NCIG
NATIONAL CENTRE FOR INDIGENOUS GENOMICS

ANNUAL REPORT 2020

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

An initiative of

Australian National University

CONTENTS

FROM THE CHAIR

COVID-19 has changed us all, and we acknowledge the disruption, trauma and loss of life across the world.

Strains and structural weaknesses in the social fabric of Australia and elsewhere are being pressure tested. Black Lives Matter and 434 Aboriginal deaths in custody in Australia since the Royal Commission in 1991 remind us that there is much work to be done in the way we do business together.

The National Centre for Indigenous Genomics (NCIG) is giving a national voice to the ancient polities that exist on this land, linking their wisdoms to modern technologies and, in doing so, showing there *is* a way of doing business together. Aboriginal self-determination and decision-making is embedded into the philosophy and operations of NCIG. NCIG should not be a landmark or outlier for this way of operating in 2020, yet it is. Importantly, the substance of NCIG's work is as transformational as the way it operates.

Genomics has the potential to change the lives of some of the most vulnerable people in this country, and NCIG can help. Inspired by that conviction, I acknowledge and thank my fellow Board members and the staff of NCIG who have worked hard throughout 2020 to seek funding in a fiercely constrained fiscal environment, and to continue the challenging work of establishing a foundational data platform for Indigenous genomic research in Australia.

Glenn Pearson
Chair

FROM THE DIRECTOR

In September 2020 NGIG's founding Director, Professor Simon Easteal, retired from the Australian National University (ANU), and consequently stepped down after seven years at the helm of NCIG.

Simon's stewardship of the Aboriginal blood samples that form the foundation of NCIG extends back three decades. This rare and precious collection – holding in its DNA the life stories of thousands of Aboriginal Australians – has been preserved and protected by Simon since the 1990s. We owe him a debt of gratitude for his steadfast commitment to doing the right thing by those samples and Aboriginal people as a whole. NCIG was established from his determination, and under his leadership, in 2013. It was conceived as a model that bakes in Indigenous leadership and ethical, respectful engagement and inclusion of Indigenous Australians in genomics. NCIG stands as Simon's signature achievement in an already distinguished career. I speak for all current and former Board members when I offer my deep thanks and congratulations to Simon for his tireless work and remarkable achievement in establishing and leading NCIG.

I am honoured to take up the role of Director of NCIG. I will work closely with the Board and the staff of NCIG to progress our goal of establishing a world-class Indigenous genomic data platform, and to identify the scientific and technical leaders who will take NCIG onward.

This has been an extraordinarily challenging year, which has disrupted the work of NCIG and our research colleagues locally, nationally and internationally. Of most significance, it has not been possible to visit any Indigenous communities this year due to travel restrictions and border closures. We were unable to conduct the annual 'reporting back' visits nor implement our planned round of engagement in the Kimberley, Western Australia. On the other hand, the NCIG team was able to attend a remarkable number of local, national and international conferences, workshops and meetings online, continue to research, write and plan for the future, and in general get on with job. This report records those achievements.

Professor Graham Mann MBBS PhD FRACP
Director, The John Curtin School of Medical Research, and
Interim Director, National Centre for Indigenous Genomics

THE YEAR OF COVID-19

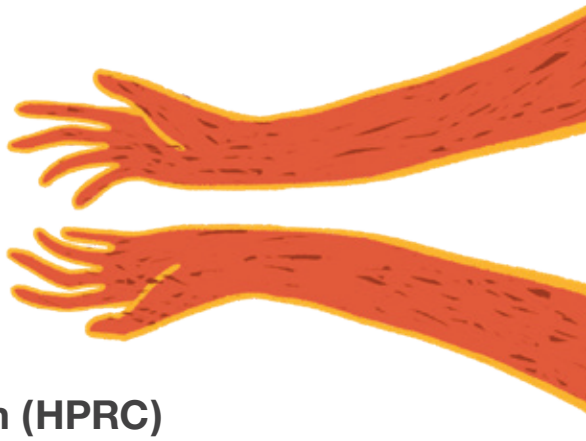
The NCIG staff and student body worked from home for a significant period in 2020. The ANU campus was closed intermittently in January and February due to bushfire smoke and hail damage. The campus then closed fully in March in response to COVID-19. Members of the team began to return to the office in August.

