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NATIONAL CENTRE FOR INDIGENOUS GENOMICS

ANNUAL REPORT 2023

NCIG is now in the unique position to lead Australia's Indigenous genomics agenda, at a time when national and international efforts have identified the critical role that ancestrally diverse populations can play in the next era of genomic science.

Professor Alex Brown Director, NCIG

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The Australian National University acknowledges, celebrates, and pays respects to the Ngunnawal and Ngambri people of the Canberra region, and to all First Nations Australians upon whose traditional lands we meet and work, and whose cultures are among the oldest continuing cultures in human history. We acknowledge that Aboriginal sovereignty was never ceded. It always was and always will be, Aboriginal land.

OUR PURPOSE

The role of the National Centre for Indigenous Genomics (NCIG) is to ensure Indigenous Australians are at the forefront of health benefits that will flow into the national health system from the integration of genomic knowledge and technology.

OUR VALUES

- Integrity, Trust, Respect NCIG acts with integrity, builds trust and behaves ethically. NCIG gives respect and strives to earn it in return.
- Confidentiality, Privacy and Security NCIG protects the privacy of participants and the confidentiality and security of data and information about participants.

OUR VISION

To create a **National Centre of Excellence** – at the interface of Indigenous culture, knowledges, genomics and data science – to transform the health, wellbeing and prosperity of Indigenous Australians.



ABOUT THE NCIG COLLECTION



This is a very important story about our Mob's health

The National Centre for Indigenous Genomics (NCIG) was created in 2013 to manage and expand a collection of 7,000 historical Indigenous blood samples – now known as the NCIG Collection – held at the John Curtin School of Medical Research, and to develop a research reference resource from the DNA in the samples for genomic health and medical research.

Genomics is at the heart of new discoveries and technologies that are transforming medical practice. However, the lack of genomic data from Indigenous Australians means that research, data and the implementation of healthcare genomics are biased and inaccurate for Indigenous Australians.

The National Centre for Indigenous Genomics at the Australian National University (ANU), established by a federal statute is creating Australia's national Indigenous genomic data resource under a unique governance and research model, placing Indigenous Australians in charge of their genomic data and its use. NCIG is creating telomere-2-telomere Indigenous Australian reference genomes, and population scale genetic variation maps for partnering communities. This growing resource will encompass a remarkable depth and breadth of information useful for genomic medicine applications. Cover art: This illustration encapsulates the mission of NCIG to create a reference resource for Indigenous genomics for the benefit of Australia's First Nations Peoples. The vast red dirt plains and expansive blue skies, symbolising the rich and diverse environment where First Nations peoples have thrived for tens of thousands of years. The complex genomic code, represented by adenine (A), cytosine (C), guanine (G), and thymine (T) are often described as the letters of the long story in our DNA. The genetic code was sequenced for individuals of four Indigenous communities to make a library. The DNA stories captured in these books were read by scientists to describe the rich tapestry of genetic heritage for Indigenous Australians. The work of the Centre, and this artwork, represent the coming together of cuttingedge genomics with the knowledges held by Australia's First Nations peoples and their unparalleled contribution to our collective history.

Cover art design: Riemke Aggio-Bruce Page ii: Maryvale Road to Titjikala Photo: Azure Hermes

See NCIG's collection sites here





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FROM THE CHAIR



My term as Chair of the Board of the National Centre for Indigenous Genomics ends as 2023 draws to its close. I look back with pride on the recent achievements of the Centre, culminating with the publication of two papers in the journal *Nature*. The work of our talented researchers and collaborators is making a difference to the health and wellbeing of Indigenous Australians. Samples that were taken in perhaps not the best of circumstances will now go on to reap benefits for Indigenous communities around Australia.

During my tenure as Chair, I have been privileged to meet and work with inspiring individuals including Mick Gooda, Misty Jenkins and Simone Reynolds. I have also had the honour of witnessing the return of samples to community and by samples, I don't just mean inanimate objects, but samples that culturally are considered a missing part of our people, so to be involved with reuniting them with their families and country was deeply humbling and incredibly powerful.

Being appointed to this NCIG Board has been a life-changer for me – I've had some fantastic opportunities to work in an area in which I have great hope for – the health and wellbeing of our kids. I've met some amazing people and we've had a heck of a journey being involved with this ground-breaking Centre.

Under the leadership of Prof Alex Brown and Assoc Prof Azure Hermes, I know the Centre will go from strength to strength from here on and become what we envision, a National Centre of Excellence to transform the health, wellbeing and prosperity of Indigenous Australians. I'm excited to watch it happen in the years to come.

Professor Glenn Pearson Chair, NCIG Board



FROM THE DIRECTOR



It is an exciting time to be joining the NCIG family, and my first 12 months supporting the team has flashed by in a heartbeat. Suffice to say we have much to be proud of. Since its establishment, NCIG has traversed the challenging terrain of culture, ethics, and science, following the identification and requirements for appropriate care of legacy samples, and the acknowledgement of the uncomfortable past treatment of Indigenous Australians in the name of science.

NCIG is now in the unique position to lead Australia's Indigenous genomics agenda, at a time when national and international efforts have identified the critical role that ancestrally diverse populations can play in the next era of genomic science – one fully cognisant of what our understanding of under-represented populations can mean for the health of humanity. Our commitment remains resolute. To developing and communicating the culturally appropriate management of a unique Indigenous genomic data resource; the establishment of Indigenous Australian sovereignty over their genomic data and its use; and the provision of a safe, sustainable, *'keeping place'* for the collection.

It is now time to extend the ambition and reach of NCIG to become Australia's national reference point for the investigation, conduct, translation and communication of Indigenous genomics research, underpinned by culturally appropriate governance mechanisms and utilising an Indigenous-led, world class genomics ecosystem.

Whilst these new ambitions represent an exciting time, they remind us that change is inevitable. In this context, I wish to thank the long-standing and irreplaceable support and wisdom of those who have or will move on from the day-to-day efforts of NCIG in the months ahead. Early in the year Lyndon Ormond-Parker retired from the board after serving since 2019. Glenn Pearson steps down from the role of Chair of NCIG after 5 years of humble service. Graham Mann and Keith Nugent are stepping into different stages of their lives and leave their roles in the heart of NCIG. No words can express how important their contributions have been, or the gratefulness we have for their wise words, commitment, kindness, and friendship.

The NCIG Project Manager and Board Secretariat, Prue Beckett, left the team in March 2023 and was replaced by Deb Marburg in June. The NCIG team has grown significantly with more than 10 new staff members joining us in 2023. I warmly welcome our new appointments, details of which can be found further along in this report.

To Deputy Director Azure Hermes and the team at NCIG thank you for all you have done in 2023. Keep up the good work. Our communities deserve everything we can bring to better meet their needs and aspirations.

Professor Alex Brown Director, NCIG



VALE DR SHARON HUEBNER



Photo supplied by Sharon Huebner



In December we received the sad news of the passing of Dr Sharon Huebner after a long illness. Sharon was a Discovery Early Career Researcher Award (DECRA) Fellow since 2022 in the National Centre for Indigenous Genomics, and a much-loved long-term friend and colleague of the NCIG family.

Sharon earned a PhD from Monash in 2016 in First Nations research ethics and principles, cultural and intellectual property rights, and First Nations repatriation and self-determined governance. She was a Chief Investigator in recent National Health and Medical Research Council (NHMRC) Synergy and Medical Research Future Fund (MRFF) grants led from ANU. She passed away only a short time before a paper on which she was an author (and one of a pair of papers from NCIG) was published in *Nature*.

Sharon's passion for addressing the barriers faced by First Nations people in healthcare genomics and her sharp intellect will be missed.

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CARING FOR THE COLLECTION

The NCIG Collection comprises biospecimens, historical documents archive, genomics data, social science artefacts and audio-visual stories of Indigenous peoples securely located within the John Curtin School of Medical Research (JCSMR) at the ANU under the custodianship of the Indigenous-majority NCIG governance board.

