



NCIG

NATIONAL CENTRE
FOR INDIGENOUS
GENOMICS



ANNUAL REPORT 2021

ABOUT NCIG

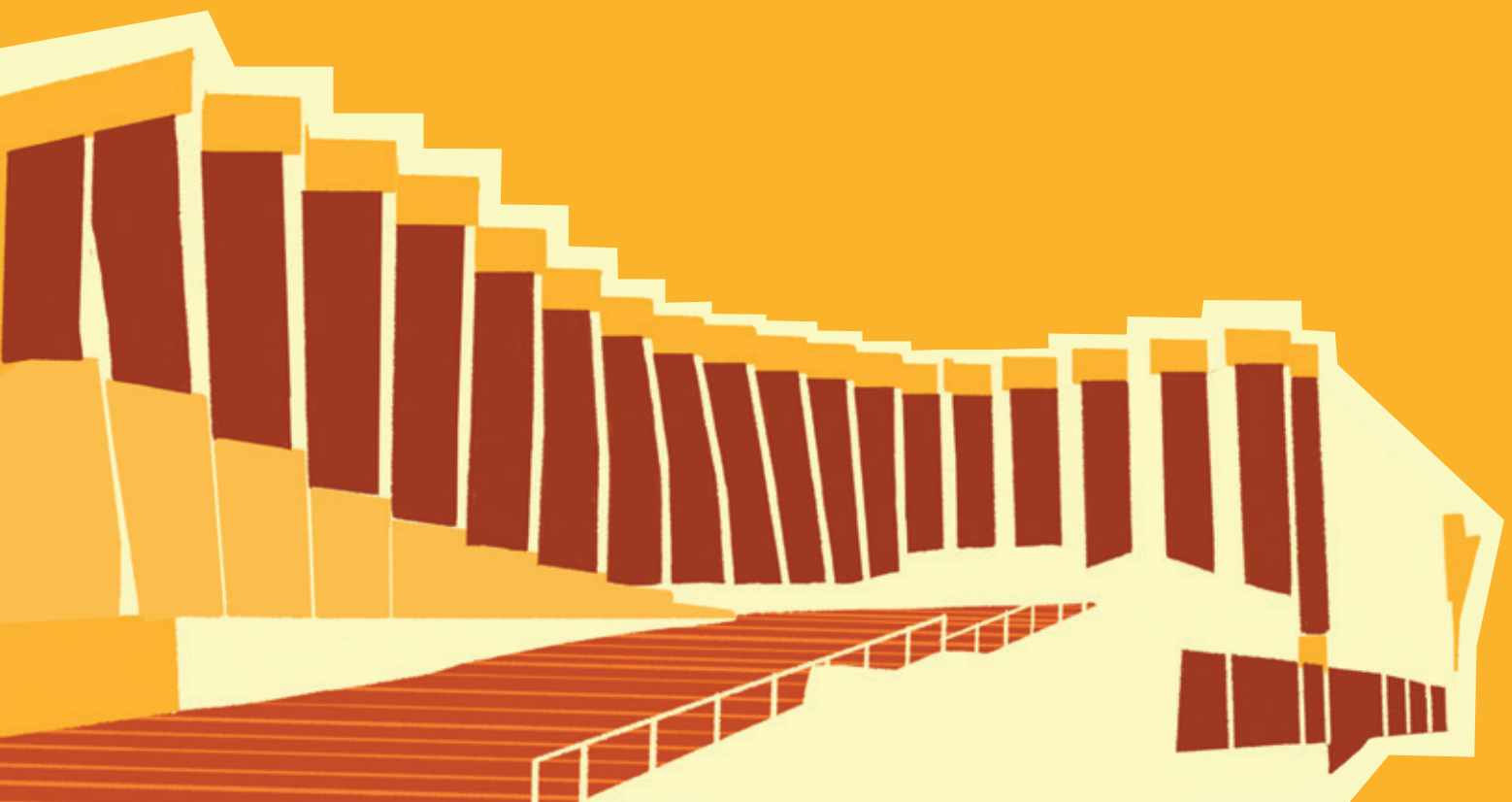
The National Centre for Indigenous Genomics (NCIG) was created in 2013 to manage and expand a collection of 7,000 historical Indigenous blood samples – now known as the NCIG Collection – held at the John Curtin School of Medical Research, and to develop a research reference resource from the DNA in the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

NCIG is overseen by a Governance Board constituted by statute with an Indigenous majority and Indigenous chairmanship. The Board is the custodian of NCIG Collection and sets the strategic goals, policies and operational plan for the Centre. For the first time, Indigenous Australians have a mechanism by which they can contribute to and benefit from genomic research with full agency, in a climate of understanding and respect.

NCIG rests on solid foundations of Indigenous support and governance; community consultation; a sound scientific research program; and a clear understanding of the technological platforms required to deliver and sustain Indigenous genomic inclusion. Under Indigenous governance, NCIG's focus is facilitating genomic research that provides benefit to Indigenous Australians.

NCIG's approach to community engagement and consultation is built on truth and time. Everything is open. Nothing is rushed. It accommodates Indigenous knowledge and ways of knowing into genomic science. From this respectful and creative space, great solutions emerge. Given the opportunity, the Indigenous communities working with NCIG have found pathways that navigate between Indigenous cultural belief systems and modern genomic science.

Together, NCIG and Indigenous Australians have created a research model for the use of DNA, the most sensitive kind of material imaginable, and in doing so are bringing the potential of genomic research to bear on the heavy burden of ill health borne by First Australians.



CONTENTS

From the Chair	2
From the NCIG Leadership Team	3
A nation-leading role	4
The year in review	6
Galiwin'ku Burial Poles	11
A coordinated national approach to Indigenous genomics	12
Committee membership and other community service	14
Research publications	15
Invited talks, conference presentations, interviews	16
Competitive funding awarded	17
Governance Board	18
2021 financial snapshot	20
NCIG Team	22

FROM THE CHAIR

Participating in countless online meetings this year from the relative safety of Western Australia, I reach the end of 2021 to salute my 'over-east' colleagues for their resilience and achievements in Year 2 of Covid.



