

# **ANNUAL REPORT 2019**

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Harnessing the science of DNA to improve the health and wellbeing of Australia's First Peoples

An initiative of



Clockwise from top left: Galiwin'ku leaders visit New Parliament House, the National Computational Infrastructure, and the laboratories of The John Curtin School of Medical Research, November 2019



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## **FROM THE CHAIRS**

We are proud to present the National Centre for Indigenous Genomics (NCIG) 2019 Annual Report. We served as Chairs of the Board of NCIG during the year, Misty from January until retiring from the Board in June, and Simone and Glenn as Co-Chairs for the remainder of the year. This arrangement provided continuity as the Board's inaugural members retired over the latter half of 2018 and early 2019.

The Board's policies assure informed choice by Indigenous Australians over their samples and genomic data in the NCIG Collection. This approach has built deep engagement with the Indigenous communities NCIG works with, leading to genuine trust and support. In 2019 NCIG achieved a breakthrough in partnership with the community of Galiwin'ku that truly sets an example of best practice in Indigenous engagement in genomics. Our research team also uncovered new and profoundly important data about the genomic diversity of Indigenous Australians. These stories are told in this annual report.

During our years with this Board, we have had the opportunity to contribute locally, nationally and internationally to the development of national genomics initiatives. Misty participated in several rounds of community consultation in the Kimberley and on Elcho Island, and represented NCIG at the Summer Internship for Indigenous Peoples in Genomics (SING) in the United States in 2016. Simone represented NCIG on the Australian Health Ministers' Advisory Council Aboriginal and Torres Strait Islander Advisory Group throughout 2019, and was an independent member of the Genomics Health Futures Mission Steering Committee in 2018–19. Glenn was a key contributor to the NCIG Governance Framework in 2017, and describes his role in the repatriation of samples to Galiwin'ku in 2019 as one of the most moving and important experiences of his life.

All NCIG work to date has successfully established the foundation for a resource that can improve the health and wellbeing of Aboriginal and Torres Strait Islander peoples. But challenges lie ahead for NCIG and for Indigenous genomics in Australia more generally. Funding uncertainty risks undermining hope for a sustained focus on Indigenous genomics, and its ultimate translation to clinical care.

A continued specific focus on Indigenous Australians is essential. As this annual report illustrates, NCIG is at the forefront of efforts to ensure Indigenous Australians are included in the benefits that will come from Australia's large and growing investment in genomic medicine.



**Dr Misty Jenkins** Member of the Consultative Committee on the Indigenous Collection (2012) Board Member (2013–2019) Chair of Board (January to June 2019)



**Dr Simone Reynolds** Board Member (2014–2019) Co-Chair of Board (July to December 2019)



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Mr Glenn Pearson Board Member (from 2018) Co-Chair of Board (from July 2019)

## FROM THE DIRECTOR

My professional association with NCIG's remarkable collection of blood samples extends back more than two decades, beginning with my appointment as head of the Human Genetics Group at The John Curtin School of Medical Research in 1994. In this role I became the custodian of a large and important collection of biological samples collected from people around the world. The samples from Indigenous Australians within this collection now form the 'NCIG Collection'.

My colleagues and I at The John Curtin School of Medical Research former Department of Human Genetics were interested, in broad terms, in questions of human wellbeing, and how this was shaped by adaptability to environment. I was then, and remain today, inspired to generate knowledge not for its own sake, but to be useful. But, at its essence, the relationship between me and the biological samples I worked with was transactional.

The Board embedded an Indigenous perspective into the fabric of NCIG that changed this relationship. It was always clear to the Indigenous members of the Board that the samples were of the people, by the people, and for the people – and the policies and strategies the Board established express this perspective.

Many of the people whose samples are held in the NCIG Collection are still alive today. Those who have passed away have relatives in their community who speak for them. These people have welcomed us into their communities and have visited NCIG in Canberra. The essence of genomics could hardly be articulated with more clarity than it was by a member of Yarrabah community who said about the samples, "That's our personal life. That blood is from us."

