

Position Description

Position Title:	Software Engineer (Rare Disease)
Salary Range:	Individual Contract
Reporting Manager:	Cas Simons (Rare Disease Program Lead)
Direct Reports:	None
Home Group:	Centre for Population Genomics

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 translate research into clinical care quickly.

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 the Centre for Population Genomics?

The Centre for Population Genomics (CPG) is a collaborative initiative of the Garvan Institute of Medical Research (Garvan) in Sydney and Murdoch Children's Research Institute (MCRI) in Melbourne. The Centre is directed by Daniel MacArthur who previously led a team at the Broad Institute of MIT and Harvard in Boston that was responsible for the development and open release of the Genome Aggregation Database (gnomAD), a collection of genetic data from over 140,000 individuals that has become one of the most widely-used reference databases in human genetics.

The Centre is focussing on developing cutting-edge tools and resources to facilitate the conversion of genomic data into improved diagnosis and treatment. Ultimately, the Centre will generate and manage the largest sets of genomic and clinical data ever assembled in Australia and apply these to solve various scientific and medical problems.

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 an 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 are central to our staff reaching their full potential.

Position Overview

The Centre for Population Genomics is seeking a full stack software engineer to join our Rare Disease Team. In this role, you will be responsible for developing and supporting web-based tools, such as <u>seqr</u>, to provide intuitive access to complex genomic datasets. You will be embedded within our team of computational biologists and genomic scientists and work in collaboration with clinicians and researchers across Australia. By working closely with these domain experts, you will play a crucial role in developing analysis methods, tools, interfaces, and visualizations that will directly contribute to the discovery and diagnosis of rare genetic disorders.

You will collaborate closely with the Centre's software engineering team to ensure that all solutions are secure, resilient, and integrated with our broader engineering infrastructure. Additionally, all software developed by the Centre is open-source and designed using best practices to ensure widespread use by the scientific community and rapid translation into impact on human biology and medicine.

This role offers a unique opportunity to work in an environment that combines the intellectual freedom and collaboration of academia with rigorous, open software development practices. You will also have the chance to collaborate with partners such as the Broad Institute and Microsoft Research to ensure that our solutions prove useful for the wider genomics community.

You will be reporting to the Rare Disease Program Lead, Cas Simons. Close collaboration with other members of the Centre's software team, genomic analysis teams, and computational groups at Garvan and MCRI will also be essential to the success of this role.

Prospective candidates will ideally be located at the Garvan Institute in Sydney or the MCRI in Melbourne. While CPG champions a remote-first work model, we also value office interactions and quarterly face-to-face events. The perfect fit will be someone who flourishes in a virtual workspace and balances remote work with the office.

Key Accountabilities

- Work closely with genomic analysts and computational biologists to translate their requirements into software solutions that present and summarise genomic data to assist rapid interpretation and enable patient diagnosis.
- Work with other engineers to ensure our systems are scalable and performant
- Apply user-centred design principles to create intuitive and accessible interfaces for diverse user groups, including clinicians and researchers
- Implement, debug, optimise, deploy, monitor, and maintain your solutions, picking the most appropriate technologies for the task
- Ensure adherence to best practices in security, particularly when handling sensitive genomic data
- Create comprehensive documentation for developed tools and systems to facilitate ease of use and onboarding for new users and team members
- Foster a culture of knowledge sharing by contributing to internal wikis, conducting workshops, and providing mentorship to junior staff
- All staff are required to comply with MCRI's workplace policies (as amended from time to time) and especially MCRI's Work Health & Safety (WHS) Policy, including working in accordance with the WHS management system at all times, as safety is MCRI's number one priority

Selection Criteria

- A BCST / BIT / BE degree in computer science, software engineering, information technology, bioinformatics engineering, biomedical engineering or a related field
- Solid experience with Python and Typescript/Javascript, including a deep understanding of building and testing performant, decoupled, and maintainable code
- Proven experience in developing or supporting web applications using the Django/React web stack or similar modern toolchains, demonstrating proficiency in leveraging the capabilities of these technologies to deliver robust and user-friendly applications
- Proficiency in working with databases such as Postgres, MariaDB, and MySQL, including expertise in query optimisation techniques, such as indexes and joins
- An appetite to learn the principles of genetics and genomic analysis. Existing knowledge or experience in genomics or bioinformatics would be advantageous but is not essential
- Ability to prioritize across multiple simultaneous projects and keep stakeholders in the loop
- Keen interest in open source and desire to learn new technologies and understand / enhance large existing code bases
- A pragmatic approach to software driven by project needs and timelines rather than a predetermined commitment to specific technical stacks
- Proven ability to work effectively in a fully remote team, including exceptional written and verbal communication skills

Conditions of Employment

- Working with Children & National Police Clearance (if appointed) in compliance with the Victorian Government's Child Safety Standards
- The right to reside and work in Australia and you meeting any applicable visa conditions

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

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.