Biospecimen collection

There are over 7000 biological samples preserved at the JCSMR in –80°C freezers and liquid nitrogen tanks until their use is determined by donors and their families. In 2023, Carly Conlan joined the team as our first dedicated Senior Technical Officer. Carly is responsible for the development and implementation of policies and standard operating procedures for the processing and management of biological samples. She works closely with Jiaxin Yuan, NCIG Data Manager, to perform auditing of samples, ensuring that NCIG Biobank Operations – the handling, processing, storage, and documentation of NCIG biospecimens and their associated data – meets national and international standards.

MediaFlux Data platform implementation

NCIG has partnered with the National Computational Infrastructure (NCI) to implement the MediaFlux data platform. This initiative is a major step to align with international best practices in technical aspects of genomics data management. In addition, the platform enhances transparency, auditing, and security of the NCIG's genomics data collections. We have successfully ingested most genomics data into the platform and are continuously refining data models and security features. Integration with the FreezerWorks sample management system and the researcher access platform is underway, aiming to streamline data access.

Collection Access and Research Advisory Committee platform

NCIG is collaborating with ANU's Service Solutions team within the Planning and Service Performance division to progress the development of the Collection Access and Research Advisory Committee (CARAC) platform. This innovative system is set to transform the way researcher requests for access to the NCIG Collection are managed, bringing efficiency and transparency to the process. The CARAC platform is strategically built on the Endpoint IQ Conduit system, which is also utilised by the ANU's Human Research Ethics Committee for ethics application workflows. Here we capitalise on ANU's established capabilities to ensure long-term alignment with ANU services and support for effective management of resources.

Migration to Nextflow analysis platform

Our ongoing collaboration with the NCI focuses on transitioning our genomic analysis workflows to the Nextflow platform. This platform ensures reproducibility and interoperability in complex genomics data analyses. Kosar Hooshmand is successfully migrating the NCIG's variant calling analysis workflow to Nextflow. We plan to expand this to encompass data ingestion, management, quality assessment, and to support pangenome graph-based analyses. These enhancements aim to produce high-quality, comprehensive variant profiles for Indigenous Australians.

ANCIENT DIVERSITY, MODERN SCIENCE: REVEALING THE GENOMIC HERITAGE OF INDIGENOUS AUSTRALIANS

In 2023, NCIG reached an important milestone in genomic research with the publication of two important studies in the prestigious *Nature* journal. These studies represent a major stride in understanding the genetic diversity of Indigenous communities.

Employing the latest DNA sequencing technologies and advanced data analysis methods, the research brought together teams from multiple institutions. This collaborative effort using data from the NCIG Collection was dedicated to unravelling the complex genetic landscape of four Indigenous communities, comparing their genetic diversity with other global populations.

The findings of these studies are profound: up to 25% of the genetic variation identified in these Indigenous communities is unique to Indigenous Australians.

The discovery and genomic resources created through both studies carries significant implications for a multitude of health aspects:

- 1. these genomic resources will be critical in diagnosing rare diseases for Indigenous families
- 2. provide a foundation for understanding the genetic basis of complex diseases
- 3. improve our understanding of the human biology.

Both studies lay a critical foundation for ongoing research efforts. The aim is to ensure that the rich genetic diversity of Indigenous Australians is comprehensively represented and effectively utilised in advancing genomic-driven healthcare. This research underscores our commitment to equitable research practices and the delivery of tangible health benefits to Indigenous communities.

Indigenous Australian genomes show deep structure and rich novel variation

Matthew Silcocks, Ashley Farlow, Azure Hermes (Gimuy Walubara Yidinji), Georgia Tsambos, Hardip R. Patel, Sharon Huebner, Gareth Baynam, Misty R. Jenkins (Gunditjmara), Damjan Vukcevic, Simon Easteal, Stephen Leslie & The National Centre for Indigenous Genomics.

Nature, Volume 624, 13 December 2023



Dr Matthew Silcocks – Research Fellow Photo supplied by Matthew Silcocks

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Our study has revealed the immense degree of genomic diversity amongst Indigenous Australian communities and has laid a solid foundation for meaningful medical research in the future. **99**

Abstract

The Indigenous peoples of Australia have a rich linguistic and cultural history. How this relates to genetic diversity remains largely unknown because of their limited engagement with genomic studies. Here we analyse the genomes of 159 individuals from four remote Indigenous communities, including people who speak a language (Tiwi) not from the most widespread family (Pama-Nyungan). This large collection of Indigenous Australian genomes was made possible by careful community engagement and consultation. We observe exceptionally strong population structure across Australia, driven by divergence times between communities of 26,000-35,000 years ago and long-term low but stable effective population sizes. This demographic history, including early divergence from Papua New Guinean (47,000 years ago) and Eurasian groups, has generated the highest proportion of previously undescribed genetic variation seen outside Africa and the most extended homozygosity compared with global samples. A substantial proportion of this variation is not observed in global reference panels or clinical datasets, and variation with predicted functional consequence is more likely to be homozygous than in other populations, with consequent implications for medical genomics. Our results show that Indigenous Australians are not a single homogeneous genetic group and their genetic relationship with the peoples of New Guinea is not uniform. These patterns imply that the full breadth of Indigenous Australian genetic diversity remains uncharacterised, potentially limiting genomic medicine and equitable healthcare for Indigenous Australians.

A plain English summary of this paper can be found here.

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The landscape of genomic structural variation in Indigenous Australians

Andre L. M. Reis, Melissa Rapadas, Jillian M. Hammond, Hasindu Gamaarachchi, Igor Stevanovski, Meutia Ayuputeri Kumaheri, Sanjog R. Chintalaphani, Duminda S. B. Dissanayake, Owen M. Siggs, Alex W. Hewitt, Bastien Llamas, Alex Brown, Gareth Baynam, Graham J. Mann, Brendan J. McMorran, Simon Easteal, Azure Hermes, Misty R. Jenkins, The National Centre for Indigenous Genomics, Hardip R. Patel & Ira W. Deveson.

Nature, Volume 624, 13 December 2023



Dr Andre L. M. Reis – Bioinformatics Research Officer Photo supplied by Andre Reis

Abstract

Indigenous Australians harbour rich and unique genomic diversity. However, Aboriginal and Torres Strait Islander ancestries are historically under-represented in genomics research and almost completely missing from reference datasets. Addressing this representation gap is critical, both to advance our understanding of global human genomic diversity and as a prerequisite for ensuring equitable outcomes in genomic medicine. Here we apply population-scale whole-genome long-read sequencing to profile genomic structural variation across four remote Indigenous communities. We uncover an abundance of large insertion-deletion variants (20-49bp; n=136,797), structural variants (50b-50 kb; n=159,912) and regions of variable copy number (>50 kb; n=156). The majority of variants are composed of tandem repeat or interspersed mobile element sequences (up to 90%) and have not been previously annotated (up to 62%). A large fraction of structural variants appear to be exclusive to Indigenous Australians (12% lower-bound estimate) and most of these are found in only a single community, underscoring the need for broad and deep sampling to achieve a comprehensive catalogue of genomic structural variation across the Australian continent. Finally, we explore short tandem repeats throughout the genome to characterise allelic diversity at 50 known disease loci, uncover hundreds of novel repeat expansion sites within protein-coding genes, and identify unique patterns of diversity and constraint among short tandem repeat sequences. Our study sheds new light on the dimensions and dynamics of genomic structural variation within and beyond Australia.

A plain English summary of this paper can be found here.

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This research is crucial because it reveals the unique genetics of Indigenous Australians, [which is] often overlooked in genomics. Their absence from global genomics hampers understanding and fairness in genomic healthcare in Australia.

The developed analysis methods and reference data are pivotal for the future of genomic medicine in Australia, enabling the identification of disease-causing DNA variation through comparison with a large unaffected group of similar ancestry. **99**

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CELEBRATING 10 YEARS OF INDIGENOUS GOVERNANCE – NCIG SUMMER ORATION 2023

To celebrate the work of the last 10 years, NCIG held their Summer Oration at the National Film and Sound Archive, in early December 2023. This event, held on a perfect balmy Canberra evening, featured a Welcome to Country by Aunty Caroline Hughes, followed by the keynote speech from ANU Vice Chancellor, Professor Brian Schmidt, a question-and-answer session with NCIG Director, Deputy Director and Board Chair plus the launch of the R M Wunungmurra Community Repatriation Fund (see <u>page 13</u> for details). Afterwards, over 100 invited guests gathered in the courtyard for refreshments and live music by Will Kepa and Monika Duggan.

