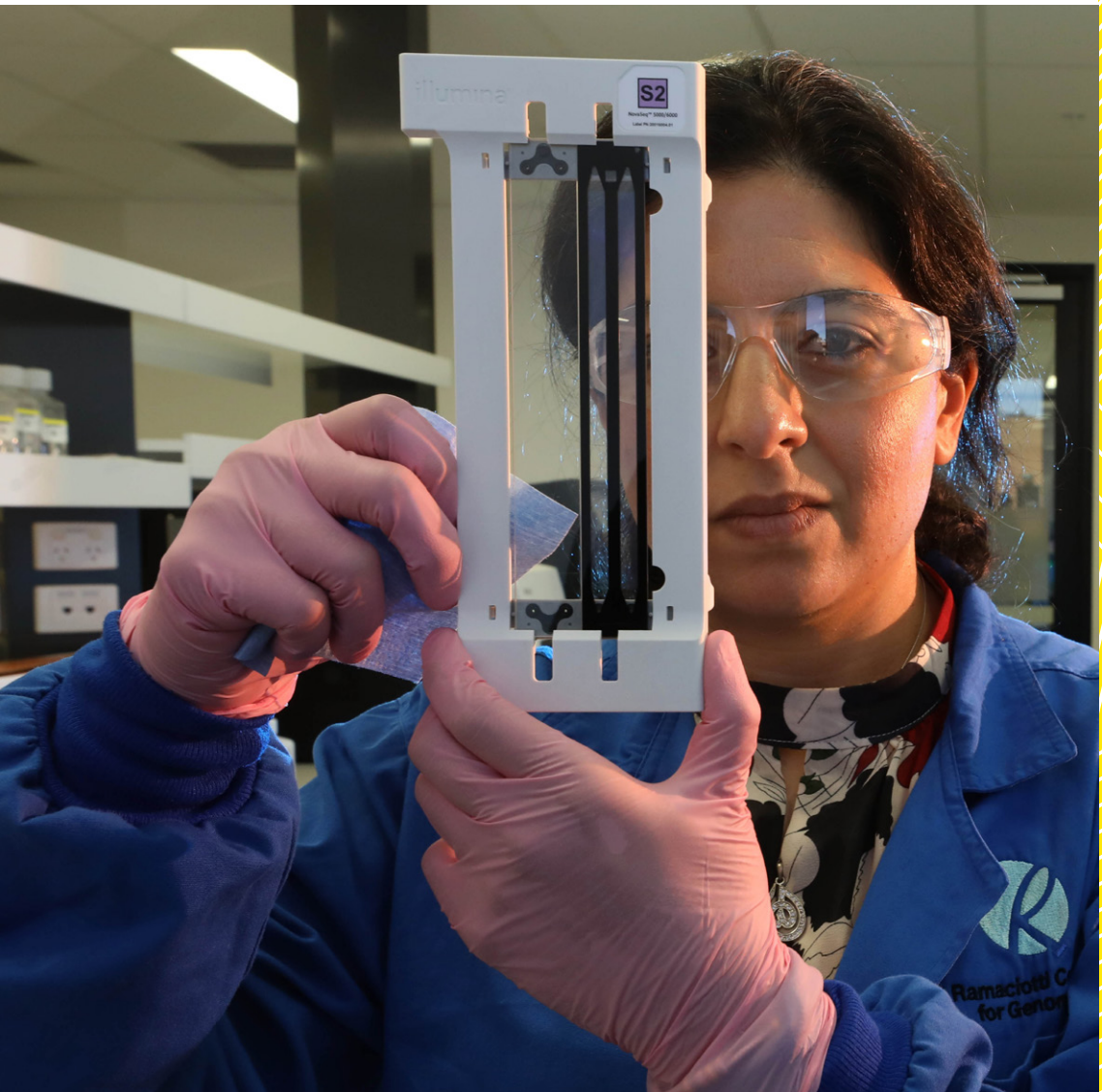
Ramaciotti Centre for Genomics

Annual Report 2018/19

UNSW SYDNEY

Next generation science.
Working for you.



Ramaciotti Centre for Genomics
Biosciences South Building (E26)
UNSW Sydney NSW 2052 Australia
Tel: +61 9385 1241
Web: ramaciotti.unsw.edu.au
Email: ramaciotti@unsw.edu.au
Find us on social media: @ramacgenomics

Acknowledgement of Traditional Owners
We would like to acknowledge the Bedegal people that are the Traditional Custodians of the Land on which the Ramaciotti Centre for Genomics is located. We pay our respects to the elders, past, present, and future, and recognise their continuing connection and contribution to this land.