My approach to research has been transformed by viewing genomics through an Indigenous cultural lens. I no longer see samples as resources or commodities that are there to do research upon. The samples are representations of the people who provided them. This simple acknowledgement has set up new patterns of community consultation and gaining consent. Without ethically, legally and culturally strong approaches to inclusion in genomics research, Indigenous Australians are more likely to avoid or withdraw from this critical field of research. This will be a severe setback to research and clinical services that would otherwise help reduce the burden of disease over time. Through an exemplary community consultation process, built on the core truth that DNA is personal, NCIG has overcome entrenched suspicion toward genetics and genomics and achieved extraordinary engagement with those whose samples are held in the NCIG Collection. As personally rewarding as this is, I am even more excited by the vastness of new knowledge that is beginning to emerge from our work, and its potential to bring benefit to Indigenous Australians.



Silver Fatal

**Professor Simon Easteal, PhD, MBA** Director, National Centre for Indigenous Genomics



# THE NCIG STORY

NCIG is built around a remarkable legacy of 7,000 biological samples collected from Indigenous Australians between the 1960s and 1990s. Our interest is in genomic research with a medical and health application. We are based in The John Curtin School of Medical Research, one of Australia's preeminent medical research institutes.

NCIG gives control of decision-making to Indigenous people. We recognise that genomic research about Indigenous Australians must have the support of Indigenous Australians to succeed. This has been the case from the beginning, when The Australian National University (ANU) established NCIG in 2013 in a format recommended by Indigenous Australians, centred on a strong model of Indigenous governance and Indigenous community engagement.

The core principle is that no sample in the NCIG Collection is 'reactivated' for use in research unless new consent is obtained. To date, the NCIG Indigenous Community Engagement Coordinator, Ms Azure Hermes, has worked with the communities of Yarrabah in Queensland, and Titjikala, Tiwi Islands and Galiwin'ku (Elcho Island) in the Northern Territory, to locate the people whose samples are held in the NCIG Collection. She has provided information and support as they made

their decisions about the future of their sample or, where applicable, the sample from their now deceased family member.

In 2016 ANU established the National Centre for Indigenous Genomics Statute to ensure appropriate Indigenous custodianship, engagement and consent. As a result, NCIG gives Indigenous Australians assurance of legally-binding, accountable Indigenous oversight and protection of their genomic data, samples and choices. The program of community engagement for NCIG genomic research is an outstanding example of practice that has resulted in a 90 per cent rate of consent by donors to the use of their samples for research.

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The program of community engagement for NCIG genomic research is an outstanding example of practice that has resulted in a 90 per cent rate of consent by donors to the use of their samples for research.

Decision-making occurs differently in each community, and this is accommodated in the NCIG governance framework and model of engagement. For example, some communities have preferred to engage and decide through communal meetings, others have asked for engagement with clan groups, and others family-by-family. Some communities have organisations to coordinate with researchers such as NCIG. Other communities defer to the leadership of respected seniors. The most important ingredients in NCIG's approach to community engagement and consultation are truth and time. Everything is open. Nothing is rushed.

Based on this unique approach, and with funding from ANU, the National Health and Medical Research Council (NHMRC) and Bioplatforms Australia, NCIG is assembling the first Indigenous genomic reference data resource. This is a pioneering and accessible research resource that will underpin innovative Australian genomic research and clinical practice into the future.



# **GENOMIC RESEARCH**

2019 was a year of consolidation of NCIG's internal research program. There was steady progress on our two main genomics projects: the creation of population reference data; and long-read genome assemblies.

During the year, we reduced the volume of new genome sequencing to allow greater focus on analysing the data already collected and developing data management systems and pipelines for data analysis.

The first round of analysis for NCIG's two main projects is now well advanced. The results are revealing important features about genomic variation in Aboriginal and Torres Strait Islander communities. We are preparing the results for presentation in plain language to participating communities, and for scientific publication.

Not only are the emerging results significant in their own right, they are also allowing us to refine the conceptual and technical development of processes for making data available for further research. Providing appropriate access to relevant data is an integral part of the developing national health genomics framework.

In addition, NCIG has joined with clinical and biomedical researchers in The John Curtin School of Medical Research and elsewhere in a project that will investigate the causes of renal failure in the Tiwi community, as part of the ANU Indigenous Health and Wellbeing Grand Challenge program. This project was in the planning stage in 2019 and is expected to commence during 2020.

#### Population reference data project

At the beginning of this project we expected, based on what we knew about people from other parts of the world, that information about genomic variation of Aboriginal and Torres Strait Islander peoples was likely to have important implications for healthcare. However, we couldn't be sure.