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Milestones like this [10-year celebration] allow us to come together to reflect on the successes we've achieved, the challenges we've overcome, and the future we hope to write. **99**

Professor Brian Schmidt



Excerpt from Summer Oration delivered by Professor Brian Schmidt



Left: Prof Brian Schmidt; right: Prof Brian Schmidt & Evelyn Djotja Photos: Tracey Nearmy

It's an honour to have been invited to deliver the second National Centre for Indigenous Genomics Summer Oration on this special occasion: the Centre's 10-year anniversary.

Milestones like this allow us to come together to reflect on the successes we've achieved, the challenges we've overcome, and the future we hope to write

NCIG are the custodians of a precious knowledge, overseeing 7,000 historical blood samples from 35 diverse Indigenous communities around Australia. There are many stories contained in those tiny samples. Stories about the people the samples came from and stories about how and why the samples were taken and stored.

It is because of this that NCIG and the University are recommitting ourselves on this tenth anniversary:

- to deepening our understanding of the cultural, historical, and scientific significance of Australia's First Peoples and their profound wisdom
- to being a trusted caretaker of precious and highly significant collections of Indigenous knowledges, histories, biological samples, and gifts
- to becoming an international leader and global centre of excellence in understanding genomic variation among Indigenous peoples
- to applying Indigenous knowledges and genomic sciences to deliver meaningful and direct benefit to the health and wellbeing of Indigenous Australians now and into the future
- to implementing a comprehensive, culturally appropriate plan for the repatriation of biological samples back to their respective communities.

The collection contains materials that were collected mostly in the 1960s and 70s by people seeking answers to some big questions about who we are, and where we've been. But in the 1990s, a moratorium was imposed on their use, due to evolving ethical considerations, and concerns for the agency of Indigenous peoples.

In 2012, an Indigenous-led consultative committee was formed to advise ANU on both the management and use of the samples. They determined that the collection was of 'immense cultural, historical and scientific importance'.

This committee made eight recommendations on how to manage the collection and ANU accepted them all. Two main recommendations were to establish a national centre to care for the samples and the need to form an Indigenous-majority Governance board, hence in 2013, the National Centre for Indigenous Genomics or NCIG was born.

The inaugural meeting in 2013, guided by Mr Mick Gooda, embarked on a mission to empower Indigenous communities to have control over the genomic data held at the ANU. The aim was to return the authority over this historical collection to the very participants whose information it contained. The board's primary responsibilities were centred on overseeing the strategic course of the NCIG, handling high-level issues, formulating communication policies and providing support for the operational team.

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A third important recommendation was the need to engage with all 7,000 individuals, or their families, whose samples were in the collection. It was recognised that the fate of the samples - whether they should be retained, returned to communities, or respectfully put to rest – should be determined in a manner both ethically and culturally appropriate. Ms Azure Hermes, a Gimuy Walubara Yidinji woman from Far North Queensland, joined the team in 2015 as a Community Engagement Officer and started the mammoth task of working with the communities whose samples are housed in the collection. It is a slow and meticulous process which can only be advanced at the speed of trust. The initial hard conversations in the Yarrabah community set a benchmark for conducting outreach in other communities. There are now 2000 cases in which community members have given consent, with 90 per cent of participants willingly agreeing to be part of the NCIG collection.

Early external partnerships with Canberra Medical Society and Bioplatforms Australia provided funds that enabled community resources, such as the NCIG animation and the genomic sequencing of 400 samples to be carried out. This support, along with community engagement, laid the foundation for the first Aboriginal Biobank. More recently, the National Computational Infrastructure at ANU provides invaluable support with storing and managing 500 terabytes of genomic data securely and with significantly less cost than would otherwise be the case.

The last ten years of hard work and collaboration have built an incredible foundation for NCIG's future. In the years ahead we will be able to unlock the potential of genomics to deliver direct and meaningful health and social benefit to Indigenous Australians. The Centre has plans to enrich its research by focusing on:

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- supporting First Nations people to become leaders in diverse areas of genomics research
- expanding their educational and teaching offerings for community and health stakeholders
- developing and harnessing a deep understanding of the uniqueness of Australia's First Nations peoples and harnessing these understandings to build powerful tools for precision medicine.

Thank you to the Indigenous-led consultative committee, past and present board members, Associate Professor Azure Hermes, all staff past and present, and to our community partners. Your commitment, collaborative spirit, and hard work have been essential in achieving the Centre's goals and fostering a supportive working environment. Join me in congratulating the National Centre for Indigenous Genomics on ten remarkable years.

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NCIG ANNUAL REPORT 2023



Left: Monika Duggan & Will Kepa; middle: Latisha Baker, Prof Brian Schmidt & Evelyn Djotja; right: Prof Brian Schmidt and Prof Alex Brown Photos: Tracey Nearmy

Reflection on the early work of the NCIG Board

At the 2023 NCIG Summer Oration, inaugural Deputy Chair of the NCIG Governance Board, Associate Professor Misty Jenkins, provided a reflection on the early work of the Board.

Misty recalls that ...

... at the time, there were few biomedical Indigenous researchers in Australia, and it was also a time when the power of genomics was being realised. It was evident that there was a need to tackle chronic disease and intergenerational disadvantage in Indigenous communities. Deep consultative community engagement would be the cornerstone of the Centre and it would take true partnerships between communities and NCIG to develop policies that would build the most culturally safe architecture for the proper handling of the samples.

Over the years, with the support of ANU Deputy Vice Chancellors and senior staff of the John Curtin School of Medical Research, many grant applications were written, countless months of meetings to engage with NHMRC and Philanthropy were held to secure the sustainability of the organisation. The focus was always firmly squared on Indigenous health. The core values of NCIG were to provide an Indigenous lens to genomics, to hopefully one day be able to integrate Indigenous genomics into clinical pathways so that our mob are not left behind the revolution of personalised medicine.



Professor Misty Jenkins AO. Photo supplied by Misty Jenkins

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It has been very special to watch the organisation grow and strengthen in its Indigenous leadership and [become] a voice to ancient wisdom but bringing it together with modern technologies like genomics. **99**



Mr R M Wunungmurra sitting with baskets filled with repatriated samples in Galiwin'ku. Photo: Jamie Kidston

Launch of the R M Wunungmurra Community Repatriation Fund

At the closing of the NCIG Summer Oration in December 2023, ANU Vice Chancellor, Professor Brian Schmidt, launched the R M Wunungmurra Community Repatriation Fund, as a tribute to the influential late Mr. Ross M. Wunungmurra, a distinguished Yolŋu man.

Mr. Wunungmurra's vision and dedication instigated real change, bridging worlds in Yolŋu Nation, including the Galiwin'ku community, Northeast Arnhem Land. His advocacy for the repatriation of NCIG samples, whilst recognising the benefits of genomics for his people, led to the return of over 200 samples in 2019.

By promoting this fund to philanthropic organisations and the wider community we hope to raise enough funds to continue this vital work, returning samples to their rightful place and perpetuating Mr. Wunungmurra's vision of unity and progress for generations to come.



Mr. R M Wunungmurra's consent for using his blood samples and belief in genomics' benefits for his people's health were pivotal. Known for his charisma, intelligence, and strong will, he formed lasting friendships and inspired many.

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Evelyn Djotja and Litisha Baker, NCIG Summer Oration, 4 December 2023



To make a donation, please scan the QR code or visit www.anu.edu.au/giving



CONFERENCES, PRESENTATIONS & COMMITTEES

Conferences

Indigenous Genomics Data Workshop

On June 13th and 14th, 2023, NCIG hosted a pivotal two-day Indigenous Genomics Data Workshop. This workshop gathered data scientists working with Indigenous genomics data in Australia, specifically within the NCIG, CONNECT and ALIGN programs. The workshop was structured around four major themes, with the primary aim of pinpointing questions needing community input.

