ALIGN HDR Opportunities

Funded higher degree in research (HDR) opportunities through the ALIGN network focused on Indigenous Genomics

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Pathways to benefit for Indigenous Australians in Genomic Medicine

The potential for genomics to deliver health benefits is clear, however, the process for ensuring that Indigenous Australians realise these benefits is less certain. Understanding how to deliver equity and benefit to Indigenous Australians through genomics requires a purposeful alliance between community, health, research, government, and industry that privileges Indigenous leadership and sovereignty.

ALIGN is a national consortium, led by the Indigenous Genomics Group at Telethon Kids Institute and the Australian National University, in partnership with Aboriginal and Torres Strait Islander stakeholders, peak bodies and Communities, as well as research, clinical, industry and institutional partners from across Australia.

Indigenous governance will both underpin and lead ALIGN's work, and be instrumental in bringing forward the voices, values, and priorities of Aboriginal and Torres Strait Islander peoples, locally and nationally. ALIGN's national Indigenous Governance Council will include membership from each of the jurisdictional Indigenous governance committees as well as invited Indigenous leaders with specific knowledge and skills that can help drive our national Indigenous genetic and genomic health governance agenda.

ALIGN Opportunities

Thanks to funding provided by the Medical Research Future Fund and our generous national network of partners, ALIGN has funded higher degree research (HDR) projects available for the right candidates.

These projects span the breadth of Indigenous genomics research, from biomedical science, public health and policy, data science and law and ethics.

This document provides a brief summary of the available projects, and the contact details of the primary investigator for candidates to directly pursue the opportunities that they are interested on.

For general information, candidates can contact the ALIGN team at:

align@telethonkids.org.au

ALIGN Projects for higher degree in research candidates

Primary Care and Genomics – ATSICCHO and genetic health services' capacity building and service integration

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Investigators: Professor Alex Brown, Professor of Indigenous Genomics, Telethon Kids Institute and ANU; Ms Louise Lyons, Senior Manager, Strategy and Policy - Indigenous Genomics, Telethon Kids Institute; Mr Gregory Pratt, QAIHC; Dr Gareth Baynam, WA Health; Dr Julie McGaughran, GHQ

Location: The candidate will be hosted by the Telethon Kids Institute and ideally based in Adelaide, however candidates based in Perth, Queensland, Western Australia or Canberra will also be considered.

Project: Indigenous people underutilise genomic testing compared to non-Indigenous Australians. With genomic testing becoming standard practice, there is a growing need to strengthen referral pathways and coordinated care between primary and tertiary healthcare providers. This requires accessible, culturally safe, responsive, integrated and family-centred approaches. Aboriginal and Torres Strait Islander Community Controlled Health Organisations are central to this agenda. This project will explore the readiness, barriers and opportunities of Aboriginal and Torres Strait Islander health services nationally to implement the "Integrated Genetic Health Care" (IGHC) model developed or supported by QIMR, QAIHC, Queensland Genomics and the Queensland Govt. The project will include:

- Engaging with NACCHO and the jurisdictional peak bodies to assess readiness to review and implement IGHC model;
- Identifying barriers to and opportunities for increasing access for Aboriginal and Torres Strait Islander people to genetic and genomic health services;
- Identifying ATSICCHO staff and service capacity building requirements implement the IGHC model;
- Identifying jurisdictional modifications to the IGHC model to better suit the ATSICCHO sector and Community requirements;
- Identify or develop culturally appropriate genetic and genomic health resources for ATSICCHOs and genetic health services/clinicians;
- Develop a culturally appropriate evaluation and impact framework that can measure the success of the IGHC's implementation; and
- Develop policy and advocacy briefs to support ATSICCHO funding and resourcing requirements.

Funding: This project will be offered as a fully funded PhD scholarship + top up for 3 years.

Contact: Professor Alex Brown alex.brown@anu.edu.au

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Developing culturally-appropriate genomics data systems for a new-era of Indigenous Genomics

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Investigators: Dr Sam Buckberry, Telethon Kids Institute and ANU; Associate Professor Jimmy Breen, Telethon Kids Institute and ANU; and Professor Alex Brown Telethon Kids Institute and ANU

Location: The project will be led by ANU, and the candidate will be hosted by the Telethon Kids Institute and ideally based in Adelaide or Perth, however remote candidates will also be considered.