We had to answer questions such as: How much genomic variation is there in Indigenous communities? How does the amount of variation compare with populations in other parts of the world? How much of the variation is likely to affect health? How much does genomic variation differ among communities in different parts of Australia? The results of our initial analysis of the genome sequences of 160 people from four communities is providing answers to these questions. Importantly, the results show that:

The amount of genomic variation within Australian Indigenous communities is slightly less, overall, than most populations in Europe and Asia. Variation in populations in all three places is substantially less than in African populations.

Approximately 25 per cent of all DNA variants in the genome of an Australian Indigenous person are unknown in people from outside Australia. This degree of region-specific variation is similar to populations in other parts of the world. It remains to be investigated how many of these Indigenous-specific variants are associated with health and disease.

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Of the Indigenous-specific variants, approximately 40 per cent are likely to be found in a single region or community. Overall, genomic differences among communities across Australia are as great as those between populations across Europe and Asia combined. In medical genomics terms, some Australian Indigenous communities are as different from each other as communities as far apart as North West Europe and South East Asia. So, using information about people from the Northern Territory, for example, as a basis for treating people in South Western Australia, would be equivalent to treating people in the United Kingdom based on information about people from Cambodia.

This analysis provides a critical foundation for further investigation of the health and medical importance of genomic variation in Aboriginal and Torres Strait Islander communities. We will carry out further sequencing in 2020, and work with participating communities to develop processes and methods for making the data available for research and clinical use.

The work outlined above is funded by BioPlatforms Australia (\$0.5 million, ongoing from 2017).

### Long-read genome assembly project

The rationale for this project is the biased nature of the human reference genome that is fundamental to all genomic analysis. The reference genome has a strong African and European bias, which evidence indicates can negatively impact the quality of genomic analysis and subsequent data interpretation for people from other parts of the world.

Our aim is to *de novo* assemble the genomes from Indigenous Australian people and assess how well use of these genomes as a reference mitigates this bias. It is also important to assess whether multiple references are justified because of genomic differences between communities in different parts of the country.

We have completed four assemblies, one from each of the communities we are working with. Analysis of these is well advanced and we are writing up the results for plain language presentation to communities and for research publication.

Analysis so far indicates differences between communities that justify the use of multiple, regional reference genomes in communities across Australia. Our data also provides information to substantially improve the quality of the existing reference genome by characterising the sequence in 25 per cent of places in the genome where the sequence is currently not known.

An accurate reference genome is particularly important for characterising variation resulting from novel insertion or loss of DNA segments in the genome. Our initial characterisation of this 'structural' variation indicates unusually high levels of difference among the communities, adding further weight to the need for multiple reference sequences from different regions.

The work outlined above is funded by a National Health and Medical Research Council (NHMRC) Project Grant (\$1.4 million, 2018–2020).

## OTHER FUNDED RESEARCH PROJECTS

Roadmap to integrate Indigenous genome assemblies into a national instance of the International Human Reference Genome Resource

(Australian Research Data Commons (ARDC) Discovery activities, \$50,000, 2019)

Professor Simon Easteal and Dr Hardip Patel prepared a report, which they presented at the eResearch Australasia Conference, 21–22 October 2019.

The report covered NCIG's investigation of the current state of the human reference genome: how it is being used in research and clinical practice; how it is likely to develop over the next few years; and the current state of research and development in Australia. The key findings were:

- there is a lack of standardisation both in Australia and internationally in the use of the human reference genome
- there is a critical unmet need for a national genome reference resource
- for Australian ancestral diversity to be included within a global human reference genome resource, a national facility needs to be developed.

#### ANU grand challenges 2019

#### (ANU, \$1 million over five years, 2020–2025)

NCIG was awarded funding for a collaborative project with Associate Professor Brendan McMorran, Dr Simon Jiang and the Tiwi Land Council to extend an established, long-running study of renal failure.

Chronic kidney disease (CKD) is at epidemic levels in Indigenous Australians and is particularly widespread in remote communities. Our project focuses on a specific Indigenous group: the Tiwi Islanders, some 2,500 individuals who are the Indigenous inhabitants of Bathurst and Melville Islands. Approximately one third of all Tiwi exhibit the clinical signs of CKD. Metabolic conditions as well as poor diet, lifestyle and access to health services contribute to the burden of renal disease in Indigenous Australians. However, there is a major knowledge gap in explaining the aetiology of CKD. While we know CKD is exacerbated by other chronic and metabolic conditions, and the impact of recent acculturation, it is important to understand if the Tiwi have any underlying genetic susceptibility to CKD. This project aims to:

- identify CKD-causing genes in the Tiwi
- determine the functions of the CKD-causing genes
- develop an Indigenous-led CKD data and genomics resource for the Tiwi and researchers.