- 1. Data Storage and Archiving Standards: a key focus was on evaluating the Global Alliance for Genomics and Health (GA4GH) standards for data storage and archiving. Discussions centred on how to maintain a principled approach in data storage and archiving, technical infrastructure needs and priorities to address.
- 2. File Classification and Data Management: the workshop discussed the categorisation of various file types based on the level of individual identification they contain, ranging from raw DNA sequence data per individual to summarised data on groups. This systematic categorisation aimed at enabling appropriate data management practices.
- 3. Emerging Standards for Genomic Data Analysis: discussions were aimed at collaboratively evaluating emerging standards in genomic data analysis, such as reference genome types and sources and workflow management systems. The goal was to develop a roadmap for establishing a community of practice and standards, enhancing consistency and interoperability of findings across different research groups.
- Research Data and Clinical Decision Making: this was aimed at understanding how research data can inform clinical decision-making, identifying priorities, and potential workflows to format research outputs for clinical deployment.

This workshop represented a significant step for NCIG in advancing the integration of Indigenous genomics data into broader research and clinical contexts, focusing on ethical, technical, and practical aspects of data handling and application.



Workshop participants outside the National Computational Infrastructure building. Photo: Hardip Patel



Genomic Diversity – Insights from Long-Read Sequencing – two-day symposium

In July 2023, NCIG co-hosted an insightful two-day symposium titled 'Genome Diversity – Insights from Long-Read Sequencing', in collaboration with the Centre for Biodiversity Analysis at the Research School of Biology, Australian National University. This event was a deep dive into the latest developments in third-generation genomic sequencing platforms like PacBio and Oxford Nanopore Technology.

The symposium's agenda was comprehensive, covering various aspects of genome assembly, haplotypes, pan-genomes, and structural variations. It provided a unique opportunity for attendees to gain both biological and technological insights, across plants, animals, and humans. This focus on diversity underscored the rapidly expanding scope of genomic research and its applications.

NCIG staff and students were among the attendees, seizing the chance to enrich their experience and expand their knowledge base. Their participation in this symposium not only enhanced their understanding of cutting-edge genomic technologies but also highlighted the importance of integrating diverse genomic data into research. This event exemplified NCIG's commitment to staying at the forefront of genomic research and education.



Left to right: Karen Miga (UC Santa Cruz), Bastien Llamas (NCIG), Ann McCartney (UC Santa Cruz) and Joel Keen (NCIG) at the National Botanic Gardens, Canberra. Photo: Hardip Patel

XXIII International Congress of Genetics

In July 2023, NCIG played a pivotal role at the XXIII International Congress of Genetics in Melbourne. Under the overarching theme 'Genetics & Genomics: Linking Life & Society', this premier global forum was a melting pot of ideas and groundbreaking research in genetics and genomics. The NCIG notably contributed to two symposia, underscoring its commitment to fostering international collaboration and knowledge exchange.

The symposia featured four distinguished speakers: Phil Wilcox, Karen Miga, Ann McCartney, and Nadine Carone. These renowned scientists shared their cutting-edge work in genomics, particularly highlighting their partnerships with Canadian First Nations and Maori populations. Their presentations delved into the creation of diversityinclusive reference resources and strategies to ensure equitable access to genomic advancements. Additionally, each symposium included four other speakers, selected based on the merit and relevance of their abstracts. These speakers added a diverse range of perspectives and insights into human genomic diversity, further enriching the discussions. Their contributions exemplified the NCIG's commitment to fostering a broad and inclusive scientific dialogue, ensuring a comprehensive representation of the latest advancements and challenges in the field of genetics and genomics. This initiative by the NCIG not only showcased the latest in genomic research but also emphasised the importance of inclusivity and collaboration in the scientific community.



Speakers at XXIII International Congress of Genomics; top: Karen Miga; bottom left: Phil Wilcox; bottom right: Azure Hermes Photos: Hardip Patel

Presentations

Dr Alex Brown, Director, NCIG

- Towards a National Indigenous Genomics Network, International Society of Stroke, Healesville, March 2023
- Towards a National Indigenous Genomics Network, Lowitja Health and Wellbeing Conference, Cairns, June 2023
- Indigenous Genomics and the First Scientists, DISER Reconciliation Week Lecture, Canberra June 2023
- Towards Indigenous Sovereignty in Genomics, ALIGN Satellite Meeting – XXIII International Congress of Genetics 2023, Melbourne, July 2023
- Leveraging bioinformatics capacity to implement African genomics for healthcare, XXIII International Congress of Genetics 2023, Melbourne, July 2023
- Equity & Diversity in the Application of Human Genomics, XXIII International Congress of Genetics 2023, Melbourne, July 2023
- Empowering Indigenous Communities in Genomics, Queenstown Research Week, Queenstown NZ, August 2023
- Genomics and Indigenous Health Equity, Monash School of Epidemiology and Public Health, Melbourne, September 2023
- ALIGN a national Indigenous Genomics Network, Human Genetics Society of Australia, Adelaide, September 2023
- Indigenous Genomics a National Agenda, American Society of Human Genetics, Washington DC USA, November 2023

Assoc Prof Azure Hermes, Deputy Director, NCIG

- CPG Seminar, Centre for Population Genomics, Online March 2023
- NCIG, ANU Students Guest Lecture, March 2023
- Indigenous Genomics in the Precision Medicine Era, XXIII International Congress of Genetics 2023, Melbourne, July 2023
- Empowering Communities in Genomics, ALIGN Satellite Meeting – XXIII International Congress of Genetics 2023, Melbourne, July 2023
- Importance of community engagement in remote Indigenous communities in Australia, American Society of Human Genetics, Washington, November 2023

- Importance of community engagement in remote Indigenous communities in Australia Australasian Gastro-Intestinal Trials Group, Online, November 2023
- Introduction to Indigenous Genomics, EMBL [European Molecular Biology Laboratory] Australia PhD, JCSMR, November 2023

Dr Hardip Patel, Bioinformatics Lead NCIG

- Progress in Genome Biology and the Development of Reference Resources, ALIGN Satellite Meeting – International Congress of Genetics 2023, Melbourne, July 2023
- Enabling the inclusion of First Nations peoples in genomics, Genome Diversity – Insights from longread sequencing, ANU Research School of Biology, Canberra, July 2023
- Introduction to Genomics for NCIG Board Members, September 2023
- Enabling the inclusion of First Nations peoples in genomics, Nanopore Community Meeting, Singapore, September 2023
- Data developments in international genomics, Indigenous Genomics Data Workshop, Adelaide, October 2023
- FAIR and CARE for genomics data, Australian Alliance for Indigenous Genomics (ALIGN) capacity building seminar series, October 2023

Kirat Alreja

 Manua curation of genome assemblies for Australian Reptiles and Amphibians, XXIII International Congress of Genetics 2023, Melbourne, July 2023

Dr Michael He

 Indigenous Genomics in Indigenous Hands: Learnings from ANU NCIG, ANU Centre for Biodiversity Analysis Stakeholder Engagement Workshop, ANU Research School of Biology, Canberra, November 2023

Dr Kosar Hooshmand

 Reference Genomic Resources for Indigenous Australians, XXIII International Congress of Genetics 2023 Melbourne, July 2023

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Joel Keen

- Grounding international students' learning experiences within Australian Indigenous perspectives, Centre for Chinese Contemporary Studies: pilot study outcomes, Online, October 2023
- Indigenous Genomics in Indigenous Hands: Learnings from ANU NCIG, ANU Centre for Biodiversity Analysis Stakeholder Engagement Workshop, ANU Research School of Biology, Canberra, November 2023
- PhD Presentation: The Bounded Westernised Relationship with Knowledge, CONNECT Scientific Retreat, Kioloa, November 2023

Alice McCarthy

- Progress and aims of the Building knowledge of genomics in partnership with Indigenous Communities and Health Services, Centre for Population Genomics, Garvan Institute of Medical Research and Murdoch Childrens Research Institute, Online, March 2023
- Progress and aims of the Building knowledge of genomics in partnership with Indigenous Communities and Health Services, Australian Genomics Policy Network, Online, August 2023
- Progress and aims of the Building knowledge of genomics in partnership with Indigenous Communities and Health Services, CONNECT Scientific Meeting, Online, August 2023
- Progress and aims of the Building knowledge of genomics in partnership with Indigenous Communities and Health Services, Australian Genomics National Steering Committee, Online, August 2023