NCIG Director, Professor Simon Easteal, retired in September after 33 years at ANU. Professor Easteal has taken an Emeritus appointment and will continue to support NCIG in a scientific capacity. Professor Graham Mann, the Director at The John Curtin School of Medical Research, accepted the additional role of NCIG Director, taking leave of absence from the Board for the duration. Ms Azure Hermes, NCIG's Indigenous Community Engagement Coordinator, was promoted to the role of Deputy Director and will provide significant support to Professor Mann.

The shutdown of NCIG and external laboratories for part of the year, and the inability to travel to communities for consultation and fieldwork, delayed some elements of NCIG's work program. In acknowledgement, the National Health and Medical Research Council granted a 12-month extension for the project grant held by NCIG.

Other work continued, however, including important progress formalising ethical arrangements and approvals with the Northern Territory Aboriginal Ethics Committee and the Western Australian Aboriginal Health Ethics Committee.

BUILDING STRONG COLLABORATIONS



Human PanGenome Reference Consortium (HPRC)

NCIG has formally partnered with the HPRC, a United States based initiative to address the lack of diversity in the human reference genome. It will also facilitate the transition of the latest DNA sequencing technologies to generate the most accurate representation of the human genome possible. HPRC is funded by the National Human Genome Research Institute. NCIG's participation will help ensure Indigenous priorities are woven into the fabric of this foundational genomic resource and identify opportunities to develop capacity in pangenome reference development in Australia.

National Computational Infrastructure (NCI)

In 2020, NCIG entered a formal collaboration with the NCI to build the first genomic data repository in Australia. This repository will serve the unmet, yet critical, need for data sovereignty and long-term usage of genomic data for improvement of public health. This repository will act as a foundation for implementing international standards in the management and use of sensitive genomic data and supporting the systematic integration of genomics into healthcare.

Educational activities

During the year's brief opportune time , NCIG hosted its third intern from the Aurora Project *(*https://www.auroraproject.com.au/*)*. Ms Sahamar (Sami) Ruben, an Indigenous student studying a Bachelor of Medical Laboratory Science at Queensland University of Technology, spent four weeks with NCIG in February. Along with a range of experiences from NCIG's program, Ms Ruben gained a taste of some of the fascinating research and laboratory support services that are part of the daily life of The John Curtin School of Medical Research.

Ms Ruben wrote about her experiences at NCIG:

In February 2020 I was privileged to have the opportunity to participate in the Aurora Internship Program. I have been aware of this program for a while but had been hesitant to apply as I can be shy in new situations. However, with encouragement from influential people in my life, I decided to apply. I currently study a Bachelor of Medical Laboratory Science at Queensland University of Technology in Brisbane.

I got the most amazing offer to undergo a 4-week placement at the National Centre for Indigenous Genomics (NCIG) at the Australian National University in Canberra. I was very nervous at first going to Canberra as I had only previously been there on a school trip. However, the help from the Aurora placements team made the adjustment incredibly easy and affordable. I found Canberra to be very similar to Brisbane with its low-set buildings and its spacious streets. My favourite part about the city though was the many museums and tourist locations, and I visited as many as I could.

NCIG is an organisation under Indigenous governance, which has constructed a biobank of samples of mostly blood but also some saliva. NCIG's ultimate goal is to create a database of Indigenous genomic data, and as part of their process they will repatriate the biological samples back to their traditional owners or the owner's family if that is what they want. These samples had been gathered and held at ANU since the 1960s.

During my time at NCIG I learnt a lot about the bioinformatics behind genetic sciences. In my degree I mainly get to experience the hands-on side of genomics, typically in the laboratory, extracting DNA myself.