Ross Mandi Wunungmurra, Chair of Yalu Marnggithinyaraw Indigenous Corporation, with the repatriated samples. Credit: Jamie Kidston/ANU.

### THE GALIWIN'KU REPATRIATION

#### **NAVIGATING BETWEEN TRADITIONAL & CONTEMPORARY**

In November 2019, ANU repatriated blood samples from deceased donors from Galiwin'ku (Elcho Island, Northern Territory) to their families. The repatriation was the culmination of two years of patience, persistence, innovation and vision. It was an inspired, community-led navigation between Indigenous cultural belief systems and modern genomic science.

### How the blood samples came to be with NCIG

In 1968 an outbreak of typhoid occurred on Galiwin'ku (Elcho Island) and the Northern Territory Health Department began taking blood samples from everyone on the island. Approximately 1,200 samples were obtained during two collection rounds in 1968 and 1969. Extensive documentation was generated. Dr Bob Kirk was the group leader of the Human Biology Department of The John Curtin School of Medical Research at that time. Dr Kirk's research interests were wide, and can be broadly described as the characterisation of genetic variation in Aboriginal populations. Dr Kirk coordinated with and obtained permission from the Northern Territory Health Department to obtain the samples, which remained at The John Curtin School of Medical Research for 50 years.

#### The consultation

The Galiwin'ku samples are the largest set in the NCIG Collection from a single community/ site, and have the most detailed information retained about the study participants. This wealth of information was instrumental in opening and conducting community consultation in 2018 and 2019. The people of Elcho Island acted through their community body, Yalu Marnggithingaraw. Yalu gave cultural and strategic advice about the community consultation and helped organise family (clan) meetings. Support was also provided by the Arnhem Land Progress Association, East Arnhem Regional Council, Shepherdson College, and the cultural law group Dalkarra Djirrikay Authority.

In Yolngu culture it is very important that once a person has died their body and spirit is returned to the land they came from. The initial reaction of many people on Elcho Island was distress, shock and even anger that blood samples were held at ANU. At first, many wanted the samples withdrawn from genomic research and returned for burial. But many also saw an opportunity, and felt an urgency, to harness genomic research to help lift the burden of disease borne by many Indigenous Australians.

#### The community-led outcome

Over several months a range of options were considered and, in early 2019, a proposal emerged that answered the dual calls of culture and science. NCIG would extract the DNA from the 215 samples from deceased donors<sup>\*</sup>, perform DNA sequencing, and keep the genomic data in the NCIG data repository. The residual material would be grouped by family and repatriated to Country into the care of their family. The NCIG Board and ANU were delighted to support this proposal.

\*The DNA and data from samples of people still living was managed according to the wishes of those individuals. Most chose to permit DNA and data to be extracted and instructed NCIG to dispose of the residual material.



Aunty 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.

#### The repatriation

We undertook careful work in the laboratory to extract DNA for later sequencing. Ashley Eriksmoen from the ANU School of Art and Design made small boxes to hold the repatriated material using timber reclaimed from the former Burgman College on campus. Each box was etched with the name of the family. The material was handled in two groups according to their Yolgnu moiety: Yirritja; and Dhuwa.

Ngambri-Ngunnawal elder, Aunty Matilda House, then conducted a smoking ceremony on the ANU campus to farewell the samples from Ngunnawal country.



Left to right: Simon Easteal, Shane Dhawa Bukulatipi, Brian Schmidt, Rosemary Gundjarranbuy, David Yangaririny Munyarryun, Matilda House, Ross Mandi Wunungmurra, Graham Mann.



#### **Coming home**

NCIG Board and senior ANU representatives were honoured to be the guests of the community of Galiwin'ku at the sample homecoming. Hundreds of community members staged a joyous Buŋgul (dance). Clan after clan assembled, the didgeridoo and clapping sticks commenced, and the families danced forward towards two ceremonial burial poles to receive their box from ANU Deputy Vice Chancellor Research and Innovation, NCIG Board Chair and NCIG Director.

The artistry of the burial poles is stunning. The Yirritja pole (left) was painted by Judy Manany Gurruwiwi and the Dhuwa pole was painted by Peter Datjing Burarrwanga. The poles were installed in a newly created commemorative garden, which has become a place of pride in the town centre.