Disclaimer
The information contained in this publication is correct at the time of printing but may be subject to change without notice. Please check the Centre's website for the latest information. UNSW assumes no responsibility for the accuracy of information provided by third parties.

Design
Slingshot Design

About Us

The Ramaciotti Centre for Genomics is Australia's leading genomics research and resource centre with the capacity to deliver data, analysis, guidance and advice of the highest quality. It is funded as National Collaborative Research Infrastructure and is a focus for the development and application of genomics in Australia. It was established in 1999 and is the largest genomics facility at any Australian University. It is comprehensively equipped with the latest next-generation sequencing technology, high throughput microarray systems and robotics to allow projects to be done at scale.

Our Mission

Is to deliver internationally competitive genomic services, of the highest quality, using leading edge technology.

Our Objectives

- To provide our users with access to enabling technology and services, facilitating internationally competitive research.
- To provide genomics and related services of the highest possible quality.
- To provide support for bespoke and niche projects.
- To provide services at competitive rates.
- To build the genomics community in the state of NSW and beyond.

Our Values

- Excellence
- Collaboration
- Innovation
- Diversity
- Respect

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Director's Report

I'm very pleased to report that the Ramaciotti Centre for Genomics has had two exceptional years. We have consolidated our role as the largest provider of genomics services in the state of NSW, and were again acknowledged as national infrastructure by the award of a substantial four-year NCRIS grant. We formalised our quality systems and secured NATA accreditation for clinically-relevant aspects of our services. We completed the Centre's single largest project, involving the genotyping of a 15,000 patient cohort; one of the biggest such screens undertaken in Australia. We became a founding partner in the UNSW Futures Institute in Cellular Genomics – a multidisciplinary initiative focused on the use of single cell genomics for research and precision medicine. We also acquired one of the first MGI sequencers in Australia and expanded our platform of Illumina sequencers to increase capacity. The hard work previously undertaken on the koala genome project was ultimately published in Nature Genetics.

In 2018 and 2019, demand for the Centre's technology and services has continued to grow. This has been from biomedical researchers in universities, medical research institutes, publicly funded research agencies and from industry. Our researcher base of ~500 labs kept us incredibly busy. Short read sequencing continued to grow dramatically, with the Centre analysing a total of 66,012 samples by next generation sequencing over the two years. The ASPREE genotyping project contributed to our greatest ever throughput in microarray, involving 20,726 samples in 2018 and 2019. Demand for Sanger sequencing remained also very strong with 58,247 samples submitted and analysed in the two years. A further 4,413 samples were analysed by other techniques. To streamline our interactions with researchers, we adopted a CRM (customer relationship management) platform; this helped with the execution of ~1,000 different projects per year. We also continued to increase our use of robotics, with the acquisition of two new major automation systems.

The Centre's financial position remains robust. Substantial fee-for-service income, NCRIS funding, support from the NSW State Government and the University of New South Wales positions us well for further technology acquisition and provides sustained funding for our staff. With the Australian Research Council's LIEF scheme, we will continue the acquisition of new technology.

The Centre's growth and ongoing success has only been possible through the hard work of the entire team. I thank them for their ongoing attention to detail, for the pride they take in their work and the care they take with every precious sample that is entrusted to us. Their professionalism is outstanding and their dedication to the research community makes them all unsung heroes! The fact that the Centre was acknowledged in 280 publications in 2018 and 2019 illustrates the high regard that researchers have for our staff's efforts and expertise.

Finally I would like to acknowledge our senior management and steering committee. I remain in awe of Dr. Helen Speirs, Dr. Helena Mangs, Jason Koval, Tonia Russell and Dr Jackie Chan and thank them for their outstanding management of so many projects, in a technically demanding field, and in the delivery of exceptional outcomes to biomedical researchers. Our steering committee provides excellent advice on our direction and strategy and are key to our ongoing technology acquisition via the ARC LIEF scheme. I warmly thank them and also acknowledge Professor Ian Dawes, the chair of our steering committee, for ongoing good humour and for sage advice.

We look forward to further supporting the genomics community in 2020.

Professor Marc Wilkins
Director

Our People

The Centre's professional team of scientists have numerous years of experience delivering genomic services. Many of our staff are PhD qualified or have industry experience.

They bring knowledge and skills from areas including biomedical research, biotechnology, the environment and conservation.

The Ramaciotti Centre is a nationally recognised genomics facility thanks to our dedicated team. We thank staff past and present who have contributed to its ongoing success.