Dr Lyndsay Newett

- Empowering Communities in Genomics, Australasian Association of Bioethics and Health Law (AABHL)
 Conference: Change, Impact and Opportunities, QUT, Brisbane, November 2023
- Genomics, Human Research Ethics Committees, and Waivers of Consent, 4th National Human Research Ethics Committees (HREC) Conference, Online, November 2023

Jiaxin Yuan

- Multi-system Infrastructure for Indigenous Genomics: empowering transformative advancements in biomedical research and clinical applications, International Congress of Genetics 2023, Melbourne, July 2023
- CARAC platform overview, NCIG Board Meeting, December 2023

Current Committee membership

Aboriginal and Torres Strait Islander Advisory Group on Health Genomics

Australian Genomics Policy Network

Connect Executive Committee

Global Alliance for Genomics & Health (GA4GH) – Advisory Group

Human Cell Atlas Outreach Australia

Human Genetics Society of Australasia – Indigenous Genomics Steering Group

Indigenous Health and Wellbeing Grand Challenge Executive Committee

JCSMR Scholarship Committee

Multi Stakeholder Advisory Group – University of Queensland

Sing Australia Working Group

Undiagnosed Disease Network – Australia: Indigenous Advisory Committee

Undiagnosed Disease Network – Australia: Steering Committee

Yalu Research Committee



RESEARCH UPDATES

Respecting the Gift: Empowering Indigenous Communities in Genomics Medicine

Professor Alex Brown is leading a consortium which was awarded \$5 million under the National Health and Medical Research Council's Synergy Grant program (2022–2026). The ambitious multi-institutional program of work planned under the grant is spread across five research domains. NCIG participates in 3 domains.

Chief Investigators: Prof Alex Brown, ANU, Telethon Kids Institute; Ms Azure Hermes, NCIG

Project Leads: ANU and NCIG: Prof Graham Mann; Emeritus Professor Simon Easteal; Dr Sharon Heubner; Dr Hardip Patel

Domain 1 Empowering Communities in Genomics

Genomics brings advantages and risks to all people. Still, this area of research has the potential to benefit, and negatively impact Indigenous peoples, disproportionately.

The social research component of this project is being designed to investigate Aboriginal and Torres Strait Islander peoples' views regarding the ways genomics is, and can be, undertaken in Australia. Working with communities, we plan to examine and compare Indigenous peoples' perspectives on consent, genomic data sharing, and other practices relating to this research field, as well as identify the questions and concerns that people have in relation to genomics. The knowledge we build from this component will contribute to the establishment of principles that can be used to assess, and generate, genomic guidelines.

With the support of Gurriny Yealamucka Health Services Aboriginal Corporation, we will continue to work with community members of the Yarrabah Aboriginal Shire on this research. We have also received support from Yalu Aboriginal Corporation to undertake this project component in Galiwin'ku, and hope that in 2024 it will be possible for us to work with other communities in the Northern Territory, as well as Western Australia, to extend on this research.

Domain 2 Genomic Data Sciences

Genomics data management and sharing with researchers and clinicians is an active area of development globally. This domain of work focuses on identifying ways to create appropriate data stewardship mechanisms and data representation frameworks that embed Indigenous data sovereignty at the core and enable genomics driven healthcare and research. NCIG staff contributed to the workshop organised by Prof Kalinda Griffiths in Adelaide and discussed "Data developments in International Genomics". Dr Patel also presented about "FAIR and CARE principles in genomics" in the capacity building seminar series to a diverse audience interested in Indigenous genomics.

In 2023, we ingested NCIG genomic data collections into the secure and flexible MediaFlux data management platform. Since then, we have focused on creating participant-centric data models that are aligned with international genomics data repositories to ensure effective management of genomic data. This system will have the ability for NCIG participants and their representatives to have fine-grained control over their data.

Domain 3 Variation

This domain focuses on enhancing Indigenous genomics resources. The NCIG has successfully completed genomic data generation for over 550 consenting participants. New data will allow for the creation of accurate population variation maps and the construction of reference genomes. To enhance the reproducibility and scalability of our analysis workflows, we are transitioning to the Nextflow system. This move, in partnership with the National Computational Infrastructure, has allowed us to refine our Nextflow workflows for increased efficiency and scalability, making it possible to process thousands of samples effectively. After conducting further testing and validation, our analytical platforms will feed into the development of a comprehensive variant database for Indigenous communities.





Building Genomic Knowledge in Partnership with Indigenous Communities and Health Services

Project Lead: Ms Azure Hermes, NCIG

This public health program is designed to complement the Indigenous Genomics Ecosystem research platform funded through the NHMRC Synergy grant led by Prof Alex Brown. Australian Genomics provided support to this project through their genomic implementation project funding.

Through this project, NCIG, in partnership with community-controlled health organisations and other community representatives, is bringing the perspectives and knowledge of communities, community organisations, scientists and doctors together to build knowledge of genomic medicine and what it could mean for Aboriginal and Torres Strait Islander peoples. This project is contributing towards NCIGs goals of equity in genomic medicine by contributing to efforts identifying and aligning genomic medicine and research with community priorities.

The project has developed strong collaborative relationships with community partners across 5 communities and 3 states and has attracted pro-bono support from other institutions. We have drafted a suite of culturally informed resources about genomics and the experience of engaging with genomic health services. We have leveraged the initial funding to secure ongoing support and are approaching this work as an ongoing priority.



Community partners at project workshop, June 2023, Kaurna country, Adelaide. Left to right: Louise Lyons (TKI), Carl Nixon (Tiwi Islands), Mitchell Garawitja (Yalu Aboriginal Corporation), Jim O'Shea (VACCHO), Alice McCarthy (NCIG), Amanda Richards-Satour (TKI), Selina Puruntatameri (Tiwi Islands), Olivia Payne (VACCHO), Katrina Connolly (Gurriny), Zahra Zainal, Azure Hermes (NCIG), Therese Bourke (Tiwi Islands), Rubijayne Cohen (NCIG), Laura Purcell (Australian Genomics), Keri Finlay (Australian Genomics), Lyndsay Newett (NCIG). Photo: Johanna Barclay

Tiwi Islanders – Kidney Disease Project, ANU Indigenous Health and Wellbeing Grand Challenge (IHWGC) 2020 – 2025

Project Lead: Assoc Prof Brendan McMorran, ANU; Dr Simon Jiang, ANU; Dr Hardip Patel, NCIG; Ms Azure Hermes, NCIG

NCIG manages data for the Tiwi kidney disease project. Through our efforts, we have established an Identity Management and Tracking system using FreezerWorks to ensure accurate chain-of-custody records to enable provenance tracking of data and samples. Using these newly developed models for identity tracking, we have begun the process of organising genomic information and samples. Relevant standard operating procedures and policy documents are developed. We aim to work with Tiwi peoples to ingest their genomic data, clinical records and other important health data. In addition, Garett Ball completed an undergraduate research project to investigate structural variations in genomic loci associated with kidney function revealing markers for further testing.

LINEAGE social research project



NCIG is part of LINEAGE, a social research project that is being conducted to explore aspects of genomic data governance in Australia. Specifically, this project is designed to obtain Aboriginal and Torres Strait Islander peoples' perspectives on genomic data storage procedures, and the return of unexpected findings. It is underpinned by a Participatory Action Research approach, which allows us to work with Indigenous research partners, and communities, to identify the benefits and risks associated with genomics and Australian research protocols, as well as culturally appropriate ways to move forward. Currently, this project has support from Gurriny Yealamucka Health Services Aboriginal Corporation and Yalu Aboriginal Corporation, to be undertaken in the Yarrabah Aboriginal Shire, and Galiwin'ku.

Australian Alliance for Indigenous Genomics program

ALIGN Astralian Allance for Indigenous Genomics

NCIG with other partner organisations and researchers from the ACT region are contributing to the Australian Alliance for Indigenous Genomics (ALIGN) program, funded by MRFF. The ACT node is in the process of forming the ACT Indigenous Governance Committee (ACTIGC), which will be supported by AIATSIS, Winnunga Nimmityjah Aboriginal Health and Community Service, and local Aboriginal leaders.