However, my time at NCIG opened my eyes to a different side of genomics, namely about the data which is produced from the procedures I perform in the lab. I got to experience how they use the data, how it is stored (a trip to the high tech computer facility at ANU called the National Computational Infrastructure) and overall how the data assists in the repatriation process.

I travelled to Melbourne with one of their communications coordinators to shadow her during meetings. This was an amazing opportunity and gave me a real insight into what goes on behind the scenes to write an academic paper. I got to conduct an interview for a book chapter they are writing and then transcribe the interview the next day. Transcribing takes way longer than you expect but it was actually pretty fun to do.

I got the pleasure of working alongside an honours student in the fruit fly lab within The John Curtin School of Medical Research. I observed and assisted in making a nutrient agar appropriate for the flies to live off. We then went to the fly lab to move the flies into new containers, as they need to be changed every four weeks. I then got to observe injecting a specific gene into the eggs of the flies, with the tiniest needle I have ever seen.

During my time at NCIG I met some of the most amazing people. The team there are so incredibly welcoming and assisted me with anything I needed whether it was work related or after hours, including talking to me about my post graduate studies and the best route to take. I never once felt uncomfortable while away from home and it was a big thanks to that team.

I highly encourage anyone looking at the program to jump in and take every opportunity. The Aurora team are so incredibly helpful and make the transition so easy. It allows you to gain real world experience in the field you are studying, which is a once in a lifetime chance.

Sami Ruben
Currently undertaking: Bachelor Medical Laboratory Science

SING – Summer Internship for Indigenous Genomics

SING is a week-long workshop and mentoring program that covers technical and laboratory skills in genomics, as well as theory, ethical and social implications of genomic research and the benefits and risks of genomics for Indigenous peoples. Originating in the United States 10 years ago, the concept is now franchised around the world through a consortium structure. In Australia, the first SING workshop was held in 2020.

NCIG has maintained a relationship with the SING network for some years. NCIG Board Member, Dr Misty Jenkins attended the 2015 SING Conference in the United States, and Board members and staff of NCIG are members of the SING Australia organising committee. NCIG sponsored Indigenous genetics/archeology student Dawn Lewis to attend the Aotearoa SING Indigenous Genomics Conference 2020 (https://www.singaotearoa.nz) in New Zealand in January.

Ms Lewis reflected on her experience:

I would like to thank the NCIG Board for sponsoring me to attend the Aotearoa SING conference. This was my first ever opportunity to interact directly with First Nations Canadian, Native American or Maori geneticists and I found the experience invaluable. Although I didn't realise until I arrived, I was the only Australian at the Alumni event, which was a little confronting but ultimately useful for the Consortia to hear about our inaugural SING.

The first three days were an alumni event, which allowed the Consortia to come together and discuss different aspects of Indigenous Genomics. It was informal, and a series of antagonistic questions put to the group allowed us to discuss the complexities of many issues. There wasn't necessarily an intention to come to a cohesive conclusion or even debate merits of each 'side' so much as an opportunity to share knowledge and experience. I think everyone walked away from these discussions with some food for thought.

At the conference itself every presentation was amazing but a couple stood out particularly in my mind. Firstly, Maui Hudson presented his work with biocultural labels, which provide clear communication around the ethical background and potential for future work with Indigenous culture including native flora and fauna. The idea that we could evaluate the integrity of an academic paper, I feel, is particularly useful when engaging with a community already struggling with the inaccessibility of language in these papers.

Secondly, and in line with my feelings the more I engage with Aboriginal and Torres Strait Islander genomics, Chip Colwell's keynote address on ancient DNA and repatriation in the United States really struck a chord. He did a wonderful job articulating the potential pitfalls of ancient DNA studies for Indigenous people, alongside the potential positive outcomes. For example, Chip asserts DNA studies involve the risk of:

- *undermining water or land rights*
- *amplifying faulty constructs of race*
- *challenging sovereignty*
- *prompting scepticism of clinical health studies in Indigenous communities*
- *creating bias towards scientific expertise over Indigenous expertise*
- *causing distrust of researchers*
- *sowing discord within Indigenous communities*
- *leading to missed opportunities for collaborative research.*

This has been a truly invaluable experience. I will be able to bring my experience in Aotearoa back to the Australian SING organising committee. I would also like to thank Rebecca McWhirter and Azure Hermes for their (extensive) help and patience.

