

## POSITION DESCRIPTION

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<b>Position Title:</b>	Computational Biologist (Genomic Medicine)
<b>Theme or Workgroup:</b>	Genomic Medicine Lab, Centre for Population Genomics
<b>Reports to (Title):</b>	Owen Siggs (Head, Genomic Medicine Lab)
<b>Job Classification &amp; Grade:</b>	SRO1 – SRO3
<b>Approved by (Title):</b>	Owen Siggs (Head, Genomic Medicine Lab)
<b>Date:</b>	December 2022

### INTRODUCTION

As part of the **Centre for Population Genomics** (CPG, Directed by Daniel MacArthur), and with the generous and long-term support of Snow Medical, the **Genomic Medicine Lab** is developing an ambitious program to understand the clinical impact of genetic variation. Our focus is to use large-scale population, clinical, and cellular genomics to understand, predict, treat, and prevent disease. A key interest is understanding the impact of somatic variation and clonal haematopoiesis in diseases of inflammation and ageing, and the interaction of somatic variation with other genetic and non-genetic risk factors.

### THE ROLE

An integral part of our program is to understand the impact of inherited and acquired genetic variation in disease risk and progression, through the analysis of large-scale genomic and clinical datasets. This will include the design and implementation of scalable somatic variant calling and statistical analysis pipelines across large-scale clinical and genomic datasets (both short- and long-read WGS, as well as single-cell genomic datasets), deployed across cloud-based computational environments.

The successful applicant will have opportunities to lead and publish projects in translational genomics, including national and international collaborative efforts. This role also comes with the benefit of membership of the Centre for Population Genomics (CPG): a joint initiative of the Garvan Institute of Medical Research and the Murdoch Children's Research Institute (MCRI), with a mission to collect and analyse genomic data at a transformative scale. In this role, you will have ample opportunity to work closely with professional and academic CPG colleagues in analysis and methods development, software engineering, and project management, and with the large and unique genomic datasets and analysis tools they curate. We also have a strong clinical focus with an extensive network of clinical collaborators, maximising opportunities for the candidate to have real-world clinical impact.

### ROLE FLEXIBILITY

Members of the Genomic Medicine Lab (and the CPG) are geographically distributed across Australia, and we operate under a remote-friendly model. However, office space is available for use at the Garvan Institute in Sydney, with in-person events held at least twice per year. This role is therefore highly amenable to flexible work, including:

- **Hours of work flexibility** (such as flexible start and finish times); and
- **Remote working flexibility** (with regular opportunities for in-person interaction)

### ESSENTIAL DUTIES AND RESPONSIBILITIES

The Computational Biologist will be responsible for, the following key areas:

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- Working closely with colleagues to design and implement quality control pipelines for large-scale somatic and germline variant callsets and accompanying clinical data.
- Leading the development and implementation of appropriate statistical analyses.
- Actively contributing to the development of study hypotheses, the interpretation of results, and the preparation of high-impact scientific manuscripts and their presentation to internal and external audiences.
- Positively engaging with internal and external colleagues on collaborative projects, and contributing to the overall scientific strategy of the team.
- Actively contributing to a culture of scientific excellence through adherence to computational best practices, rigorous quality control, and reproducible science.
- Cost-effectively managing and integrating code repositories and datasets across cloud-based platforms and on-premise hardware.

### KEY COMMUNICATIONS

*Internal:* This role will report to Owen Siggs (Head, Genomic Medicine Lab), and will also work closely with other academic and professional staff from the Genomic Medicine Lab, CPG, and Garvan.

*External:* National and international collaborators, including researchers, clinicians, and other stakeholders.

### KNOWLEDGE, SKILLS AND QUALIFICATIONS REQUIRED

In order to be successful in this position, you will demonstrate the following key skills and attributes:

- Formal qualifications in a quantitative discipline such as Computational Biology, Statistical Genetics, Computer Science, or a related field.
- Strong proficiency in at least one coding language (Python and R preferred), and direct experience in high-performance and/or cloud computing, and collaborative coding.
- Highly engaged and motivated: able to define and execute computational strategies across a wide range of technical areas, from basic sequence quality control, to analysis of large genomic and clinical datasets.
- Excellent written communication skills, with a demonstrated ability to lead the production of high-quality academic manuscripts, grant applications, and/or technical reports.
- Excellent verbal communication skills, with a demonstrated ability to effectively represent teams internally and externally.
- Highly collaborative, with a demonstrated ability to build and maintain relationships within and between teams, and engage in complex discussions remotely.
- A problem-solving mentality, with an ability to navigate complex technical obstacles, and work effectively with other team members and colleagues to overcome them.

### DESIRABLE SKILLS

- Direct experience with cloud computing platforms (e.g. GCP, AWS) and large-scale genomic datasets (e.g. UK Biobank) is a plus.