As the Genome Biology Flagship node, we will establish the role of Indigenous reference genomes and panels of variation in the pathway to health benefit by:

- 1. empirically evaluating the required steps in the roadmap to secure and deploy reference resources
- 2. establishing reproducible analytical standards for genomic analysis using pan-human genome references accounting of Indigenous diversity
- 3. developing the future research agenda for the Network in this domain.

Research publications in *Nature* demonstrate the requirement of increasing the diversity representation in current databases. We completed genomic data collection in 2023 for all consenting participants (>500) and have implemented scalable analytical workflows using the Nextflow framework at the National Computational Infrastructure (NCI). The establishment of appropriate governance including local Indigenous leadership will be critical for advancing precision medicine and health outcomes for Indigenous communities in the ACT region and beyond.

Structural variation project

NCIG is a partner in a research project funded by MRFF that utilises Oxford Nanopore long-read sequencing technology to establish a comprehensive structural variation map for Indigenous Australians. The first phase of this research was published in *Nature* in 2023. We are establishing required analytical workflows at the NCI to scale this project and perform reanalysis using graph-based pangenomes to improve accuracy of variant calls. In addition, we initiated important discussions with the Machado Joseph Disease Foundation and the PROPHECY program to identify potential collaborations.

GRANTS AND PUBLICATIONS

Grants

- Genomics Health Futures Mission 2022 MRFF Genomics Health Futures Mission – Stream 2, June 2023 – May 2026, MRF2025085, Funding amount: \$1,973,206
 - Title: Integrated Genetic HealthCare Improving Access to Quality Genetic Services for Aboriginal and Torres Strait Islander Patients

Chief investigators:

Gregory Pratt, Julie McGaughran, Gareth Baynam, Elizabeth Palmer, Ray Mahoney, Kristen Nowak, Renee Williams, Alex Brown, Julie Rogers

- Associate Investigators: Louise Lyons, Amanda Collins-Clinch, Rebecca D'Souza, Tiffany Boughtwood, Hardip Patel, Christina Bernardes
- Genomics Health Futures Mission 2022 MRFF Genomics Health Futures Mission – Stream 5, June 2023 – May 2026, MRF2024837, Funding amount: \$2,959,806
 - Title: Advancing health equity for Indigenous Australians through pharmacogenomics: building an end-to-end discovery pipeline

Chief investigators:

Klaus-Martin Schulte, Alex Brown, Hardip Patel, Azure Hermes, Gareth Baynam, Graham Mann, Elizabeth Gardiner, Amee George, Ruth Arkell, Rita Ferreira, Nadine Hein

Publications

Ahmad, M., et al. (2023). "Uncovering the genetic and molecular features of Huntington's Disease in Northern Colombia." *Int J Mol Sci* 24(22).

Hermes, A., et al. (2023). "'We are Taking It Back to Our Homeland; We are Free to Move On': repatriation of blood samples to the Galiwin'ku Community." In Cressida Fforde et al. (eds), *Repatriation, Science and Identity*. Routledge, London

McWhirter R., et.al. (2023). "Community Engagement and the Protection-Inclusion Dilemma." *Am J Bioeth* 23.

Reis, A. L. M., et al. (2023). "The landscape of genomic structural variation in Indigenous Australians." *Nature* 624

Silcocks, M., et al. (2023). "Indigenous Australian genomes show deep structure and rich novel variation." *Nature* 624

Wagner, S., et al. (2023). "Gene expression of male pathway genes sox9 and amh during early sex differentiation in a reptile departs from the classical amniote model." *BMC Genomics* 24(1): 243.



COMMUNITY ENGAGEMENT

NCIG has been working closely with the Anindilyakwa people of the Angurugu and Umbakumba communities in the Groote Archipelago this year in partnership with the Anindilyakwa Land Council. The discussions in these communities have been productive and thoughtful, and families are steadily progressing towards decisions about the management of the samples of their loved ones.

NCIG was able to visit Galiwin'ku and Yarrabah to provide updates on the Centre's progress and discuss the publication of research papers. It was important to be able to connect with communities again about the Centre's work.



Azure Hermes and community leaders: Edith Mamarika, Mildred (Millie) Mamarika, Elaine Mamarika, Edna Bara, Colleen Mamarika and Hannah Moody at Groote Eylandt. Photo: Mark Hermes

NCIG has also begun initial engagement with communities including Galiwin'ku, Yarrabah and Groote Eylandt to introduce new projects, garner interest and align priorities, including for the work of PhD candidates (Rubi and Bridie) and for the Domain 1 project.

NCIG was also delighted to host visits to the Centre from representatives from Yarrabah, Groote Eylandt, Galiwin'ku and the Tiwi Islands this year.



Azure Hermes with Mayor Ross Andrews and Deputy Mayor Lucresia Willett. Photo: Tracey Nearmy



Left to right: Carly Conlan, Selina Puruntatameri, Andrew Howard, Allan Williams. Photo: Tracey Nearmy





Communities visited by NCIG representatives

National Indigenous Summer School

In December, NCIG hosted another cohort of the National Indigenous Summer School where First Nations students from around the continent participate in a showcase of STEM (science, technology, engineering, mathematics) activities over a week at the ANU. The students were engaged and thoughtful as they learned about the Centre and the work we do. Participants heard from our Deputy Director and First Nations PhD candidates, created DNA structures, and undertook a blood typing activity in the lab.



National Indigenous Summer School participants and NCIG staff. Photos: Luka Vertessy





	ANU NCIG Operational Funding	External Grant Funding	Philanthropic Donations
Open Balance	\$-	\$339,349.73	\$54,860.38
Income			
Operating & Project Grants	\$485,050.00	\$1,136,934.01	
Operating Expenses			
Salaries	\$270,439.69	\$435,663.70	
Equipment	\$36,104.95	\$-	
Travel and fieldwork	\$95,621.48	\$79,573.85	
Expendable research materials	\$4,714.45	\$93,178.34	
Consultancies	\$6,375.00	\$7,000.00	
Consumables	\$24,905.08	\$2,734.88	
Other expenses	\$29,887.35	\$3,670.98	
Grant Share to Others		\$131,499.50	
Net result	\$17,002.00	\$722,962.49	\$54,860.38

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Grant / Funding Source	Funding 2023	Remaining Period	Total Remaining Funding
MRFF Synergy - Domain 1	\$200,000.00	4 Years	\$800,000.00
MRFF Synergy - Domain 2	\$65,680.00	4 Years	\$262,720.00
MRFF Synergy - Domain 3	\$51,000.00	4 Years	\$204,000.00
MRFF GHFM S4 (Brown)	\$199,950.00	3 Years	\$599,850.00
MRFF GHFM S6 (Jiang)	\$90,000.00	1 Year	\$90,000.00
ANU Grand Challenge	\$150,000.00	2 Years	\$300,000.00
MRFF GHFM S3 (Newson)	\$80,000.00	4 Years	\$320,000.00
MRFF GHFM S6 (Deveson)	\$149,410.00	1 Year	\$149,410.00
MRFF GHFM S5 (Brown)	\$1,600.00	4 Years	\$6,400.00
NHMRC Idea Grant - (Patel)	\$798,709.00	2 Years	\$1,750,778.00
NHMRC Idea Grant - (Farlow)	\$336,752.00	2 Years	\$548,504.00
ANU JCSMR Operational Funding	\$320,000.00	2 Years	\$640,000.00
ANU DVCRI Operational Funding	\$150,000.00	2 Years	\$300,000.00
Grand total	\$2,593,101.00		\$5,971,662.00



GOVERNANCE BOARD

Meetings and attendance 2023

Board Member	Meeting 1 27 March 2023 via Zoom	Meeting 2 26 June 2023 via Zoom	Meeting 3 27 Sept 2023 via Zoom	Meeting 4 4 Dec 2023 ANU
Assoc Prof Glenn Pearson – Chair	\checkmark	\checkmark	\checkmark	\checkmark
Slade Sibosado		First meeting	\checkmark	\checkmark
Dr Lyndon Ormond-Parker	Retired at this meeting			
Dr Karla Canuto	First meeting	\checkmark	X Resigned	
Prof Gareth Baynam	Apologies	\checkmark	\checkmark	\checkmark
Prof Keith Nugent	\checkmark	\checkmark	\checkmark	\checkmark
Ms Erica Kneipp	\checkmark	\checkmark	Apologies	\checkmark
Prof Yvette Roe	\checkmark	Apologies	Apologies	\checkmark
Mr Benjamin Murray	\checkmark	\checkmark	\checkmark	\checkmark
Ms Janine Mohamed	Apologies	Apologies	\checkmark	Apologies



New appointments

Dr Karla Canuto, Associate Professor in Aboriginal and Torres Strait Islander Health at Flinders University joined the Board in January 2023 and resigned in August 2023.