Link to write up from the university of Waikato: https://www.waikato.ac.nz/news-opinion/media/2020/indigenous-genomics-under-the-microscope-at-sing-conference

Dawn Lewis
BA/Sc (2019)
Currently undertaking: Bachelor of Science (Honours). Molecular genetics and archaeology.

COMMUNITY SERVICE (COMMITTEE MEMBERSHIP)

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

Hermes A. Mackenzie's Mission Indigenous Advisory Committee, Australian Genomics.

Hermes A. Aboriginal and Torres Strait Islander Health Survey Advisory Committee, Australian Bureau of Statistics.

Hermes A. Indigenous Data Governance Working Group, Indigenous Data Network, University of Melbourne.

Hermes A, Easteal S. ANU College of Health & Medicine Reconciliation Action Plan Committee.

RESEARCH PROJECTS

Population reference data project

The NCIG population variation project aims to investigate and describe medically relevant genomic variation and the processes that drive the nature of this variation within the NCIG sample. This work is fundamental for producing a valuable and useful genomic data resource from the NCIG sample. It sets the context for the collection and ensures its utility for other researchers. 'Medically relevant' in this context means both:

- directly – the variants that are known or predicted to be harmful (this is difficult to address using a population sample)
- indirectly – the background 'natural' genetic variation that must be accounted for when studying or investigating a disease (a key feature that can be readily addressed using the NCIG data collection).

We have undertaken a large and comprehensive range of analyses to address these aims, in addition to underpinning work on data preparation and quality control. All analyses are completed except one, which is substantially complete. A manuscript for publication is close to completion.

Long-read genome assembly project

The current human reference genome has a strong African-European bias, and evidence from studies in other populations indicates it is likely to be a suboptimal template for genomic studies in Indigenous populations. By assembling a high quality Aboriginal Australian Reference Genome, we aim to understand and mitigate this bias.

This project will deliver a set of resources that improve clinical insight based on the current reference genome and on novel genome assemblies from Aboriginal individuals. To achieve this, we will release two resources, each with two components.

Resource 1: An Aboriginal Australian augmentation of the current reference genome:

- A set of genome intervals (in Browser Extendable Data format[1]) on the human reference genome, the GRCh38 coordinate system, that demarcate regions requiring special care. Requiring the least amount of specialist knowledge, this will signal regions that need a more nuanced interpretation. It will include annotations.
- Variant calls (process of identifying variants from sequence data) of particular importance. This dataset will include genetic variation that is typically recovered with standard tools but is either novel or at unusually high frequency in Aboriginal populations. This is produced in combination with the population sample data from the Population Reference Data Project. Importantly, however, it will contain variants that require the long read assemblies to identify.

Resource 2: Standalone Aboriginal genome assemblies and 'alt-contigs':

- The set of novel sequences that are not represented within the current reference. Regions of novel sequence, or regions that are highly divergent from the reference, will be made available as alternative-sequence-patches (alt-contigs) to the current human reference.
- Genome assemblies for each individual. In addition to the above resources, we continue to undertake analysis that describes the quality of genetic variation these assemblies allow us to recover, the biological or historical causes for this variation, and the technical and potential clinical consequences.

All four components were near completion at the end of 2020. A manuscript for publication is planned for 2021.

Tiwi renal disease project

In partnership with the McMorran Group at The John Curtin School of Medical Research and the National Computational Infrastructure, NCIG has been funded under the ANU Indigenous Health and Wellbeing Grand Challenge to conduct a five-year project on the genetic basis of renal disease among Tiwi Islander Australians.

The project commenced in September 2020, with the first stage of work focused on designing the 'data management engine' that will underpin the project: a customised instance of the MediaFlux platform installed under license at the National Computational Infrastructure.