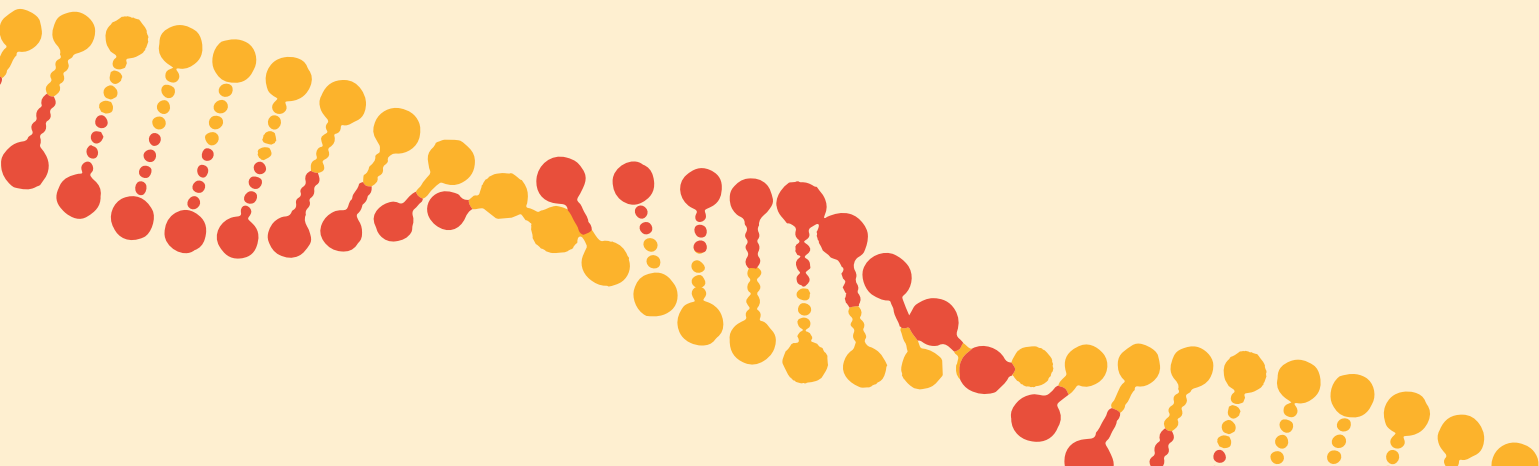
Like so many others, the team at the National Centre for Indigenous Genomics has persevered through the difficulties and uncertainties that have characterised this moment in history. For the second year in a row the team was unable to visit Aboriginal communities, with state borders shut for long periods. And yet, it has been a year of steady achievement. NCIG has emerged from this year stronger, focused and ready to move forward.

It was a highlight of my life to visit the community of Galiwin'ku in 2019 and a great disappointment to be confined to Western Australia when esteemed Galiwin'ku community leaders visited Canberra to participate in an ANU-led Indigenous Genomics Roundtable and to unveil duplicate burial poles on 27 May 2021.

On that day, genomic and medical research scientists, government officials and university leaders gathered face-to-face, and via the ubiquitous Zoom, in the company of our close partners from Galiwin'ku. We spoke about the imperative for our nation to empower Indigenous Australians to engage in and lead genomics research, and its translation into health benefit, for Indigenous peoples and for all. This important discussion was followed by a moving traditional ceremony to unveil burial poles.

As I write this in December 2021, my fellow Board members and the NCIG staff are trying to find a sense of balance after the unexpected passing of Ross Wunungmurra (Mr M), an outstanding Aboriginal leader and Galiwin'ku elder, without whom our close research collaboration and deep friendship with the community would not have been possible. We mourn the loss of a very fine man. NCIG will never stop working towards unlocking the benefits of 'precision' medicine for Aboriginal people. The commitment of Mr M and others like him remind us of our purpose.

Glenn Pearson
Chair



FROM THE NCIG LEADERSHIP TEAM

It has been a pleasure to see long-term work by NCIG's staff and research partners come to a hard-won maturity towards the end of 2021. The imminent completion of seminal research, described in this Annual Report is a credit to all involved.

The retirement of NCIG's founding Director triggered new management arrangements in 2021. Azure Hermes stepped up to the role of Deputy Director and spearheaded a year-long, energetic dialogue and analysis to identify NCIG's future direction. NCIG's research agenda and financial sustainability have been re-set, and its place in the emerging national Indigenous genomics coalition is established.

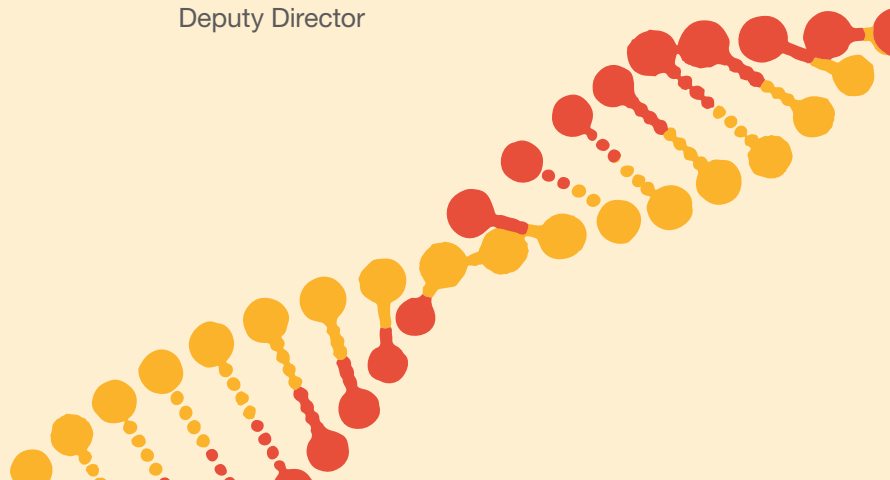
We are buoyed by the knowledge that the work NCIG is doing will make a difference as it is disseminated through the medical research and clinical community. The insatiable curiosity of scientists drives discovery, but overlaying this is the distinguishing characteristic of NCIG: the expectation that scientific effort be focused specifically on *delivering benefit* to Indigenous Australians. It is the vision of making a difference that has kept us focused this year.



Graham Mann
Interim Director



Azure Hermes
Deputy Director



A NATION-LEADING ROLE

The National Centre for Indigenous Genomics was established at The John Curtin School of Medical Research (JCSMR) in 2013.

JCSMR was one of the founding Schools within The Australian National University (ANU). The ANU was established to be a resource for the nation by conducting research which will transform the lives of people across Australia. The power of education and research is its capacity to change the way we understand the world and make it a better place for all people.