Project: The vast amount of genomics and personal data collected across the Australian healthcare system has an enormous potential to improve the lives of Indigenous Australians. Recent, very public, examples of data breaches at Australian Healthcare and Telecommunications companies only highlight the importance of storing identifiable information safely and securely. Given the lack of genomics information on Indigenous Australians, data breaches may incur an extreme risk to the identity of Aboriginal and Torres Strait Islanders, having implications in law enforcement and community protection. The mitigation of this risk is a major aim of ALIGN's mission, crafting culturally appropriate, ethical genomic data storage and management practices that promote precision medicine for Indigenous Australian populations.

This project will be an enormous opportunity to work across multi-institutions including ANU, CSIRO, database management start-up company Pacific Analytics and TKI's Indigenous Genomics Data Science teams, working on a variety of projects focusing on data sharing protocols and methods, cryptography and file compression and biocultural and indigenous knowledge labelling.

Funding: This project will be offered as a fully funded PhD scholarship + top up for 3 years.

Contact: Dr Sam Buckberry sam.buckberry@telethonkids.org.au

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Investigating the sex differences in Cardiovascular and Type-2 Diabetes disease risk in Aboriginal and Torres Strait Islanders

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Investigators: Associate Professor Jimmy Breen, Telethon Kids Institute and ANU; Dr Stevie Pederson, Telethon Kids Institute and ANU; and Dr Liza Kretzschmar Telethon Kids Institute and ANU

Location: The project will be led by ANU, and the candidate will be hosted by the Telethon Kids Institute and ideally based in Adelaide or Perth, however remote candidates will also be considered.

Project: Complex diseases impact Indigenous people disproportionately compared to non-Indigenous people. Up to 30% of the adult Indigenous population have T2D and suffer nine times the mortality, >10 times the rates of T2D-related end-stage kidney failure, three times the rates of heart disease and 38 times the rates of lower limb amputations than non-Indigenous Australians. Interestingly, disease risk varies significantly between biological sexes within Indigenous people, which impacts how individuals are treated in the clinic. In this study, we aim to quantify and investigate the sex differences in T2D and CVD disease risk using the Aboriginal Diabetes Study (PROPHECY) cohort. Using multiomics profiles of 1,245 individuals and reference datasets from the Genotype-Tissue Expression (GTEx) project, we will investigate the genetic impact of sex on adipose tissue and other related tissues within Indigenous participants. We aim to establish predictive models to accurately define CVD and T2D risk in Indigenous Australians and establish computational tools that could be implemented within a primary healthcare setting.

Funding: This project will be offered as a fully funded PhD scholarship + top up for 3 years.

Contact: Associate Professor Jimmy Breen Jimmy.Breen@telethonkids.org.au

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Standardising technical standards and approaches to supporting data sharing in Indigenous genomics

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Investigator: Associate Professor Kalinda Griffiths, Director Poche SA+NT & Research and Education Lead; Emeritus Professor Simon Easteal, College of Medicine and Public Health, ANU; Professor Daniel MacArthur, Centre for Population Genomics, Garvan Institute of Medical Research; Dr Hardip Patel, National Centre for Indigenous Genomics, ANU.

Location: The project will be led by Flinders University, and the candidate can be based in Darwin or Adelaide.

Project: The aim of this PhD project is to develop technical data standards in the collection and sharing of Aboriginal and Torres Strait Islander genomics and genomics related data. This project will identify existing guidelines, protocols, and formats for the generation, analysis, and interpretation of genomic data. It will also involve developing a set of agreed-upon rules and standards that ensure consistency, compatibility, and interoperability across different organisations, health care services, research institutions, and databases.

The work will centre Aboriginal and Torres Strait Islander voices and support cultural, ethical, and legal considerations within Aboriginal and Torres Strait Islander communities in the context of genomics drive these developments. It will emphasise collaboration, consent, cultural safety, and community engagement to ensure that Indigenous peoples' rights and interests are protected.

This is a national project that will support the development of a national Indigenous data governance framework aligned with international best practice to strengthen public trust; ensure appropriate data collection and sharing that reflects Indigenous principles; and develop standards for data collection, sharing, analysis, and reporting. Further it will work to enable seamless sharing and integration of genomic data, fostering collaboration, reproducibility, and accelerating scientific discoveries in the field of genomics. Individuals with experience in the data sciences, genomics and Indigenous health are encouraged to apply.

Funding: This project will be offered as 1 year fully funded with support to apply for RTP Scholarship with a top up to equal a $35,000 stipend over the 3years.