1 A text file format used to store genomic regions as coordinates and associated annotations.

Harvard collaboration

In September 2020, Mr Renzo Balbo, PhD candidate, started a collaboration with Dr Shilpa Garg, a research fellow at Harvard Medical School and the Dana-Farber Cancer Institute in Boston, Massachusetts. Mr Balbo is currently working on a project to accurately detect structural variation in segmentally duplicated regions within the human genome. This research will help uncover structural variation within complex unresolved regions of the human genome that have pivotal roles in clinical and biodiversity studies. It will also be useful in detecting somatic structural variation within cancer genomes.

Pharmacogenomics for Indigenous peoples

Dr Hardip Patel collaborated with the team led by Dr Shiv Nagarajan and Arvind Jaya Shankar (PhD student) at the Queensland University of Technology to understand and describe properties of genes involved in drug metabolism. Adverse drug reaction and non-responsiveness to a drug is a common problem in healthcare. Large-scale genomics is helping to unravel the underpinnings of an individual's response to common drugs. Through careful analyses of genomic data from Tiwi Islanders, this team was able to identify mutations that may have an impact on drug metabolisms for Tiwi Islanders. Overall, 22 variants relevant to how an individual responds to drugs were discovered. These variants relate to 37 clinically actionable guidelines, with implications for drug dosing and treatment of conditions including cancer, depression and diabetes. Targeted follow-up research focused on Indigenous peoples will allow an actionable plan to be developed.

RESEARCH PUBLICATIONS

NCIG staff, students and board members contributed to the following research publications.

D'Angelo CS, **Hermes A**, McMaster CR, Prichep E, Richer É, Van Der Westhuizen F, Repetto GM, Gong M, Malherbe H, Reichardt J, Arbour L, Hudson M, du Plessis K, Haendel MA, Wilcox PL, Lynch SA, Rind S, **Easteal S**, Estivill X, Thomas Y, **Baynam G**. (2020). Barriers and considerations for diagnosing rare diseases in Indigenous populations. ***Frontiers in Pediatrics: General Pediatrics and Emergency Care*** (accepted for publication 2 November 2020).

Easteal S, Arkell R, **Balboa RF, Bellingham SA**, Brown AD, Calma T, Cook MC, **Davis M**, Dawkins HJS, **Dinger ME**, Dobbie MS, Farlow A, Gwynne KG, **Hermes A**, Hoy WE, Jenkins MR, Jiang SH, Kaplan W, Leslie S, Llamas B, **Mann GJ**, McMorran BJ, McWhirter RE, Meldrum CJ, Nagaraj SH, Newman SJ, Nunn JS, **Ormond-Parker L**, Orr NJ, Neil, Paliwal D, **Patel HR, Pearson G**, Pratt GR, Rambaldini B, **Russell LW**, Savarirayan R, Silcocks M, Skinner JC, Vinuesa CG, **The National Centre for Indigenous Genomics, Baynam G** (2020). 'Equitable expanded carrier screening needs Indigenous clinical and population genomic data'. ***American Journal of Human Genetics*** 107(2):175–182. (DOI: 10.1016/j.ajhg.2020.06.005).

Baynam G, Groft S, van der Westhuizen FH, Gassman SD, du Plessis K, Coles E, Selebasto E, Selebasto M, Gaobinelwe B, Selebasto T, Joel D, Llera VA, Vorster C, Wuebbels B, Djoudalbaye B, Austin CP, Kumuthini J, John Forman J, Kaufmann P, Chipeta J, Gavhed D, Larsson A, Stojiljkovic M, Nordgren A, Roldan E, Taruscio D, Wong-Rieger D, Nowak K, Bilkey GA, **Easteal S**, Bowdin S, Reichardt JKV, Agulló SB, Kosaki K, van Karnebeek CDM, Gong M, Shuyang Z, Shai R, Adams DR, Puri RD, Zhang F, Pachter N, Muenke M, Nellaker C, Gahl WA, Cederroth H, Broley S, Kym M. Boycott KM, Posada M. (2019). 'Rare Diseases in Africa and the Protea Declaration – collaborative approaches for the unmet needs and potential benefits for Africa'. ***Nature Genetics*** (DOI:10.1038/s41588-019-0552-2; PMID: 31873296).