We can know this, and also know that there is an ugly history of science being used as a weapon against First Nations people. NCIG was created and structured to be a deliberate intervention in legacy research models. The way NCIG works is as innovative as its scientific discoveries.

Its governance model and operating protocols are built on the principle that Indigenous people must be engaged at all levels of programs that specifically target their health, wellbeing and communities. This means involving Indigenous people in all decision-making processes.

Genomics has the potential to enable treatment for diseases like cancer or diabetes to be precisely calibrated to an individual person's genomic makeup. If so, we can make radical improvements to patient experience, and save lives. At the population level, First Nations communities experience more disease and have poorer health outcomes. So, the promise of genomics is particularly important to Aboriginal and Islander people.

Transformational results are possible by investing funds and effort in matters which Indigenous people identify as their priority. An integrated collaborative national ecosystem is required, encompassing: genomic research, both fundamental and disease-focused; advanced platforms for data management, governance and sovereignty; clinical translational pipelines; and community engagement and consultation. NCIG proudly contributes on all fronts.

"I understand that people will read the story and feel in two minds about it because genomics and genetics and DNA is a really taboo subject for Indigenous people because we've got politicians and media personalities out there always trying to define ... Aboriginality," she said.

"If we really want to Close the Gap, there has to be recognition genomics is a serious player in this space.

"Right now with NCIG we want to work collaboratively with Indigenous researchers and form a national agenda where we are leading the conversation, leading the push, and leading the research when it comes to genomics.

"In order to do that we really need to have a conversation with government about, not only just setting up that national agenda, but funding it properly. We as researchers, we're going to push ahead and do it, but we'd really love it if [government] was standing next to us."

Azure Hermes, speaking to the National Indigenous Times, 27 May 2021



We are determined in our resolution to do the right thing, and we see that commitment as part of our unique responsibility — and privilege — as the national university.

Vice Chancellor Professor Brian Schmidt, 27 May 2021.



THE YEAR IN REVIEW

Because Aboriginal and Torres Strait Islander cultures are the longest surviving in human history, and because those cultures are so strongly connected to place, genomic diversity between Indigenous communities in Australia is unparalleled. Our seminal research projects are reaching finalisation, and will be the platform for ambitious new directions.

Population variation project

Over the last 4 years NCIG and its research collaborators (Professor Stephen Leslie and his colleagues at the University of Melbourne) have analysed the genomes of 160 individuals from four Indigenous Australian communities from distinct regions of Australia. The purpose of this study is to identify the extent to which genomic variation in Indigenous Australian communities differs both between communities, and from variation in other parts of the world in ways that impact the application of genomic data in medical research and clinical practice. About one quarter of the genetic variation observed in the Indigenous samples is absent from global reference datasets, and nearly half of it is private to the individual communities studied. Related research completed by NCIG indicates that 1.5% of the human reference genome has significant bias. This must be corrected to improve the accuracy and effectiveness of genomic analysis for Indigenous Australians.

In 2021, NCIG PhD candidate Mr Renzo Balboa extended existing genomic analyses to detect larger structural variations in Indigenous Australian genomes. This research shows that Indigenous Australians have the most structural variation diversity of all population groups outside Africa. This is a significant advance, providing clues to the nature and processes of structural variation in human genomics that will improve our understanding of the functional consequences of structural genomic variation in health and disease.



Mr Renzo Balboa

Reference genome (de novo assembly) project

It is well-established in the scientific literature that the current human reference genome is incomplete, contains errors and that its data is biased towards people of European ancestry. This renders it suboptimal for genomic studies of Indigenous and other ancestrally diverse peoples. NCIG and its research collaborators (Dr Ashley Farlow and his colleagues at the University of Melbourne) have begun to create high quality Aboriginal Australian Reference Genomes to address these limitations.

In the first phase, high quality DNA from one individual from each of four communities was used to generate long read DNA sequence data. These long read data were assembled using state-of-the-art software to reconstruct the genome of each individual. Very long stretches of chromosomes extending up to 104 million base pairs were assembled. These Phase 1 genomes have filled 90 gaps in the human reference genome and revealed unique sequences that were not previously present. Critical evaluation of the gain is underway.

Human Pangenome Reference Consortium

Importantly, NCIG became an external collaborator with the Human Pangenome Reference Consortium (HPRC), which is leading efforts to sequence 350 diverse genomes from around the world by 2025. The linear reference genome in current use will be replaced with a graph-based reference genome which will more accurately represent human diversity and provide a better template for genomic analysis.

HPRC, in partnership with the Global Alliance for Genomics and Health (GA4GH), will develop standards for the use of the new pangenome reference. NCIG is contributing to the development of these standards, especially for data access and reuse. Based on this collaboration and the importance of staying connected with international agencies, the NCIG Board approved a project to create the first telomere-to-telomere genome for Indigenous Australians. The generation of high-quality ultra long read sequence data for four individuals commenced during 2021.

Genomic structural variation project

A pilot project was conducted in 2021 to assess the suitability of Oxford Nanopore Technologies (ONT) sequencing technology for addressing limitations in standard short read sequence data. The investigation was led by Dr Hardip Patel (NCIG) and Dr Ira Deveson (Garvan Institute), and his team, Dr Andre Martins Reis, Ms Jillian Hammond, Dr Hasindu Gamaarachchi and Mr James Ferguson.

With funding from the Garvan Institute's Kinghorn Foundation, samples from 16 individuals were sequenced using ONT. To date, 44 terabytes of data has been generated. The high-performance compute and storage capability of the National Computation Infrastructure was used to handle the quantity and complexity of data. Preliminary results indicate that this technology delivers significant additional benefit when seeking to describe structural variation, compared to short read data. The catalogue of variation emerging from the ONT project will accelerate the accurate description of variation in Indigenous Australians.

A second iteration of the project is being planned for 2022 and beyond, to sequence additional individuals, and to evaluate benefits in rare disease diagnosis.

This project has been a practical demonstration of a successful partnership between NCIG and an external research group. It will guide the way data and samples are managed under Board oversight in the future.



Dr Hardip Patel

A family tradition

Dr Ira Deveson, co-leader of the ONT pilot project, is the grandson of the founder of the NCIG Collection, Professor Bob Kirk.

Professor Bob Kirk was the person responsible for collecting most of NCIG's historical biological samples. The collection began in the late 1950s when he was at the University of Western Australia and was continued by him when he joined the Human Biology Department at JCSMR, ANU in 1967 until his retirement in 1986.

