



creating
possible

Position Description

Position Title:	Genomic Curation Research Specialist
Salary Range:	To be advised based on experience
Reporting Manager:	Head of Clinical Bioinformatics
Direct Reports:	None
Home Group:	Clinical Bioinformatics

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.

About The Project - A national large scale automated reanalysis program to increase rare disease diagnosis

Genomic testing technologies have transformed rare disease diagnosis. Unfortunately, more than half of those undergoing genomic testing currently remain without a diagnosis.

Reanalysis of existing data has repeatedly been shown to deliver additional diagnostic yields in the range of 10-15%. Despite strong support at professional and health policy levels, translation into practice is currently limited by reliance on a heavily manual process. Coupled with shortages of a skilled genomics workforce and limited funding, most existing datasets benefit from little if any reanalysis, creating an inequity in diagnostic, and by extension, health outcomes.

This project will fill this gap by developing, applying and evaluating a national program for automated, systematic reanalysis of genomic data to deliver improved diagnostic outcomes for rare disease patients and their relatives with the aim of providing answers to hundreds of Australian families.

About The Team

To deliver this project, we are building a multidisciplinary team that combines clinical and laboratory genomics, population genomics, health economics, bioethics, implementation science, software design and bioinformatics expertise. This role will be based at the MCRI and work closely with team members at Victorian Clinical Genomics Services (VCGS), the Centre for Population Genomics (a joint initiative of the Garvan Institute of Medical Research and the MCRI) and other collaborators.

Position Overview

The Genomic Curation Research Specialist will play a key role in the success of the project by informing the development of automated curation logic for variants reanalysed by the system. The Genomic Curation Research Specialist will utilise their expert knowledge of clinical variant curation to advise the software teams and machine learning scientists regarding initial approach, as well as ongoing requirements as the system evolves. Through extensive detailed parallel curation of variants, they will assess the outcomes of annotation, prioritisation and filtering performed by the system and feedback suggested improvements and benchmarking results. The role will also involve identifying, evaluating and cleaning appropriate source databases to input into the relevant software and machine learning approaches. Elements requiring evaluation include the clinical validity of decisions made, usability of the system in terms of translation of the results to real clinical downstream use and understanding the clinical utility of results that are delivered. In addition to assisting development of in-house tools, the Curation Research Specialist will also evaluate third party options including both commercial and existing open source/research tools. This may involve direct negotiation with third parties to establish appropriate arrangements and facilitating execution of any agreed mutual outcomes (such as evaluation and benchmarking).

Due to the intimate connection to the software development team, it is crucial that the Genomic Curation Research Specialist is familiar and comfortable with software development processes and ideally will have some direct experience or skills in this area. Similarly, the Curation Research Specialist will interact heavily with machine learning experts who will assist to develop and deploy state of the art AI approaches to maximise the accuracy and diagnostic yield from the project.

Finally, they will use their understanding of genomic testing to help to define the overall flow of the reanalysis process, ensuring that the overall workflow meshes with the needs and expectations of downstream clinical users, accounting for possibly different pathways for different kinds of clinical results.

Key Accountabilities

- Assessing outcome of annotation, prioritisation and filtering tools for genomic variants.
- Evaluating & providing feedback about outcomes from automated interpretation tools.
- Contributing to/advising/guiding development of possible automation/machine learning approaches.
- Contributing to identifying/selecting/cleaning training data sets for use in machine learning and benchmarking of approaches.
- Evaluating the validity, accuracy, usability and clinical utility of the automated variant assessment approaches.
- Validating selected approaches for clinical use.
- Ensuring approaches are well designed for interfacing and integration to clinical laboratory workflows and staff.
- Identifying existing commercial and research tools for variant annotation, prioritisation and filtering.
- Negotiating with stakeholders for third party tools to enable evaluation and possible use in our project.
- Ensuring software developer team and machine learning experts have clear, actionable feedback regarding success of implementations and improvements to be made.
- Is aware of, and adheres to, MCRI policy on Intellectual Property/Material Transfer Agreements/Contracts/Clinical and Public Health Outcomes.

Selection Criteria

Essential

- Excellent understanding of clinical variant interpretation/prioritisation and curation processes.
- Understanding of software development principles and algorithm development.
- Methodical and target/outcome focussed with the ability to deliver on time and budget.
- Familiarity with clinical laboratory processes, NATA/NPAAC accreditation, legal requirements for clinical testing.
- Understanding of research frameworks for use of genomic data (eg: privacy, ethics, consent).
- Proven high level experience in communicating, navigating and negotiating with multiple organisations, stakeholders and professional groups from different fields and backgrounds.

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- Flexible and adaptable approach with the ability to quickly grasp and use new concepts.

Desirable

- Familiarity with relevant concepts of machine learning.
 - Experience in a computing language (e.g. Python) and Linux/Unix environments.
 - Demonstrated experience and ability to lead and mentor employees in order to deliver project outcomes.
 - Demonstrates independent judgement, initiative and influencing skills.
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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.