Hermes A, Huebner S, **Easteal S, Ormond-Parker L**, McCarthy A, Gundjarranbuy Garrawurra R, Mandi Wunungmurra R (2020). 'We are taking it back to our homeland; We are free to move on: Aboriginal participation in genomics research: Returning historical blood samples to the Galiwin'ku community'. ***The Routledge Companion to Indigenous Repatriation: Return, Reconcile, Renew.*** Fforde C, McKeown C, Keeler H (Eds.). Routledge, New York (book chapter, in press).

Huebner S, **Hermes A**, **Easteal S** (2020). 'The practice of engaging Aboriginal and Torres Strait Islander communities in genome research'. Ch 8; pp.109–126. ***Indigenous Research Ethics: claiming research sovereignty beyond deficit and colonial legacy***. George L, Tauri J, MacDonald L (Eds). Emerald Publishing, Bingley, United Kingdom (book chapter).

CONFERENCES, WORKSHOPS AND PRESENTATIONS

Easteal S. Medical genomics and Australians of Aboriginal and/or Torres Strait Islander descent. Lorne Genome Conference, Lorne, Australia. Plenary. February 2020.

Balboa R, Easteal S, Patel H. Alu repeat diversity in the human genome. Lorne Genome Conference, Lorne, Australia. Poster presentation. February 2020.

McInerney TW, Easteal S, Patel HR. Identifying blocks free of recombinant haplotypes in human genomes using character-compatibility. Lorne Genome Conference, Lorne, Australia. Poster presentation. February 2020.

Patel H, Easteal S. Genotypic sex specific use of the human reference genome. Lorne Genome Conference, Lorne, Australia. Poster presentation. February 2020.

Easteal S. The National Centre for Indigenous Genomics, Rare Voices Rare Disease Day, The John Curtin School of Medical Research. Public Lecture. February 2020.

Balboa R, Easteal S, Patel H. A comprehensive survey of Alu repeat diversity. The John Curtin School of Medical Research Bioinformatics Seminar Series. Platform presentation. March 2020.

Balboa R, Easteal S, Patel H. A comprehensive survey of Alu repeat diversity. Human Genome Organisation Conference (HUGO), Perth, Australia. Platform presentation. April 2020.

Easteal S. National Centre for Indigenous Genomics. HUGO Conference, Perth, Australia. International Plenary. April 2020.

Hermes A. Live panel discussion with Professor Ian Anderson AO and Dr Virginia Marshall for ANU National Reconciliation Week, 'In this Together'. May 2020.

Patel H. National Centre for Indigenous Genomics: 2020 Update. Genome Science Departmental Seminar. Australia, ANU. August 2020.

Hermes A. Panel discussion with Professor Marcel Dinger to present perspectives of First Nations Genomics, University of New South Wales Science Week. August 2020.

McInerney TW, Paliwal D, Raviv J, Andrews S. Update to 'A globally diverse reference alignment and panel for imputation of mitochondrial DNA variants (MitoImpute)' and 'Mitochondrial pathway polygenic risk scores are associated with Alzheimer's Disease'. Ronald M. Loeb Center for Alzheimer's Disease at the Icahn School of Medicine at Mount Sinai, New York. Presentation. August 2020.

Easteal S, Llamas B, Patel H. The National Centre for Indigenous Genomics. Human Pangenome Reference Genome Consortium Meeting, Washington DC, United States. International. September 2020.

Easteal S. National Centre for Indigenous Genomics. Telomere-to-Telomere and Human Pangenome Reference Consortium Conference, Washington DC, United States. International Plenary. September 2020.

Balboa R, Easteal S, Patel H. Alu repeat diversity in the human genome. Telomere-to-Telomere and Human Pangenome Reference Consortium Conference, Washington DC, United States. Poster presentation. September 2020.