He was a scientist of wide and varied interests, but broadly, he was interested in studying diversity in human populations, in particular, Indigenous populations of Australia and Papua New Guinea.

The ANU formed the Indigenous samples into a managed collection under the governance and custodianship of the National Centre for Indigenous Genomics in 2013. At the launch, Professor Kirk's daughters, Ira's mother Pip Deveson, and his aunt Ann Evers, were among the guests of honour. Ms Deveson, based at the ANU, is an ethnographic filmmaker working in the area of visual anthropology.

Eight years on, NCIG is working with a third generation of the Kirk family. Dr Ira Deveson is the Head of Genomic Technologies at the Kinghorn Centre for Clinical Genomics at the Garvan Institute of Medical Research. Like his grandfather, Dr Deveson is a scientist with many interests. In addition to his work with NCIG to enrich the quality of its Indigenous genomic data, Dr Deveson and his team are working on other important contemporary research questions. Recently, Dr Deveson led an important study to evaluate the analytical performance of five commercially-available assays used for tumour DNA sequencing, important in the fight against cancer. And, currently he is tracing the genomic evolution of coronavirus.

Pip Deveson & Ann Evers standing next to photo of their father, Professor Bob Kirk at the NCIG launch, JCSMR, 2014



Dr Ira Deveson





Genomic data platform

Throughout 2021 work occurred at the National Computational Infrastructure to develop a secure, supported data management system for scalable storage and management of Indigenous genetic data, incorporating FAIR and CARE data management principles. The platform is being designed to allow Indigenous data sovereignty principles to be applied to genomic data for the first time.

In broad terms the scope of works has been to develop:

- a scalable Mediaflux platform to store current NCIG genomic data holdings and meet NCIG's future needs, and
- documentation describing the technical build, operational procedures and policy for the platform delivered.

The project is well advanced, and the substantial contribution being made by the National Computation Infrastructure is acknowledged with appreciation.

In 2021 NCIG was awarded 5 million computing service units under the National Computational Merit Allocation Scheme and 4 million service units under the ANU Merit Allocation Scheme. The value of these combined allocations was \$360,000. In addition, with the generous support of the JCSMR Director and the Office of the Deputy Vice Chancellor Research and Innovation, NCIG's data storage allocation was increased from 250Tb the previous year to 400Tb in 2021.

Since 2018 NCIG has received 16.9 million service units (1 service unit ~ 1 CPU hour). NCIG data storage has grown to 340Tb of storage on the fast file system (gdata) and 100Tb on the archival file system (massdata).

At the end of 2021 NCI and ANU together confirmed a commitment to NCIG of 10 million service units for the next five years and 500Tb of storage facility to manage growing data collections.

ANU Indigenous Health and Wellbeing Grand Challenge

The goals of this project are described in the Case Study. In 2021, a data management project officer was employed to improve the interoperability between NCIG records and data and the software platforms required for their storage, management and use.

CASE STUDY

Why a genomic data platform is needed

Tiwi renal disease project

Tiwi Islanders have an exceptionally high prevalence of chronic kidney disease (CKD) compared to other Indigenous Australians and non-Indigenous communities, even when adjusting for major risk factors such as hypertension and diabetes. CKD is also associated with chronic conditions such as heart disease, cardiac failure, stroke and death, and the impacts on Tiwi Islanders' physical and psychosocial health, lifestyle and capacity are profound. CKD and renal failure are the most common cause of illness and death on the islands. As a result, it is recognised as a priority health issue by the Tiwi Islanders and a partnership has been established and maintained for more than 20 years between the Tiwi Land Council and the project team to address these concerns.

With past support from the National Health and Medical Research Council, 750 participants were recruited, representing more than 20% of the Tiwi population, from which measures of kidney function and other chronic diseases were obtained, as well as DNA, to perform a genetic screening study of the participants. Whole genome sequences for 170 of these participants have been generated and tested for genes that may cause or increase their risk of CKD.

In 2020 the NCIG team partnered with the McMorran and Jiang Groups from JCSMR and received funding from the ANU Indigenous Health and Wellbeing Grand Challenge initiative to continue this work. The project is applying genetic and genomic methods to investigate the causes of the disease, underpinned by a MediaFlux data management platform installed under licence at the National Computational Infrastructure, recruitment of ANU Indigenous research students, and collaborations with the University of Queensland, Queensland University of Technology and Royal Darwin Hospital.

Associate Professor Brendan McMorran and Dr Simon Jiang are in partnership with NCIG to conduct the ANU Indigenous Health and Wellbeing Grand Challenge Tiwi Chronic Kidney Disease Project, 2020–2025.



GALIWIN'KU BURIAL POLES



Auntie Matilda House conducts a smoking ceremony. ANU Vice Chancellor Brian Schmidt and JCSMR School Director, Graham Mann bring the Yirritja and Dhuwa samples from JCSMR. Credit Jamie Kidston/ANU.

In the NCIG 2019 Annual Report¹, we shared the story of these samples, the highlight of which was a spectacular ceremonial acknowledgement of the samples upon their return to Galiwin'ku, and the unveiling of burial poles in the community's central square.

In a fortuitous window between lengthy lockdowns, during Reconciliation Week in May 2021, the ANU Chancellor, the Honourable Julie Bishop, and the ANU Vice Chancellor, Professor Brian Schmidt, hosted an executive briefing and Indigenous genomics roundtable before unveiling duplicate Burial Poles at JCSMR.

A party travelled from Galiwin'ku to participate in the roundtable and to lead the ceremonial event that followed at JCSMR.

These interesting and at times deeply moving events, attended by around 100 invited guests, mark the Galiwin'ku community's strong connection with the University and their ongoing commitment to research in collaboration with the University.

In the final weeks of 2021 NCIG received the devastating news that its highly esteemed friend and colleague, community leader and Chair of Yalu Aboriginal Corporation, Ross Wunungmurra, passed away. The words and images used in this report respect Yolgnu protocols by not using Ross' traditional name, nor his image.

Nearly 20 per cent of the samples in the NCIG Collection were gathered from people of Galiwin'ku (Elcho Island, Eastern Arnhem Land, 550km East of Darwin) in 1968.