Flinders University | Poche SA+NT

Contact: Associate Professor Kalinda Griffiths kalinda.griffiths@flinders.edu.au

Equity in Genomics – defining future genomics research and care to reduce health inequality

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Investigators: Professor Alex Brown, Professor of Indigenous Genomics, Telethon Kids Institute and Australian National University; Ms Louise Lyons, Senior Manager, Strategy and Policy - Indigenous Genomics, Telethon Kids Institute; and Ms Tiffany Boughtwood, Australian Genomics

Location: The candidate will be hosted by the Telethon Kids Institute and ideally based in Adelaide, however candidates based in Perth, Queensland, Western Australia or Canberra will also be considered.

Project: This research will be to define equitable pathways for Aboriginal and Torres Strait Islander peoples to engage in genetic and genomic health systems, services and research. Equitable access to genetic health services and research requires heath professional commitment, health systems changes and opportunities that allow to Indigenous Australians to envision, lead and create the governance and service models that will deliver equitable health services and improved health outcomes for all Indigenous peoples. Readiness for change will be assessed along with barriers and opportunities that need to be addressed. This project scope will require Community and ATSICCHO consultations, and close collaboration with investigators undertaking research within the Indigenous Genomics research platform. Addressing health systems barriers and opportunities will also include emerging technologies and infrastructure (eg NAGIM), data governance and sovereignty models, commercial market, IP protections, and national policy gaps.
Project aims include:

- An international/national systematic review of genetic health systems barriers to Indigenous peoples seeking and retaining essential health services;
- Identifying strategies that strengthen referral, diagnostic and treatment pathways between primary health care and genetic health service providers;
- Identifying evidence-based and best practice models that successfully and measurably address health service access barriers;
- Developing an evaluation and impact framework that demonstrates benefits to Indigenous Australians and the mainstream genetic health services;
- Developing a national policy framework that addresses existing genetic and genomic health access gaps and identifies actions and resources that require government and mainstream service providers commitment to effect equitable access for Aboriginal and Torres Strait Islander people.

Funding: This project will be offered as a fully funded PhD scholarship + top up for 3 years.

Contact: Professor Alex Brown alex.brown@anu.edu.au

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Functional genomics approach to investigate CVD variants of uncertain significance in Indigenous Australians

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Investigators: Professor Sally Dunwoodie, Head of the Congenital Heart Disease Research Program and Director of the Innovation Centre of the Victor Chang Cardiac Research Institute; Professor Jamie Vandenberg, Head of the Cardiac Electrophysiology Laboratory of the Victor Chang Cardiac Research Institute; and Professor Jason Kovacic, Head of Vascular Biology Laboratory and Executive Director of the Victor Chang Cardiac Research Institute

Location: The candidate will be hosted onsite by the Victor Chang Cardiac Research Institute in Darlinghurst, Sydney.

Project: The Victor Chang Cardiac Research Institute has developed a multifaceted Functional Genomics approach to investigate variants of uncertain significance (VUS) for cardiovascular disease (CVD). Using high-throughput functional assays and in-silico methods, VUS can be efficiently screened, accelerating the pathway to a genetic diagnosis. In addition, novel multi-omics analysis pipelines have been developed to functionally assess genes associated with CVDs. Thus our focus in ALIGN is to identify and prioritise VUS and other causal genes, and to define their role in causing CVDs in Aboriginal and Torres Strait Islander peoples, integrating genomic, transcriptomic, metabolic and proteomic datasets. A suitable PhD student for this work would have experience in basic molecular and cell biology techniques (e.g. PCR, western blot, microscopy, cell culture) and would be willing to devote their PhD efforts toward understanding how gene variation causes CVD in Indigenous populations.

Funding: This project will be offered as a fully funded PhD scholarship + top up for 3 years.

Contact: Professor Sally Dunwoodie s.dunwoodie@victorchang.edu.au

Victor Chang
Cardiac Research Institute

Indigenous Cultural Understandings of Kinship and Inheritance as a Basis for Communicating Genomics

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Investigators: Professor Alex Brown, Professor of Indigenous Genomics, Telethon Kids Institute and Australian National University; Ms Louise Lyons, Senior Manager, Strategy and Policy - Indigenous Genomics, Telethon Kids Institute; Associate Professor Azure Hermes, Deputy Director, National Centre for Indigenous Genomics; and Ms Ms Cheryl Bridge, Head, Kulunga, Telethon Kids Institute

Location: The candidate will be hosted by the Telethon Kids Institute and ideally based in Adelaide, however candidates based in Perth, Queensland, Western Australia or Canberra will also be considered. Candidates can be enrolled through ANU, the University of Adelaide or the University of WA.