In 2018 and 2019 we welcomed Crystal Cui, Scott Genner, Jessica Gurr, Gurjeet Kohli, Melissa Rapadas and Rhys Stark to the professional laboratory team, Keren Natalia as Quality Manager and Abigail Teo as Administration Officer. We said farewell to Alper Yasar who was appointed to the role of Project Officer at the office of the Pro Vice Chancellor for Infrastructure, UNSW, to Zena Kassir who took up a position with Illumina technical support and to Carolina Correa Ospina who took up a position at the Biomolecular Resource Facility, John Curtain School of Medicine, ANU.

As of the close of 2019 our team included:

Centre Director
Professor Marc Wilkins

Deputy Director – Centre Manager
Dr. Helen Speirs

Research Managers
Jason Koval
Dr. Helena Mangs

Quality Manager
Keren Natalia

Senior Research Officers
Dr. Jackie Chan
Tonia Russell

Research Officers
Dr. Erika Becker
Dr. Angela Chilton
Dr. Jessica Gurr
Dr. Gurjeet Kohli
Dr. Caitriona Murray
Dr. Kristin North
Dr. Carolina Correa Ospina
Dr. Darshi Ramesh
Stefano Vecchiarelli

Research Assistants
Dr Kylie Cairns
Crystal Cui
Scott Genner
Lisa Phan
Melissa Rapadas
Rhys Stark

Administrative Officer
Abigail Teo

Systems and Network Administrator
Robin Heron

… bring knowledge and skills from areas including biomedical research, biotechnology, the environment and conservation.

Honorary UNSW Fellowship Awarded to Emeritus Professor Ian Dawes

Former Centre Director Emeritus Professor Ian Dawes FAA FRSN was awarded an Honorary Fellowship from UNSW on the 13th May 2019. Staff, family and former colleagues and students were present to celebrate this award, presented in recognition of his long service to the research community and UNSW.

Professor Dawes is a member of the Australian Academy of Science and has made significant contributions to eukaryotic gene regulation during cell development and responses to the environment. His studies of yeast sporulation provided a detailed understanding of metabolism during meiosis and identified the motif that regulates specific gene expression during meiosis. He was first to show that eukaryotes can mount adaptable responses to oxidative stress and by combining biochemical, genetic and genomic analyses, he has made a substantial contribution to elucidating mechanisms whereby cells adapt, maintain resistance, and respond, to oxidants. A significant outcome of this regulation research was the identification of a novel system controlling one-carbon metabolism in yeast.

Professor Dawes established the Ramaciotti Centre in 1999 with an award from the Clive and Vera Ramaciotti Foundation and served as Centre director from 1999 until 2010. He oversaw the establishment of transcriptomics using in-house manufactured spotted arrays and commercial gene arrays, a capillary Sanger sequencing service that has since analysed hundreds of thousands of samples, and the introduction of next-generation sequencing. We are fortunate to have Ian continue to serve as the independent chair of the Centre's steering committee.

Professor Ian Dawes receives his Honorary Fellowship from the Chancellor of UNSW, David Gonski.

Developing Leadership Skills – Horizon Program

UNSW, in partnership with KPMG, launched the Horizon Program in December 2017 as part of their long-term collaborative and strategic partnership, focused on the delivery of their closely aligned strategic priorities. The 18-month program aimed to inspire and develop highly talented people and enable their success in an innovative and entrepreneurial culture. The Centre's Deputy Director and Centre Manager Dr Helen Speirs was one of only six applicants – three from UNSW – selected for the program.

The program accelerated the development of the participants' professional skills through a mix of formal learning in general management, coaching on leadership behaviours and workplace secondments. Helen applied for the Horizons program to continue to develop her leadership skills, bring back new knowledge to the Centre and to challenge herself. Her first secondment to KPMG's Brand & Engagement division exposed her to different sector and consequently a different way of thinking, giving her some fresh ideas on how to position the Centre and raise its profile within the research community. Her second secondment to the UNSW3+ team, part of the UNSW 2025 Strategy, gave her the opportunity to work on the program to restructure the academic calendar from two semesters to three terms.

Helen said "Whilst the Horizon Program was challenging, it was a very positive opportunity to grow and develop at UNSW. I thank the team and the Centre Director Marc Wilkins for their support throughout the program."

Steering Committee

The Ramaciotti Centre for Genomics is affiliated with other universities and medical research centres in the Sydney region.