Slade Sibosado, Telethon Kids Institute (TKI) Kimberley Operations Manager, joined the Board in June 2023.

Retirements

Dr Lyndon Ormond-Parker retired from the Board in early 2023 after serving on the Board since January 2019.

Glenn Pearson retired as Chair and from the Board at the end of 2023 after 5 years' service.

Operational strategies and planned activities 2024

The primary goal of NCIG for the upcoming year is to cultivate Indigenous leadership in the field of genomics, adhere to ethical standards in research, and improve health and well-being through advancements in genomic studies. Key priority areas include:

- Sustainability and Resource Management: Strengthen financial foundations to support NCIG's core infrastructure and operations. This includes managing the NCIG Collection and adhering to ethical research standards.
- Community Engagement and Capacity Building: Deepen engagement with Indigenous Australian communities and strengthen partnerships. This involves enhancing community consultation and integrating these perspectives into research and grant processes.
- Advancing Research in Indigenous Genomics: Lead and participate in high-quality research focusing on Indigenous genomics, contributing to the understanding and application of genomic sciences for the benefit of Indigenous Australians.
- Contributing to Precision Medicine: *Play* a pivotal role in ANU's Precision Medicine Flagship, leveraging genomics to develop precision medicine approaches and resources, with a focus on Indigenous populations.

In 2024, NCIG's focus is on advancing Indigenous leadership in genomics, improving health outcomes through research, and maintaining ethical governance and community involvement.

OUR TEAM

Director Prof Alex Brown

Deputy Director Assoc Prof Azure Hermes

Bioinformatics Lead Dr Hardip Patel

Project Manager/Board Secretariat Ms Prue Beckett (to March 2023)

Community Engagement Coordinator Ms Alice McCarthy

Data Manager Ms Jiaxin Yuan

Administration/Board Secretariat Ms Deb Marburg (from June 2023)

NCIG new team members

Kirat Alreja



Kirat Alreja joined the NCIG team as a Bioinformatics Support Officer in March 2023, immediately after completing his Bachelor of Advanced Computing (Hons) from the Australian National University in December 2022. During his studies, he specialised in machine

learning and undertook a bioinformatics computing project at the Research School of Biology as part of his Honours program. His role at NCIG marked the beginning of his professional journey, allowing him to apply the diverse skills acquired at ANU.

At NCIG, Kirat's primary focus has been on developing workflows for the manual curation of genome assembly within the AusARG (Australian Amphibian and Reptile Genomics) project funded by Bioplatforms Australia. This national initiative collaborates with universities, museums, and research institutions to generate genomic resources for Australia's unique native amphibians and reptiles, aiding in understanding their evolution and conservation. Kirat has contributed by conducting extensive experimentation on genome assembly workflows, implementing best practices through user-friendly scripts and workflows optimised for the NCI facility. His efforts have facilitated the genome assembly of *Bassiana Duperreyi*, *Tiliqua Rugosa*, *and Pogona Vitticeps*, and he aims to further adapt these processes for broader NCIG data applications.

Garrett Ball



Garrett Ball is an ANU student who joined the NCIG halfway through 2023. He is studying a double degree in law and science, majoring in cell and molecular biology. In 2023, Garrett worked on two significant projects at the NCIG.

He completed a biology research project under the supervision of Dr. Hardip Patel. This project investigated the genetics of chronic kidney disease in the Indigenous Tiwi Islands population. The research surrounded the analysis of structural variants present in this gene pool, with a particular focus on the relationship of these variants with loci associated with kidney function.

Garrett has also supported the community engagement work, led by Deputy Director Azure Hermes. Here, he contributed to key resources that aim to ensure all Indigenous Australians have the opportunity to access information about genomics in ways that suit them.

Garrett has a special interest in intersecting the social sciences with biology and he hopes to continue work in this area in the future. He aims to use his combination of skills to improve the well-being and quality of life of Indigenous Australians with further research and legislative advocacy.

Zahra Chew



Zahra joined NCIG in early 2023 to complete a research project as part of her undergraduate studies in the Bachelor of Philosophy (Honours) at the ANU. Supervised by Dr Hardip Patel, her work focused on investigating the cystic fibrosis transmembrane

conductance regulator locus in ancestrally diverse populations, with a view to facilitating more equitable diagnosis and treatment of cystic fibrosis. Using publicly available datasets incorporating genomic data from participants across the globe, the project highlighted the immense genetic and proteoform diversity that leaves diverse populations falling through the gaps of equitable disease management in Australia and necessitates more inclusive and considerate research practices. Zahra is both honoured and excited to work in a centre dedicated to improving access to informed and culturally sensitive healthcare through biomedical research and would love to continue working in the field after she graduates.

Carly Conlan



Carly Conlan is a dedicated professional with a diverse background. Recently joining NCIG as the Senior Technical Officer, she plays a pivotal role in the processing and management of NCIG biospecimens.

Prior to joining NCIG Carly

was an educator, working as the Program Facilitator for the Year 7 Integrated Maths and Science Program at St John Paul II College in Nicholls, ACT. Her passion for education and community outreach also led her to Timor-Leste, where she served as a STEM Pedagogy Advisor, delivering STEM professional learning to teachers in regional areas.

Carly's educational journey includes a Bachelor of Science Honours in Biochemistry and Molecular Biology from the Australian National University and a Bachelor of Arts/Bachelor of Science from the University of Queensland, where she earned commendations for high achievement in areas of both Molecular Archaeology and Biomolecular Research. With a strong foundation in education and a commitment to Indigenous health, Carly is excited to be a part of the NCIG community, and to make significant contributions to genomics, education, and community engagement.

Olivia Dalton



Olivia is a second-year medical student at the ANU School of Medicine and Psychology. She completed a Bachelor of Health Science at ANU with a minor in Australian Indigenous Studies. Olivia is passionate about Aboriginal and Torres Strait Islander health and is interested in

the integral role of public policy in health outcomes. She hopes to work as a Rural Generalist in the future which is a subset of General Practice specifically tailored to the rural environment.

Olivia is working with the NCIG team conducting research on the clinical genetics services available to Aboriginal and Torres Strait Islander peoples across the country. Her project involves mapping the services across Australia that have been developed specifically for and with Aboriginal and Torres Strait Islander people, identifying the benefits and barriers to these service models, and applying these to the mainstream public genetic services available. By producing an overview and brief analysis of the services that are currently available to Aboriginal and Torres Strait Islander peoples, this project will contribute to efforts to improve access to, and experiences of clinical genetic services.

Lynne El Hassan



Lynne completed her Honours project with NCIG in 2023 and graduated with a Bachelor of Science (Advanced) (Honours). Her thesis, titled "The use of a genetic differentiation metric (F_{sT}) to improve polygenic risk scoring across ancestries", explored a promising method of

improving a type of clinical tool that aims to predict a person's genetic risk of developing certain diseases. Importantly, the project's inclusion of Indigenous Australian samples revealed previously unexplored insights that may greatly inform how such a clinical tool is developed and implemented for our First Nations people.

Lynne presented her work at the XXIII International Congress of Genetics and was ultimately awarded First Class Honours.

Dr Michael He



With a 17-year career spanning academia, public service, and the non-profit sector, Dr He brings a wealth of experience to his roles as Manager of NCIG and Coordinator of the Australian Alliance for Indigenous Genomics (ALIGN) ACT Node.