Patel H. National Centre for Indigenous Genomics, Telomere to Telomere & Human Pangenome Reference Consortium Conference, poster presentation and selected talk. September 2020.

Hermes A. University of Technology Sydney, presentation to Master of Genetic Counselling class. September 2020.

Balboa R, Easteal S, Patel H. Alu repeat diversity in the human genome. COMBINE Conference 2020, Australia. Platform presentation. October 2020.

Easteal S. Presentation to Poche Key Thinkers Forum Precision Medicine. October 2020.

Patel H. National Centre for Indigenous Genomics: An Update. Australian Genome Health Alliance Program 2 Monthly Meeting. Invited presentation. October 2020.

Hermes, A. Co-moderator of an all-Indigenous panel at the American Society of Human Genomics conference. 'Indigenous Biobanking: Global Perspectives on Keeping our Genomic Data Local'. October 2020.

Balboa R, Easteal S, Patel H. Alu repeat diversity in the human genome. ABACBS Conference 2020, Australia. Asynchronous presentation. November 2020.

Acera Mateos P, **Balboa R, Easteal S**, Eyras E, **Patel H**. PACIFIC: A lightweight deep-learning classifier of SARS-CoV-2 and co-infecting RNA viruses. ABACBS Conference 2020 (online), Australia. Asynchronous presentation. November 2020.

Sethi AJ, Acera Mateos P, **Balboa R**, Weiss E, Horvath H. A machine learning model to predict splice factor expression directly from transcriptome-wide splicing patterns. ABACBS Conference 2020, Australia. Asynchronous presentation. November 2020.

McInerney TW, Patel HR, Easteal S. Identifying blocks free of recombinant haplotypes in human genomes using character-compatibility. COMBINE Symposium 2020. Presentation. November 2020.

Hermes A. University of New South Wales Science and Engineering Indigenous Preparatory Program. Presentation. December 2020.

Patel H. National Centre for Indigenous Genomics. AMSI BioInfoSummer 2020. Invited presentation. December 2020.

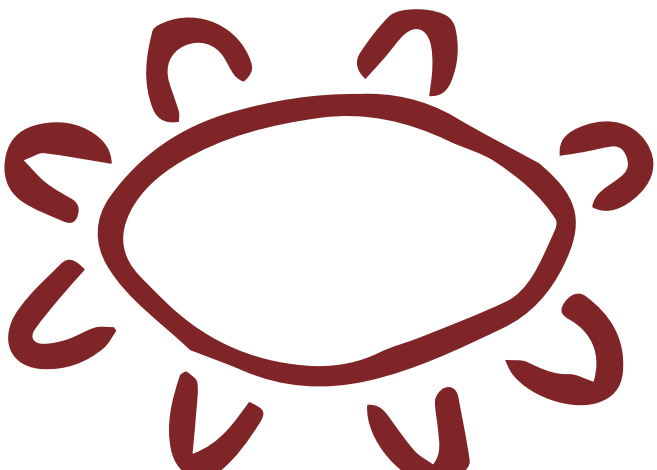
GOVERNANCE BOARD

The Board met four times in 2020 – holding all meetings online. Professor Graham Mann took leave of absence from the Board effective 5 September 2020 to take up the role of Director of NCIG. At the invitation of the Vice Chancellor, Ms Erica Kneipp, Head of Research Strategy, ANU College of Health & Medicine, accepted the seat.

Dr Shayne Bellingham's term ended on 31 December 2020. Dr Bellingham has accepted the Board's invitation to become the inaugural Chair of the Collection Access and Research Advisory Committee (CARAC), which will be launched in 2021. Dr Bellingham will take leave of absence from the Board in 2021 while fulfilling the duties of CARAC Chair.