While the cremated remains of those who have passed are traditionally interred in burial poles in this part of northern Australia, these highly public poles are not expected to be used in this way. Each family will make a

private choice about the final resting place for the repatriated remains, with many likely to be returned to homelands across the island.

#### The achievement

Our shared ability to create a research model that respects Indigenous cultural practices and genomic discovery is a significant achievement, and a gift from each to the other. It is a testament to the worth of the Indigenous-led model by which NCIG conducts genomic research.

We have each gained from the experience. The relationship between our community partner, Yalu, and other groups on the island has been strengthened and renewed. As a result of the publicity around NCIG's visit, researchers from around Australia have engaged with Yalu, seeing a community that is open to pragmatic but also innovative approaches to research. For our part, NCIG - although uncertain that useable DNA could be successfully extracted from 50-year-old blood samples - succeeded in obtaining good quality DNA. The project generated an opportunity to develop, test and prove laboratory protocols that can be applied to other samples in the NCIG Collection in the future. It means that the 'sequence-and-repatriate' model can be offered to other communities that want it.

This story generated widespread interest. It was reported 310 times in the national media, reaching a potential audience of more than 1.5 million people. We were told that the story was even seen by the (predominantly Australian) passengers on a cruise ship crossing the Pacific Ocean. The story also featured in *Nature News* (Nature 577, 11–12 (2020) doi: 10.1038/ d41586-019-03906-5).

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Something remarkable has happened. Conceptual boundaries have been shifted and the model for conducting human genomic research has been reshaped.

ANU Vice Chancellor Brian Schmidt, 2019



# FITZROY CROSSING CEMETERY PROJECT

#### UPDATE

In our 2018 Annual Report we shared the moving story of the Fitzroy Crossing Cemetery. The cemetery had been heavily eroded by river action over time and many graves holding the remains of Aboriginal people had been lost. The remaining 70 graves were relocated to a new cemetery in 2018. Of those relocated, fewer than 20 were able to be individually identified. With the help of our research collaborator, Dr Bastien Llamas from the University of Adelaide, NCIG extracted material from the exhumed remains prior to reinterment. The purpose was to gather DNA for later analysis and comparison, so the remains could be identified.

In 2019 Dr Llamas spent time in the specialist laboratories of ANU to extract DNA from the skeletal material collected in 2018. DNA was present in most samples. A pilot DNA sequencing run was performed, pending further analysis to work out quantity and quality of human DNA in these samples.

The next step is to obtain funding, in conjunction with our local partner, Kimberley Aboriginal Law and Culture Centre (KALACC). Not only must sequencing and analysis be conducted on the DNA, there must also be a program of community engagement to definitively identify the remains, and to allow families to decide the final resting place for their relative. Some families may elect to leave their family member resting at the new Fitzroy Crossing Cemetery, while others may prefer to repatriate remains to Country.



This is a reconciliation story...it also involves [white fellas] – their mob helping to put things right. I've been talking about the DNA project – people want to give saliva samples as well. I have been telling people that NCIG is an Aboriginal organisation that is not going to allow things to go off into [white fella's] hands and [be] used for wrong purposes.

Community member, Kimberley, Western Australia, 2019

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### **COMMUNITY VISITS, CONFERENCES & MEETINGS**

JANUARY	FEBRUARY	MARCH
Wet season!	<b>Garrabah</b> Annual update to community	Tiwi Islands Community consultation Galiwin'ku
	Meeting with Tiwi Land Council	Community consultation NCIG Board meeting
APRIL	MAY	JUNE
<b>Townsville</b> Summer Internship for Indigenous Peoples in Genomics (SING) Workshop <b>Titjikala</b> Annual update to community	Canberra & Galiwin'ku Repatriation planning NCIG Board meeting	Helsinki, Finland Infrafrontier/International Mouse Phenotyping Consortium Conference Melbourne Genetics Society of AustralAsia Conference Galiwin'ku Community consultation
JULY	AUGUST	SEPTEMBER
Brisbane Australian Institute of Aboriginal and Torres Strait Islander Studies Research Conference Galiwin'ku Community consultation	Wellington, New Zealand Human Genetics Society of Australasia Conference NCIG Board meeting	The Kimberley Community organisations annual general meetings Galiwin'ku Community consultation
OCTOBER	NOVEMBER	DECEMBER
Addis Ababa, E-thiopia Cell Atlas meeting	Canberra & Galiwin'ku Repatriation activities	Wet season! NCIG Board meeting
Brisbane e-Research Australasia Conference	The Kimberley Community consultation planning	
Galiwin'ku Community consultation	NCIG Board meeting	