In 2018, NCIG sought agreement from the people of Galiwin'ku to open a consultation about the future of the samples. Working with Yalu Marnggithinyaraw Indigenous Corporation (Yalu), a representative community body, a joint plan for the management and use of the samples was developed. After an innovative consultation process, the community decided to allow sequencing of the samples of deceased people followed by repatriation.



¹ 2019 Annual Report available on the NCIG website at: ncig.anu.edu.au/about/annual-report

A COORDINATED NATIONAL APPROACH TO INDIGENOUS GENOMICS

In October, the Australian National University announced a strategic partnership with the Telethon Kids Institute. The appointment of acclaimed Aboriginal researcher and clinician, Professor Alex Brown, as Professor of Indigenous Genomics is welcomed by NCIG. Professor Brown has stepped down from his role as member of the NCIG Board, and now provides strategic and research advice to NCIG as a member of the ANU research community.

Professor 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 program of work planned under the grant is spread across five research domains and eight research nodes across Australia. NCIG personnel are working in Domains I, II and IV.



Professor Alex Brown

DOMAIN I

In partnership with Indigenous communities, Domain I will explore the cultural, ethical, social and legal issues (C+ELSI) relating to genomics research and provide a roadmap for Indigenous leadership and inclusion in genomics. Domain I includes the evaluation of community engagement and governance strategies with the aim of supporting communities in decision making processes relating to consent and data sovereignty. The team of Chief Investigators include NCIG's Azure Hermes and Dr Sharon Huebner with Associate Investigators Dr Rebekah McWhirter from Deakin University and Mr Yarlulu Thomas from the University of Western Australia.

DOMAIN II

Examines how genomic information is interpreted, incorporated and translated meaningfully into the lives, experiences, and healthcare of individuals and communities. It will produce 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. The team of Chief Investigators include NCIG's Professor Simon Eastaugh with Dr Daniel MacArthur from the Garvan Institute and Dr Kalinda Griffiths from the University of New South Wales. Dr Hardip Patel (NCIG) is an Associate Investigator.



DOMAIN III Will create high-quality reference genome sequences that represent Indigenous Australian diversity; create a variant reference database to improve the diagnosis of rare diseases; and functionally annotate novel variants for further study.

DOMAIN IV Will undertake genomic, epigenomic, transcriptomic, proteomic, metabolomic, lipidomic and immune characterisation to develop a comprehensive assessment of multi-omics signatures, for further study. Chief Investigators include JCSMR Director and NCIG Interim Director, Professor Graham Mann, and ANU Professor of Indigenous Genomics, Alex Brown (Lead CIA)). The Associate Investigator is Associate Professor Misty Jenkins from the Walter and Eliza Hall Institute (WEHI), former Board Chair of NCIG.

DOMAIN V Will create and translate best-practice integrated genomics health care, through a focus on health literacy, community engagement and the development of a culturally and scientifically capable Indigenous genomics workforce.

COMMITTEE MEMBERSHIP AND OTHER COMMUNITY SERVICE

Azure Hermes

Aboriginal and Torres Strait Islander Advisory Group on Genomics, Commonwealth Department of Health.

ANU 2025 Strategy Roundtable (invited contributor)

Galiwin'ku Research Intake Committee, Yalu Marnggithinyaraw Indigenous Corporation.

Global Alliance for Genomics and Health Equity Diversity and Inclusion Advisory Group, East Asia/Pacific/Americas.

Indigenous Advisory Committee, Australian Bureau of Statistics.

Intergenerational Health and Mental Health Study Biomedical Topic Advice Group, Commonwealth Department of Health.

Indigenous Advisory Committee, Undiagnosed Disease Network.

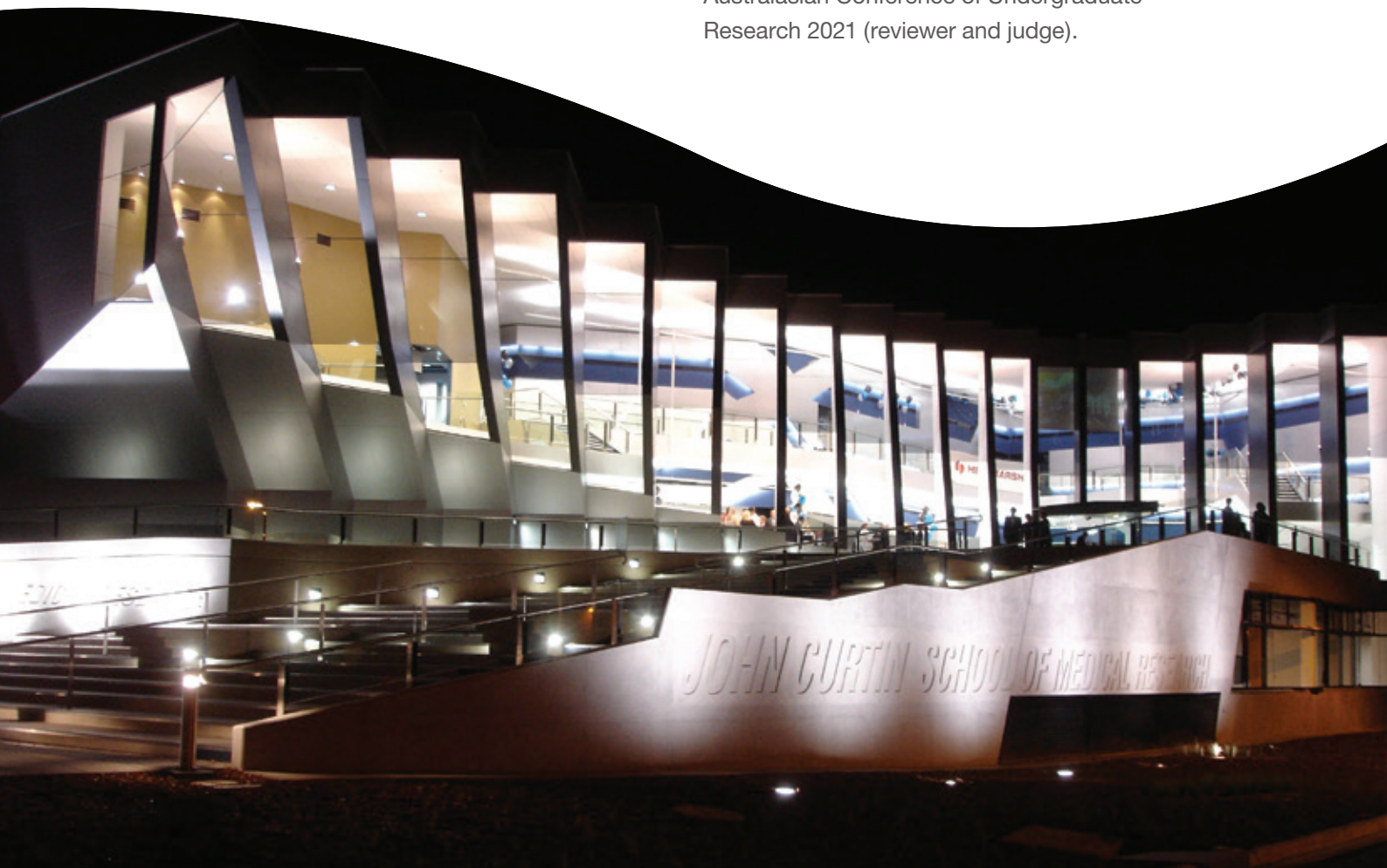
McKenzie's Mission Indigenous Advisory Board.