Project: Genomic and precision medicine represent a critical step change in health and medical sciences. Elucidation of the human genome has exposed the underlying biological architecture of human development and functioning, however diverse populations are not yet represented in, engaged with, nor do they have equitable access to, the benefits of genomic research. This is particularly true for Indigenous Australians, where much of this is due to a lack of engagement with Aboriginal and Torres Strait Islander peoples, and limited investment in enabling their leadership in genomics. Greater attention to self-determination in genomic science is long overdue, and the continued failure to respectfully engage and empower Indigenous communities runs the risk of further widening already significant health inequalities.

This project will involve deep engagement with Indigenous communities across multiple jurisdictions within Australia to explore cultural understandings of kinship, relatedness, inheritance and genetics as a foundational step in seeking common ground between Indigenous culture and genomic sciences. The goal will be to guide the development of educational and engagement resources and methods that: raise awareness of the utility and benefits of genomics for Indigenous people; explore and document Indigenous understandings of genetics and inheritance; improve and evaluate genetic literacy in communities and conversely, understanding among the genomics community of Indigenous knowledge systems and ethical approaches to genetic research; utilise Indigenous knowledge systems as a foundation to empower communities in genomic research and clinical care; and develop best-practice models or policies that increase Community participation and retentions within the genetic health service pathways

Funding: This project will be offered as a fully funded PhD scholarship + top up for 3 years.

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Contact: Professor Alex Brown alex.brown@anu.edu.au

Knowledge Tree: Unlocking the voice of Indigenous people and communities for equitable and precise health care

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Investigators: Professor Gareth Baynam, Medical Director Rare Care Centre and Honorary Research Fellow, Telethon Kids Institute; Professor Timo Lassmann, Program Head, Precision Health, Telethon Kids Institute; Professor Alex Brown, Professor of Indigenous Genomics ANU and Telethon Kids Institute; Associate Professor Azure Hermes, Deputy Director, National Centre for Indigenous Genomics; Libby Massey, Director Research and Education, Machado-Joseph Foundation; Professor Tudor Groza, Lead of Phenomics, Rare Care Centre; Professor Tom Gedeon, Curtin University, Director Optus Centre for AI; Dr Richard Palmer, School of Earth and Planetary Sciences, Lead Developer Cliniface; Professor Peter Robinson, Professor of Computational Biology, the Jax Laboratory; Mr Yarlalu Thomas, Precision Public Health Fellow and Inaugural Lyfe Languages Champion

Location: The candidate will be hosted by the Telethon Kids Institute and ideally based in Perth.

Project: Phenotyping is critical and cross-cutting for all genomics research, its translation and implementation. In a clinical context, phenotyping is a practitioner's daily work i.e., performing history, examination and investigations to diagnose and to inform culturally appropriate and safe implementation of treatment and care and its monitoring. Addressing phenotyping in a socially and culturally appropriate way is also key to diversity, equity, inclusion and scale. Phenotypic Standards (Phenopackets) have been progressed by the Global Alliance for Genomics and Health, the International Rare Diseases Research Consortium and others, and some precision phenotyping initiatives (Cliniface 3D facial analysis software and Lyfe Languages) have focused on addressing Indigenous inequity. At the level of geography tools such as Mappa (Mapping care closer to home) are advancing clinical care and integrating knowledge of Country, Lore, climatic conditions and language. However, compared to a primary focus on genotyping and other omics, a dedicated focus on phenotyping and phenomics (deep and precision phenotyping) has received comparatively little attention. Starting through the lens of rare diseases, and then expanding to more common disorders, the overarching aim is to progress phenotypic standards (Phenopackets, pedigree tools), ontologies (Human Phenotype Ontology, MAxO), and related technology interoperability and integration (omics, imaging, spatial analysis) to ensure the perspective of what communities want answers to guides and informs (gen)omic analyses to unlock pathways in health and disease.

Funding: This project will be offered as a fully funded PhD scholarship + top up for 3 years.

Contact: Professor Gareth Baynam Gareth.Baynam@health.wa.gov.au

Government of Western Australia
Child and Adolescent Health Service

Perth Children's Hospital

Rare Care Centre
Clinical Centre of Expertise for Rare and Undiagnosed Diseases

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