## **SCIENTIFIC MEETINGS** & CONFERENCES

•••••	••••••
31 March – 3 April	SING Workshop, Townsville Ms Azure Hermes
3–5 June	Infrafrontier/International Mouse Phenotyping Consortium Conference 2019, Helsinki, Finland Dr Hardip Patel
1–3 July	AIATSIS Research Conference, Brisbane Ms Azure Hermes
30 June – 3 July	Genetics Society of Australasia Conference, Melbourne Dr Hardip Patel and Mr Renzo Balboa (PhD candidate)
3–6 August	Human Genetics Society of Australasia Conference, Wellington, New Zealand Ms Azure Hermes
30 September	Biological Data Science Institute Interactive Seminar, Canberra Mr Renzo Balboa
17 October	Dragon Genomics meeting, Canberra Dr Hardip Patel
21–22 October	eResearch Australasia Conference, Brisbane Dr Hardip Patel presented ARDC Roadmap paper
31 October	Cell Atlas meeting, Addis Ababa, Ethiopia Ms Azure Hermes
2019	SING Australia, organising committee meeting, Melbourne Ms Azure Hermes
2019	Aboriginal and Torres Strait Islander Advisory Group on health genomics reporting to Australian Health Ministers' Advisory Council (AHMAC) Ms Azure Hermes (staff member), Prof Gareth Baynam, Dr Shayne Bellingham, Dr Lyndon Ormond-Parker and Dr Simone Reynolds (NCIG Board Members)

# **EDUCATION**

#### Aurora project intern

NCIG hosted an intern from the Aurora Project between 4 February and 8 March 2019. Ms Lauren Chappill, an undergraduate law and health science student from the University of Adelaide, worked on a project investigating approaches to governance for genomic data. She reflected upon her placement with NCIG positively, saying, "through my research I learnt a lot about how genetic material is gathered and stored around the world, and the different ethical protocols which are in place within the global scientific community. I also learnt how Indigenous genetic material is handled and stored around the globe and how researchers interact with their Indigenous donors. This research was very interesting as it allowed me to research both the legal implications of storing Indigenous genetic data and also the implications of maintaining Indigenous sovereignty".

#### Shell Questacon science circus

The Shell Questacon Science circus partnered with NCIG to deliver an in-school workshop during the Galiwin'ku Repatriation visit in November. Two experienced Questacon outreach staff travelled to Galiwin'ku with the NCIG team and delivered a rotation of science-themed activities to around 200 enthusiastic students at Shepherdson College. With 21 scientists on the island for the repatriation event, there was plenty of knowledge to share. The goal was to spark the students' interest and excitement about science and to show, through fun interactive activities, how science is happening all around us.

### ANU Indigenous STEM summer school

In December NCIG conducted a hands-on genomics session with the visiting students from the ANU Indigenous STEM Summer School. We are pleased to participate annually in this program.

### Undergraduate and postgraduate students

Eight students worked under the supervision of NCIG's academic team in 2019, although not all student projects were NCIG-related. We supported three ongoing PhD students, one PhB student, one Masters student and three undergraduate Honours students. Further information is provided at *NCIG team* on pages 19–20.

# **GOVERNANCE BOARD**

#### Activity

In 2019 the NCIG Board continued to advocate for better national approaches to achieving inclusion for Indigenous Australians in genomics health initiatives. Dr Simone Reynolds, Prof Gareth Baynam, Dr Shayne Bellingham and Dr Lyndon Ormond-Parker sat on the Aboriginal and Torres Strait Islander Advisory Group on health genomics convened by the Australian Government Department of Health to advise the Project Reference Group on Health Genomics, and ultimately the Australian Health Ministers' Advisory Council. The Board conducted a recruitment process mid-year. The contribution of Mr Joe Hedger, Director, Indigenous Sector Practice, Ernst & Young, as an independent external member of the recruitment sub-committee was greatly appreciated.