SING organising committee.

Hardip Patel

Australian Bureau of Statistics stakeholder consultation for Privacy Impact Statement (invited contributor).

Australasian Conference of Undergraduate Research 2021 (reviewer and judge).



RESEARCH PUBLICATIONS

A.J. Shankar, S. Jadhao, W. Hoy, S. Foote, **H.R. Patel**, V. Scaria, B. McMorran and S.H. Nagaraj. *Pharmacogenomic analysis of a genetically distinct Indigenous population*, The Pharmacogenomics Journal. (Accepted for publication). 2021. DOI: 10.1038/s41397-021-00262-4

A. McCartney, J. Anderson, L. Liggins, M.L. Hudson, M. Anderson, B. TeAika, J. Geary, B. Cook-Deegan, **H.R. Patel**, A.M. Phillippy. *Balancing openness with Indigenous data sovereignty – an opportunity to leave no one behind in the journey to sequence all of life*. 2021. DOI: 10.2139/ssrn.3922996.

P.D. Waters, **H.R. Patel**, A. Ruíz-Herrera, L. Álvarez-González, N.C. Lister, O. Simakov, T. Ezaz, P. Kaur, C. Frere, F. Grutzner, A. Georges, J.A.M. Graves. *Microchromosomes are building blocks of bird, reptile and mammal chromosomes*. Proceedings of the National Academy of Sciences. 2021. DOI: 10.1073/pnas.2112494118

T.W. McInerney, B. Fulton-Howard, C. Patterson, **D. Paliwal**, L.S. Jermini, **H.R. Patel**, J. Pa, R.H. Swerdlow, A. Goate, **S. Easteal**, S.J. Andrews, for the Alzheimer's Disease Neuroimaging Initiative. *A globally diverse reference alignment and panel for imputation of mitochondrial DNA variants*. BMC Bioinformatics. 2021. DOI: 10.1186/s12859-021-04337-8.

D.M. Prabhakaran, **H.R. Patel**, S.S.K. Chandrika, S. Thomas. *Genomic attributes differ between Vibrio parahaemolyticus environmental and clinical isolates including pathotypes*. Environmental Microbiology Reports. 2021. DOI: 10.1111/1758-2229.13000.

Y.Li, **H.R. Patel**, Y. Lin, *Kmer2SNP: Reference-free heterozygous SNP calling using k-mer frequency distributions*. Methods in Molecular Biology. Book Chapter. 2021. Accepted for publication.

J.A. Chu-Tan, Z.P. Feng, Y. Wooff, A.V. Cioanca, U. Schumann, R. Aggio-Bruce, **H. Patel**, M. Rutar, K. Hannan, K. Panov, J. Provis, R. Natoli. *Functional microRNA targetome undergoes degeneration-induced shift in the retina*. Molecular Neurodegeneration. 2021. DOI:10.1186/s13024-021-00478-9

P.A. Mateos, R.F. Balboa, S. Easteal, E. Eyra, **H.R. Patel**. *PACIFIC: A lightweight deep-learning classifier of SARS-CoV-2 and co-infecting RNA viruses*. Scientific Reports. 2021. DOI:10.1038/s41598-021-82043-4

D. Featherstone, C. Stuchbery, **S. Huebner**, L. Ormond-Parker, A. Dodd. *Archiving First Nations Media: The race to save Australia's community media and cultural collections*. Australian Aboriginal Studies. Issue 1, 2021.

C. Stuchbery, B. Gosper, **S Huebner**, A. Dodd, L. Ormond-Parker. *Above and Beyond Broadcasting: a study of First Nations Media and the Covid-19 Pandemic*. First Nations Media Australia. 2021.

A. Hermes, **S. Huebner**, S. Easteal, L. Ormond-Parker, A. McCarthy, R. Gundjarranbuy, R. Wunungmurra. *'We are taking it back to our homeland; We are free to move on': Aboriginal participation in genomics research*. C. Fforde, L. Ormond-Parker (eds.) Indigenous DNA, Craniometrics and Repatriation. Routledge. In Press. 2021.

S. Huebner, E. Flowers. *The Relational Language of First Nations Cultural Sensibilities, Principles and Storytelling Ethics as an Intercultural Approach to Script Development*. C. Batty, S. Taylor (eds.) International Perspectives, Palgrave Handbook of Script Development, Palgrave Macmillan. In Press. 2021.

D. Paliwal, **T. McInerney**, J. Pa, R.H. Swerdlow, **S. Easteal**, S. Andrews. *Mitochondrial pathway polygenic risk scores are associated with Alzheimer's Disease*. Neurobiology of Aging. 2021. Advanced Online Publication. DOI: 10.1016/j.neurobiolaging.2021.08.005

INVITED TALKS, CONFERENCE PRESENTATIONS, INTERVIEWS

Azure Hermes

JCSMR/Galiwin'ku Burial Poles. Interviews with the ABC, BBC, SBS, Canberra Times, National Indigenous Times and WIN News, 26–27 May 2021.

Indigenous Women in Leadership. Invited talk. International Women's Day, Australian National University.

Indigenous People in Science – Genomics. Invited talk. Year 12 biology class, Gilroy Santa Maria College.

Indigenous Genomics – NCIG perspective. Invited talk. Australian National University. 3rd year Genetics course.

NCIG. Invited talk. Presentation to ANU TRANSFORM Advisory Board.

