis methods.
  - Participating in publication of research articles.
  - Working closely with the Group Head and Interpretation Lead to foster a strong culture of ownership and teamwork to deliver high quality solutions that add value for stakeholders, our team and the organisation.
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### Selection Criteria

- A tertiary qualification (Masters level or higher) in Bioinformatics or a similar field, or substantial equivalent experience.
  - Knowledge of one or more data analysis and software programming languages relevant to bioinformatics (R, Python, Perl, JVM languages such as Scala, Java or Groovy).
  - Understanding of fundamentals of rare disease genetics, including Mendelian inheritance patterns, common features of pathogenic variants and the underlying biology of how these result in human disease.
  - Strong knowledge of the Linux command line, including use of HPC and cloud environments for managing data and executing genomic workflows.
  - Familiarity with modern software practises such as version control, automated testing, DevOps, etc.
  - Excellent time management, organisational and analytical problem-solving skills.
  - Demonstrated excellent verbal and written communication skills.
  - Demonstrated capacity to work independently and collaboratively in a team environment.
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### Conditions of Employment

- Working with Children & National Police Clearance (if appointed) in compliance with the Victorian Governments Child Safety Standards.
  - The right to reside and work in Australia and you meeting any applicable visa conditions.
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## Health, Safety & Wellbeing

- We are committed to providing and maintaining a working environment which protects the health, safety and wellbeing of our people, partners and the community.
- Employees conducting duties on behalf of MCRI are expected to meet the environment, health and wellbeing requirements and responsibilities specifically required for the role.
- We are committed to supporting children in their right to be safe and adhere to the responsibilities we have to ensure their protection and safety as per the Child Safety Standards Policy.
- Specified positions may be subject to medical review to ensure that the inherent requirements of the role can be undertaken safely.

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*As MCRI evolves to meet its changing strategic and operational needs and objectives, so will the roles required of its employees. As such, this document is not intended to represent the position which the occupant will perform in perpetuity. This position description is intended to provide an overall view of the incumbent's role as at the date of this statement.*