#### Rotation of board membership

во	ARD MEMBERSHIP AT 1 JAN 2019	RETIREMENTS	NEW MEMBERS	BOARD MEMBERSHIP AT 1 JAN 2020
$\checkmark$	DR SIMONE REYNOLDS (Co-Chair from July)	31 Dec 2019		
$\checkmark$	MR GLENN PEARSON (Co-Chair from July)			$\checkmark$
$\checkmark$	DR MISTY JENKINS (Chair January to June)	30 Jun 2019		
$\checkmark$	PROF GARETH BAYNAM			$\checkmark$
$\checkmark$	DR SHAYNE BELLINGHAM			$\checkmark$
$\checkmark$	PROF JOHN BEKKERS	16 Oct 2019		
$\checkmark$	PROF DON CHALMERS	30 Jun 2019		
$\checkmark$	PROF KEITH NUGENT			$\checkmark$
$\checkmark$	DR LYNDON ORMOND-PARKER			$\checkmark$
	PROF MEGAN DAVIS		From 1 Jul 2019	$\checkmark$
	PROF MARCEL DINGER		From 1 Jul 2019	$\checkmark$
	PROF GRAHAM MANN		From 17 Oct 2019	$\checkmark$
	PROF LYNETTE RUSSELL		From 1 Jan 2020	$\checkmark$

#### Meetings held & attendance

BOARD MEMBER	MEETING 9 23 MAR 2019	MEETING 10 23 MAY 2019	MEETING 11 30 AUG 2019	MEETING 12 19 NOV 2019	MEETING 13 5 DEC 2019
DR SIMONE REYNOLDS (Co-Chair from July)	$\checkmark$	$\checkmark$	$\checkmark$	$\checkmark$	$\checkmark$
MR GLENN PEARSON (Co-Chair from July)	$\checkmark$	×	$\checkmark$	$\checkmark$	$\checkmark$
DR MISTY JENKINS (Chair January to June)	$\checkmark$	$\checkmark$	_	_	_
PROF GARETH BAYNAM	$\checkmark$	×	$\checkmark$	×	$\checkmark$
DR SHAYNE BELLINGHAM	$\checkmark$	$\checkmark$	$\checkmark$	$\checkmark$	$\checkmark$
PROF JOHN BEKKERS	$\checkmark$	$\checkmark$	$\checkmark$	-	-
PROF DON CHALMERS	$\checkmark$	×	-	-	-
PROF KEITH NUGENT	$\checkmark$	$\checkmark$	$\checkmark$	$\checkmark$	$\checkmark$
DR LYNDON ORMOND-PARKER	$\checkmark$	$\checkmark$	$\checkmark$	$\checkmark$	$\checkmark$
PROF MEGAN DAVIS	_	_	$\checkmark$	$\checkmark$	×
PROF MARCEL DINGER	_	_	$\checkmark$	$\checkmark$	$\checkmark$
PROF GRAHAM MANN	_	_	✓ (observer)	$\checkmark$	$\checkmark$
PROF LYNETTE RUSSELL	_	_	✓ (observer)	✓ (observer)	×
PROF SIMON EASTEAL (Director)	$\checkmark$	$\checkmark$	$\checkmark$	$\checkmark$	$\checkmark$
MS JACKIE STENHOUSE (Secretariat)	$\checkmark$	$\checkmark$	$\checkmark$	$\checkmark$	$\checkmark$

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# **NCIG TEAM**

#### Staff

Director Professor Simon Easteal, PhD, MBA

Manager, Strategic Relationships (until September 2019) Dr Michael Dobbie

Indigenous Community Engagement Coordinator Ms Azure Hermes

**Bioinformatics Lead** Dr Hardip Patel

Administrator | Board Secretariat Ms Jackie Stenhouse

Research Assistants Ms Alice McCarthy, Mr Mark Hermes, Ms Claire Hubbard, Ms Somasundhari Shanmuganandam

#### **Students**

Professor Easteal and Dr Patel supervised the following postgraduate and undergraduate students in 2019.