Galiwinku repatriation and the installation of burial poles at JCSMR. Invited talk. Reconciliation Week, Australian National University.

Indigenous Community Consultation and Genomics. Invited talk. Masters of Genetics Counselling Course, University of Technology Sydney.

Fitzroy Crossing Cemetery project – Repatriation and Genetics. Invited speaker. Repatriation Course panel discussion, Australian National University.

Experiences and Perspectives from the National Centre for Indigenous Genomics. Invited talk. Queensland Genetic Counsellor Education Day.

Scaffolding Cultural Co-Creativity Project & Collaboratory. Podcast Guest.

Hardip Patel

Long road to long-read assembly. Interview. Nature Methods. 2021. DOI: 10.1038/s41592-021-01057-y.

Use of NCI resources in genomics, Conference presentation. BioCommons Showcase 2021. Virtual Meeting.

NCIG: National Centre for Indigenous Genomics. Invited talk. Computational Biology Seminar Series. Garvan Institute of Medical Research. Virtual.

Microchromosomes are building blocks of bird, reptile and mammal chromosomes. Conference presentation. 23rd International Chromosome Conference and the 24th International Colloquium in Animal Cytogenetics and Genomics. Virtual Conference.

Microchromosomes are building blocks of bird, reptile and mammal chromosomes, Conference presentation. Genetics Society of AustralAsia Annual Meeting. Virtual Conference.

Renzo Balboa

Structural variation in diverse human populations. Conference presentation. Genetics Society of Australasia Conference 2021. Virtual meeting.

Tim McLnerney

Genetic Legacies of the Mongols and the Steppe. Keynote Address to Mongolia Institute.

Deep histories of humans inferred from haplotype genealogies. Lightning talk, Genetics Society of Australasia Conference. (Virtual).

Identification of haplotype blocks using character-compatibility analysis and inference of deep human history. PhD Exit Seminar, Genome Sciences and Cancer Division, The John Curtin School of Medical Research.

COMPETITIVE FUNDING AWARDED

Australian Research Council Discovery Early Career Research Award (ARC DECRA), 2022–2024

Dr Sharon Huebner joined NCIG in 2021. She is an emerging leader in a field requiring trusted collaborations with First Nations communities throughout Australia to address questions critical to cultural and ethical, legal, and social implications of genomic research.

Dr Huebner's successful DECRA proposal – *First Nations Community-led approaches to Australian healthcare genomics* – will address key barriers to First Nations inclusion in Australian healthcare genomics. The project is working with First Nations people throughout Australia to evaluate, co-design, and implement culturally appropriate strategies and ethical research practices, including relationship-based consent, cultural integration of genomics; and ethical strategies for long-term management and use of biological samples and data for clinical and research purposes. The expected outcomes of this research are policy recommendations, contributions to national ethics and protocol guidelines, and the evaluation of educational materials and digital learning tools aimed at improving genomics literacy as well as research practices inclusive of First Nations people.



Dr Sharon Huebner

National Health and Medical Research Council (NHMRC) Synergy Grant 2022–2026 – Respecting the Gift: Empowering Indigenous Communities in Genomic Medicine

Led by Professor Alex Brown, who will be taking up an appointment at ANU in 2022, NCIG staff and its founding Director are Chief Investigators for three of the five research streams: Interim Director Professor Graham Mann; Deputy Director Ms Azure Hermes; ELSI Lead Dr Sharon Huebner; and Emeritus Professor Simon Easteal. This important project, described in detail elsewhere in this report, aims to empower communities in genomics through a culturally informed, and national approach to the collection, generation, storage, analysis, translation and use of genomic data.

GOVERNANCE BOARD

The Board met four times in 2021, holding all meetings online.

In March 2021 the ANU Council approved the Board's recommendation to amend NCIG's statute to increase the size of the Board from 9 to 10 members. The amended statute, named *National Centre for Indigenous Genomics Statute 2021*, is viewable on the Federal Register of Legislation at www.legislation.gov.au.

Collection Access and Research Advisory Committee

Dr Shayne Bellingham, former NCIG Board member, was appointed as Chair of the Collection Access and Research Advisory Committee (CARAC) and will lead the recruitment of members to the committee. Emeritus Professor Simon Easteal was engaged to produce procedural documentation mapping the process by which researchers, the CARAC, and the Board will manage applications for access to the research resource, project approval and project oversight. The procedural mapping forms the basis of a scope of works for an end-to-end online platform. It is planned to commission IT development work and to commence establishment of the committee in 2022.

Fundraising Committee

The Board formed a fundraising committee which met frequently in the first half of 2021 to develop strategies to promote NCIG and source funding. The fundraising committee was closely supported by the ANU College of Health and Medicine Development team, and ANU Advancement. The Indigenous Genomics Roundtable held in conjunction with the unveiling of the Galiwin'ku burial poles on 27 May was a successful event that enhanced awareness of and support for NCIG amongst senior government officials and parliamentarians. As the year continued, NCIG and ANU representatives led by the Vice Chancellor, Professor Brian Schmidt AC, were pleased to have the opportunity to meet with the Minister for Indigenous Australians, Mr Ken Wyatt AM, and to be invited to provide information to the Minister for Health, Mr Greg Hunt. A briefing was provided by invitation to Senator Pat Dodson, Shadow Assistant Minister for Reconciliation, and Shadow Assistant Minister for Constitutional Recognition of Indigenous Australians. These meetings provided information about NCIG, and its role within the emerging national coalition working towards an integrated and cooperative Indigenous genomics research and clinical ecosystem.

Recruitment Subcommittee

The Board formed a recruitment committee in the second half of the year (Mr Pearson, Chair; Dr Ormond-Parker; Professor Davis) which called for Expressions of Interest to fill the new seat created by the amendment to the Statute, and two seats becoming vacant by retirement. Professor Yvette Roe, Professor of Indigenous Health and the Co-Director of the Molly Wardaguga Research Centre at Charles Darwin University, and Mr Benjamin Murray, Senior Advisor at the Office of the Registrar of Indigenous Corporations joined the Board at its final 2021 meeting. The third seat will be filled early in 2022.