Meetings and attendance

BOARD MEMBER	MEETING 14 26 MAR 2020	MEETING 15 9 JUN 2020	MEETING 16 2 SEP 2020	MEETING 17 4 DEC 2020
Mr Glenn Pearson	✓	✓	✓	✓
Professor Megan Davis		✓	✓	✓
Dr Lyndon Ormond-Parker	✓	✓	✓	✓
Dr Shayne Bellingham	✓	✓	✓	✓
Professor Lynette Russell AO	✓	✓	✓	✓
Professor Marcel Dinger	✓	✓		✓
Professor Gareth Baynam	✓	✓	✓	✓
Professor Keith Nugent	✓	✓	✓	
Professor Graham Mann (until 5 Sep)	✓	✓	✓	---
Ms Erica Kneipp (from 5 Sep)	---	---	---	✓



Funding

In 2020 NCIG was funded by ANU, the National Health and Medical Research Council, Bioplatforms Australia and the National Computational Infrastructure (NCI).

As the momentum of NCIG's work swings towards computation and the curation, management and storage of data, we have been well supported by the NCI. NCIG was awarded more than 4 million computational Service Units and 250 Terabytes of storage capacity in 2020 via the National Competitive Merit Allocation Scheme and the ANU Competitive Merit Allocation Scheme, and we acknowledge with gratitude the ongoing support and expertise provided by NCI for the establishment of NCIG's genomic data platform.

One of many occasions NCI staff showed NCIG guests the supercomputing facility. This tour in late 2019 the last occasion preCOVID that visitors joined us in Canberra. Left to Right: Rosemary Gundjarranbuy*, Anna Tonkin, Asmi Wood (ANU), David Yangaririny Munyarryun*, Shane Dhawa Bukulatjpi*, Graham Mann (ANU), Ross Mandi Wunungmurra,* Andrew Howard (NCI/ANU).

*Community leaders from Galinwin'ku (Elcho Island), Northern Territory.

2020 FINANCIAL STATEMENT

	ANU	PROJECT GRANTS	PHILANTHROPY & OTHER	TOTAL
Income				
Carried forward	$129,116	$1,179,863	$54,667	$1,363,646
Operating grants	$538,901	$718,144	10,299	$1,267,345
	$668,018	$1,898,007	$64,966	$2,630,991
Expenses				
Salaries	$431,396	$466,738		$898,133
Equipment	$4,760			$4,760
Scholars expenses	$1,000			$1,000
Travel and fieldwork	$15,047	$93,532		$108,579
Expendable research materials	$8,606	$361,433		$370,038
Contributions		$240,790		$240,790
Consultancies	$14,284	$52,893		$67,177
Consumables	$5,676			$5,676
Internal purchases				
Other expenses	$16,453	$39,070		$55,523
	$497,220	$1,254,454	-	$1,751,675
Net result	$170,797	$643,553	$64,966	$879,316

NCIG TEAM

Staff

Director
Professor Simon Easteal, PhD, MBA (until September 2020)
Professor Graham Mann, (from September 2020)

Deputy Director
Ms Azure Hermes (from September 2020)

Indigenous Community Engagement Coordinator
Ms Azure Hermes

Bioinformatics Lead
Dr Hardip Patel

Administrator | Board Secretariat
Ms Jackie Stenhouse

Laboratory Technical Officer
Ms Somasundhari Shanmuganandam

Consultant

Dr Sharon Huebner

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

Academic visitors

Dr Ashley Farlow, University of Melbourne

Dr Sharon Huebner, 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

ACKNOWLEDGEMENTS

Funders

Bioplatforms Australia

National Computational Infrastructure

National Health and Medical Research Council

The Australian National University

The Australian National University Indigenous Health and Wellbeing Grand Challenge

Research collaborators

Professor Gareth Baynam, University of Western Australia

Dr Ashley Farlow, University of Melbourne

Associate Professor Misty Jenkins, Walter and 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

Technical service providers

The Australian Phenomics Facility, Australian National University

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

Ramaciotti Centre for Genomics, University of New South Wales

Indigenous communities and organisations

Kimberley Aboriginal Law and Cultural Centre, Western Australia

Kimberley Aboriginal Medical Service, Western Australia

SING Australia

Tiwi Land Council, Northern Territory

Top End Human Research Ethics Committee, Northern Territory

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