Dr He's academic achievements include a PhD in Sociology from the University of Sydney, acquired in 2011 with Endeavour Program support, and his contribution as the author of a monograph published by Palgrave Macmillan in 2015. His tenure as Senior Project Manager at the National Aboriginal Community Controlled Health Organisation demonstrated his commitment to understanding the social determinants affecting the health and wellbeing of Aboriginal and Torres Strait Islander communities. His current role at NCIG resonates with his dedication to Indigenous communities and advances his professional and personal growth. Dr He is also passionately committed to fostering diversity, inclusion, and equity in the workplace and actively seeks to effect positive change through collaborative and innovative methods.

Dr He proudly serves as a Jawun secondee, with placements at both the Moorundi Aboriginal Community Controlled Health Services and the Ngarrindjeri Lands and Progress Aboriginal Corporation, located within the Ngarrindjeri Nation. Beyond these contributions, back in Ngunnawal and Ngambri Country, he shares his project management skills as a mentor in the ANU Professional Staff Mentoring Program and volunteers his time as a Justice of the Peace in the ACT.

Dr Kosar Hooshmand



Kosar Hooshmand is a Bioinformatics Officer at NCIG, appointed in March 2023, with expertise in designing workflows and statistical methods for variant and benchmarking analysis using the T2T and hg38 reference genomes.

Before her role at NCIG, Kosar gained extensive experience in bioinformatics as an analyst at Pacific Analytics and as a bioinformatician and platform consultant at GenieUs Genomics. She was also a fully funded visiting research student at the Brain and Mind Centre through Tarbiat Modares University, where she developed a strong foundation in analysing various data types, including whole genome sequencing and RNA sequencing using cutting-edge tools and machine learning techniques.

Kosar is currently dedicated to exploring the intricacies of population genetics, with a particular focus on conducting in-depth studies on genetic variations within the Indigenous Australian population. She is determined to illuminate their unique genetic profiles and deepen our understanding of their valuable contributions to the human genetic diversity landscape.

As part of her research activities, Kosar performs variant analysis for Indigenous Australian samples.

With her expertise in bioinformatics and dedication to unravelling the complexities of population genetics, Kosar is an asset to the NCIG team, connecting cuttingedge research with real-world applications.

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Dr Hyungtaek Jung



Dr Hyungtaek Jung joined the National Centre for Indigenous Genomics with the primary objective of analysing extensive biological data produced through cuttingedge sequencing technologies. His role involves overcoming the current limitations of human reference

genomes by creating comprehensive genomic resources tailored to the Indigenous Australian population. This includes generating haplotype-resolved telomereto-telomere (T2T) Indigenous genomes, compiling a comprehensive catalogue of genetic variations anchored to Indigenous-T2T genomes and facilitating clinical genomics applications for Indigenous communities.

Dr. Jung brings with him a wealth of successful experience collaborating with diverse national and international laboratories. His expertise spans life and clinical sciences, aquaculture and agriculture, molecular ecology, genomics, and bioinformatics.

Joel Keen



Joel Keen is a Gomeroi scholar and educator. As a relatively new member of the NCIG team, Joel continues the active pursuit of protecting and promoting the integrity of Indigenous Knowledges, and constructing a strengthbased Indigenous genomics framework within which

Indigenous Knowledges can be safely utilised. He is additionally involved in a cross-institutional project, aimed at improving the educational experience of international university students in Australia through the design and delivery of a localised Aboriginal orientation program.

With a particular focus on the sociology of knowledge, his Honours thesis established current conceptualisations of racial equality as paradoxical, self-contradictory, and unachievable in the contemporary societal super-structure. He has published on the necessity of amplifying Indigenous voices and the unrivalled validity of place-based knowledge. His PhD is an extension of this work, performing an interrogation of the Westernised relationship with knowledge.

Deb Marburg



Deb joined NCIG in June 2023 on a part-time basis and then converted to full-time in mid-December. An alumna and long-term employee of ANU, Deb brings with her a wealth of knowledge about the administrative practices of the University. Deb started working within the Student

Administration team while studying at ANU in the 1990s. Since then she has worked in the Research Initiatives and Infrastructure team, the College of Business & Economics, the College of Arts & Social Sciences, ANU Global Programs and has been executive assistant to the Deputy Vice-Chancellor (Research & Innovation). Outside of ANU, Deb previously worked in the travel industry and has a passion for seeing the world and meeting new people. She also has 12 years' experience in the fashion industry working to help women feel comfortable and supported.

Deb provides secretariat support to the external NCIG Board and supports the NCIG Director and Deputy Director to implement the Centre's objectives and activities.

Dr Hadi Nazem-Bokaee



In late July 2023, Hadi became part of NCIG to enhance the value of NCIG Genomic Data Collections and their clinical implementation for NCIG communities. Drawing upon over a decade of combined professional experience in scientific research and operational domains, Hadi's

research centres on employing advanced numerical and statistical techniques in bioinformatics and computational biology. His research revolves around exploring genomics, deciphering structurally complex genetic variations in human genomes, and understanding their connection with protein dysfunctions and the onset of diseases.

Prior to joining NCIG, Hadi held positions as a research scientist at CSIRO, Australia, and as a research scholar at Penn State University in the United States. In these roles, he led the application and advancement of computational models and mathematical methods in the systems biology field, with the goal of engineering biological systems to achieve targeted phenotypic outcomes or uncovering the inherent genomic and metabolic capabilities within these systems. Additionally, Hadi held the position of a pharmaceutical engineer at the Institute of Pasteur Iran, where he played a key role in the large-scale production of recombinant protein drugs.

Hadi holds a Bachelor of Science in Chemical Engineering (Iran University of Science and Technology), a Master of Science in Biochemical Engineering (Sharif University of Technology, Iran), and a PhD in Biological Engineering (Virginia Tech, USA).

Dr Lyndsay Newett



Lyndsay Newett is a mixed-methods sociologist; she began her role as a Research Fellow at NCIG in March 2023. Prior to her role at NCIG, Lyndsay worked across several interdisciplinary research projects at the University of Tasmania, and as a Research Fellow

at the University of Tasmania's Centre for Law and Genetics, where she assisted with a project examining public trust in Human Research Ethics Committees within the context of medical research involving genomic data sharing. Additionally, Lyndsay was employed as a Senior Research Associate at Bellberry Limited, where she helped with research exploring communication and consent practices in relation to Australian clinical trials research.

In her new role at NCIG, Lyndsay will be working in conjunction with Centre staff, students, and community partners to explore how people feel about the ways genomics and precision medicine can be conducted, and to ensure appropriate, and best practices, are developed and engaged. In this role, Lyndsay is looking forward to learning more about Indigenous ways of thinking and approaches to research, and doing what she can to help improve how research is undertaken and experienced by those involved.

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NCIG ANNUAL REPORT 2023

ACKNOWLEDGEMENTS

Research collaborators

- Professor Gareth Baynam University of Western Australia
- Dr Ashley Farlow University of Melbourne, Australian National University
- Associate Professor Misty Jenkins Walter & Eliza Hall Institute
- Dr Simon Jiang ANU
- Professor Stephen Leslie University of Melbourne
- Associate Professor Bastien Llamas University of Adelaide
- Associate Professor Brendan McMorran ANU
- Dr Yassine Souilmi University of Adelaide
- Dr Ira Deveson Garvan Institute of Medical Research
- Dr Matthew Silcocks University of Melbourne
- Professor Stephen Leslie University of Melbourne
- Dr Rebekah McWhirter Deakin University
- Ms Louise Lyons South Australian Health & Medical Research Institute

Indigenous communities and organisations

- Anindilyakwa Land Council
- Kimberley Aboriginal Law and Culture Centre, WA
- Kimberley Aboriginal Medical Service, WA
- Ninti One, NT
- SING Australia
- Tiwi Land Council, NT
- Top End Human Research Ethics Committee, NT
- Yalu Aboriginal Corporation
- Yarrabah Aboriginal Shire Council
- Gurriny Yealamucka Health Service
- The Walpiri people of Yuendumu and Lajamanu

Technical service providers

- The Australian Phenomics Facilities, ANU
- Biomolecular Resource Facility, ANU
- Information Technology Services, ANU
- Kinghorn Centre for Clinical Genomics (Garvan Institute of Medical Research)
- National Computational Infrastructure, ANU
- Ramaciotti Centre for Genomics, UNSW







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