Meetings and attendance

Board Member	Meeting 18 24 March	Meeting 19 23 June	Meeting 20 26 August	Meeting 21 10 December
Mr Glenn Pearson	✓	✓	✓	✓
Professor Megan Davis	✓	✓	✓	✓
Dr Lyndon Ormond-Parker	✓	✓	✓	✓
Professor Lynette Russell AM	✓	✓		✓
Professor Alex Brown (until 10 December)	✓	✓	✓	Attended as guest
Professor Marcel Dinger	✓	✓	✓	✓
Professor Gareth Baynam	✓	✓	✓	✓
Professor Keith Nugent	✓	✓	✓	✓
Ms Erica Kneipp	✓	✓	✓	✓
Professor Yvette Roe (from 10 December)	---	---	---	✓
Mr Benjamin Murray (from 10 December)	---	---	---	✓

2021 FINANCIAL SNAPSHOT

Operating Grants

2021

Income

Opening Balance	\$-
2021 Allocation	\$481,155
Total Income (year to date)	\$481,155

Expenditure

Salaries	\$209,596
Equipment	\$(1,348)
Scholars' Expenses	\$2,193
Travel and Fieldwork	\$41,373
Expendable Research Materials	\$78,156
Contributions	\$19,411
Consultancies	\$1,733
Consumables	\$12,621
Other Expenses	\$4,707
Total Expenses	\$368,442

Net result **\$112,713**

NHMRC Project Grant

Life to date (2017-2021)

Income

Opening Balance	\$-
Income received in 2021	\$1,466,536
Total Income (life to date)	\$1,466,536

Expenditure

Salaries	\$684,945
Equipment	\$-
Scholars' Expenses	\$-
Travel and Fieldwork	\$93,532
Expendable Research Materials	\$252,946
Contributions	\$435,264
Consultancies	\$7,893
Consumables	\$-
Other Expenses	\$2,388
Total Expenses (life to date)	\$1,476,968

Net result **\$(10,432)**

ANU Grand Challenge Project Grant**Life to date (2020-21)****Income**

Income received in 2020	\$135,056
Income received in 2021	\$268,093
Total Income (life to date)	\$403,149

Expenditure

Salaries	\$250,143
Equipment	\$16,106
Scholars' Expenses	\$-
Travel and Fieldwork	\$5,200
Expendable Research Materials	\$91,318
Contributions	\$-
Consultancies	\$-
Consumables	\$-
Other Expenses	\$389
Total Expenses (life to date)	\$363,156

Net result	\$39,993
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Other Funds**2021****Income**

Opening Balance	\$152,256
Income received in 2021	\$-
Total Income (life to date)	\$152,256

Expenditure

Salaries	\$-
Equipment	\$-
Scholars' Expenses	\$-
Travel and Fieldwork	\$-
Expendable Research Materials	\$-
Contributions	\$-
Consultancies	\$2,137
Consumables	\$23,249
Other Expenses	\$-
Total Expenses	\$25,386

Net result	\$126,870
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NCIG TEAM

Staff

Director

Professor Graham Mann

Deputy Director & Indigenous Community Engagement Coordinator

Ms Azure Hermes

Bioinformatics Lead

Dr Hardip Patel

ELSI Lead | Research Manager

Dr Sharon Huebner

Data Manager

Ms Jiaxin Yuan (from August 2021)

Administrator | Board Secretariat

Ms Jackie Stenhouse

Research Officer

Mr Duminda Dissananyake (from September 2021)

Consultant

Emeritus Professor Simon Easteal

Students

Mr Tim McInerney

PhD candidate

Characterising haplotype blocks across global populations for inferring population dynamics

Mr Renzo Balbo

PhD candidate

Characterising Alu-SINE repeat diversity in global populations to infer evolutionary mechanisms of transposable elements

Ms Maria Silva Lara

PhD candidate

Establishing genetic links between ADHD and neurodegeneration to identify biomarkers of age-related disorders.



Ms Bridie Moy

Honours candidate

Investigating the effect of mutations of the gene variant Autoimmune Regulator (AIRE) on complex autoimmune disease including kidney disease.

Bridie Moy is a Wiradjuri woman who grew up in Batemans Bay (Yuin country). She completed a Bachelor of Science at ANU, majoring in human biology and biomedical science.

In 2021 Bridie undertook an Honours project with NCIG research collaborator, Dr Simon Jiang.

Tiwi Islanders have some of the highest rates of kidney disease in the world. It has been estimated that genetics accounts for up to 60% of the risk for kidney disease. Whole genome sequencing has identified numerous genetic variants unique to the Tiwi. One of the genes harbouring a Tiwi-unique variant is *Autoimmune Regulator (AIRE)*. AIRE mutations are associated with complex autoimmune disease including kidney disease. Bridie's project focuses on testing whether these Tiwi-unique variants are damaging, and how they may contribute to the development of kidney disease in the Tiwi.

Ms Devashi Paliwal

Medical Student

Research Project

Nuclear-encoded mitochondrial variation in Indigenous Australians.

Academic visitors

Dr Ashley Farlow
University of Melbourne

Dr Mari Kondo
University of New South Wales

Professor Stephen Leslie
University of Melbourne

Associate Professor Bastien Llamas
University of Adelaide

Dr Yassine Souilmi
University of Adelaide

Dr Rebekah McWhirter
Deakin University

Professor Alex Brown
South Australian Health & Medical
Research Institute

Ms Louise Lyons
South Australian Health & Medical
Research Institute

Funders

Bioplatforms Australia

National Computational Infrastructure

National Health & Medical Research Council

The Australian National University

The Kinghorn Foundation, Garvan Institute of
Medical Research

Research collaborators

Professor Gareth Baynam,
University of Western Australia

Dr Ashley Farlow, University of Melbourne

Associate Professor Misty Jenkins,
Walter & Eliza Hall Institute

Dr Simon Jiang, Australian National University

Professor Stephen Leslie, University of Melbourne

Dr Yu Lin, Australian National University

Associate Professor Bastien Llamas,
University of Adelaide

Associate Professor Brendan McMorran,
Australian National University

Dr Yassine Souilmi, University of Adelaide

Dr Ira Deveson, Garvan Institute of
Medical Research

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

Ramaciotti Centre for Genomics, UNSW

Indigenous communities and organisations

Kimberley Aboriginal Law and Culture Centre, WA

Kimberley Aboriginal Medical Service, WA

SING Australia

Tiwi Land Council, NT

Top End Human Research Ethics Committee, NT

Yalu Marnngithinyaraw Indigenous Corporation

Yarrabah Community







National Centre for Indigenous Genomics

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