**Mr Tim McInerney** PhD candidate *Characterising haplotype blocks across global populations for inferring population dynamics* 

**Mr Renzo Balboa** PhD candidate *Characterising Alu-SINE repeat diversity in global populations to infer evolutionary mechanisms of transposable elements*  Ms Maria Silva Lara

PhD candidate Establish genetic links between ADHD and neurodegeneration to identify biomarkers of age-related disorders

**Mr Hanif Patel** PhB candidate Polygenic risk scoring for Type 2 diabetes across populations using variant fixation Index measures

**Mr Jiajia Xu** Masters candidate *Calling variants from both short and long reads* 

**Ms Devashi Paliwal** Undergraduate honours candidate *Nuclear-encoded mitochondrial genetic variation and expression associated with mitochondrial pathways significantly influences Alzheimer's disease risk and has therapeutic potential* 

Mr Brendan Siles Undergraduate honours candidate Comprehensive evaluation of NumtS in diverse human populations

**Ms Beilei Wang** Undergraduate candidate *K-mer properties and their applications for human Y chromosome* 

#### Academic visitors

Professor Stephen Leslie, University of Melbourne Dr Ashley Farlow, University of Melbourne Dr Mari Kondo, University of New South Wales

# **2019 FINANCIAL REPORT**

	ANU	PROJECT GRANTS	PHILANTHROPY	TOTAL
Income				
Carried forward	\$61,772.18	\$76,526.00	\$70,619.18	\$208,917.36
Operating grants	\$719,735.14	\$1,162,377.62	\$9,408.45	\$1,891,521.21
	\$781,507.32	\$1,238,903.62	\$80,027.63	\$2,100,438.57
EVERNOED				
EXPENSES				
Salaries	\$324,119.51	\$306,779.04	-	\$630,898.55
Equipment	\$168.18	-		\$168.18
Scholars expenses	\$1,500.00	-	-	\$1,500.00
Travel & fieldwork	\$201,343.18	\$93,531.62	-	\$294,874.80
Expendable	\$39,295.33	\$137,779.65	-	\$177,074.98
research materials				
Contributions	-	\$144,474.00	-	\$144,474.00
Consultancies	\$16,184.28	\$7,892.81	\$2,137.50	\$26,214.59
Consumables	\$18,308.99	-	\$23,248.98	\$41,557.97
Internal purchases	\$800.00	-	-	\$800.00
Other expenses	\$34,030.68	\$25.19	\$9.17	\$34,065.04
	\$635,750.15	\$690,482.31	\$25,395.65	\$1,351,628.11
NET RESULT				
	\$145,757.17	\$548,421.31	\$54,631.98	\$748,810.46

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# **SUPPORTERS**

#### **Funders**

- Australian Research Data Commons
- Bioplatforms Australia
- National Computational Infrastructure
- National Health and Medical Research Council
- The Australian National University

#### Advisors, project partners

- Ernst & Young, pro bono
- Shell Questacon Science Circus

#### **Research collaborators**

- Professor Gareth Baynam, University of Western Australia
- Dr Ashley Farlow, University of Melbourne
- Dr Simon Jiang, Australian National University
- Associate Professor Stephen Leslie, University of Melbourne
- Dr Yu Lin, Australian National University
- Dr Bastien Llamas, University of Adelaide
- Associate Professor Brendan McMorran, Australian National University

#### Technical service providers

- The Australian Phenomics Facility/Biomolecular Resource Facility, Australian National University
- Information Technology Services, Australian National University
- Kinghorn Centre for Clinical Genomics (Garvan Institute of Medical Research)
- National Computational Infrastructure, Australian National University
- Ramaciotti Centre for Genomics, University of New South Wales

### Indigenous communities & organisations

- Dalakarra Djirrikay Authority, Galiwin'ku, Northern Territory
- Galiwin'ku community (Elcho Island), Northern Territory
- Kimberley Land Council, Western Australia
- Kimberley Law and Culture Centre (KALACC), Fitzroy Crossing, Western Australia
- Marra Worra Worra Aboriginal Corporation, Fitzroy Crossing, Western Australia
- Northern Land Council, Northern Territory
- SING Australia
- Titjikala community, Northern Territory
- Tiwi Land Council, Northern Territory
- Yalu Marnggithinyaraw Indigenous Corporation, Northern Territory
- Yarrabah community, Queensland



Top: NCIG Indigenous Community Engagement Coordinator, Azure Hermes, and Board member, Lyndon Ormond-Parker, ran an information stall at the Tiwi Island Aussie rules grand final, a major community event that enabled them to connect with people from outlying districts.

Below clockwise from top left: The artist cuts the tree for the burial pole, Galiwin'ku, NT; community members conduct ceremony at the new Fitzroy Crossing Cemetery, WA; The John Curtin School of Medical Research, Canberra, ACT.



#### National Centre for Indigenous Genomics

#### The John Curtin School of Medical Research

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