These include the University of Sydney, Macquarie University, the University of Technology Sydney, the University of Newcastle, the Victor Chang Cardiac Research Institute and the Garvan Institute of Medical Research. Representatives from each sit on the Centre's scientific advisory committee. The committee helps guide the Centre's strategic direction and objectives.

Representatives on the committee at the close of 2019 included:

Emeritus Professor Ian Dawes (Chair)
UNSW Sydney

Professor Marc Wilkins (Director)
UNSW Sydney

Dr Helen Speirs (Deputy Director)
UNSW Sydney

Professor Paul Munroe (Deputy Dean Faculty of Science) – UNSW Sydney

A/Professor Mark Cowley
Children's Cancer Institute, Sydney

Professor Aaron Darling
University of Technology, Sydney

A/Professor Sarah Kummerfeld
Kinghorn Centre for Clinical Genomics

Professor Ian Paulsen
Macquarie University

Professor Brett Neilan
University of Newcastle

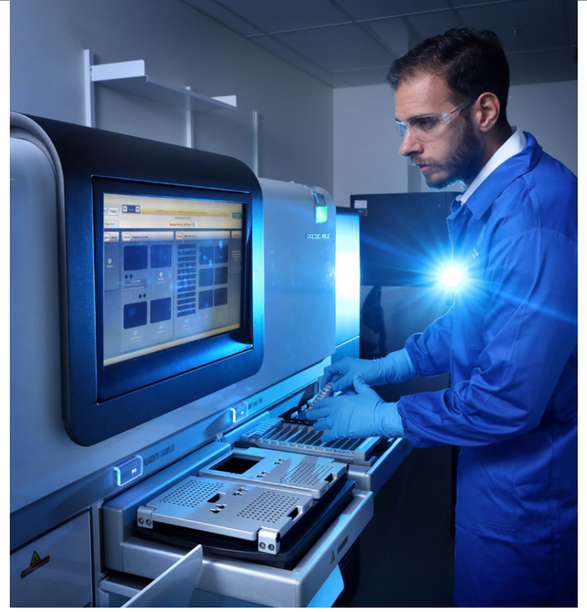
Alternates

Professor Steven Djordevic
University of Technology, Sydney

Professor Claire Wade
University of Sydney

The Centre thanks past committee members Professor Peter Lovibond (UNSW), Professor John Mattick (Garvan Institute of Medical Research), Professor Deborah Marsh (Kolling Institute), Professor Eddie Holmes (USyd) and Professor Nicki Packer (Macquarie University) for their service on the committee.

Research Collaboration Highlights



Koala Genome Project: Australian scientists crack the genetic code

Centre staff were among a team of Australian and international scientists who sequenced and assembled the genome of the koala. This has provided new insights into the unique biology of the koala and will inform conservation efforts, aid in the treatment of diseases, and help to ensure the koala's long-term survival.The consortium of scientists collaborating on the project comprised 54 scientists from 29 different institutions across seven countries, led by Professor Rebecca Johnson, Director of the Australian Museum Research Institute and Professor Katherine Belov, University of Sydney. The findings were published in Nature Genetics in August 2018.

The koala genome, which is slightly larger than the human genome (3.5Gb v 3.2Gb), was sequenced by the Centre using PacBio long-read sequencing technology. The project took 10 months to complete, required 272 PacBio SMRT cells and generated 200 Gb of data.

The genome was assembled by the UNSW Systems Biology Initiative (SBI). "We sequenced and then assembled the genome with supercomputers, allowing the consortium to study the >26,000 genes of this unique species," said Professor Marc Wilkins, Director of both the Ramaciotti Centre and the SBI.

"We did so using long-read sequencing allowing us to produce a very high-quality genome assembly – meaning that the result is the best marsupial genome to date, and one that is on par with the human genome in terms of its quality, which is incredibly exciting".

"Because of its high quality, this genome is now a fundamental resource for all the other marsupial genomes which have yet to be generated and studied. We will be able to use this as a reference for the entire marsupial community."

Professor Johnson said: "The Koala Genome Consortium has been an ambitious journey, affording us great insights into the genetic building blocks that make up a koala – one of Australia's, as well as the world's, most charismatic and iconic mammals.

"This milestone has come from our vision to use genomics to conserve this species. The genetic blueprint has not only unearthed a wealth of data regarding the koala's unusual and highly specialised diet of eucalyptus leaves, but also provides important insights into their immune system, population diversity and the evolution of koalas," she said.

According to Professor Belov, another important discovery was the characterisation of the composition of koala milk. Like all marsupials, koalas do most of their development in the pouch. They are born without an immune system after 34-36 days gestation and spend ~6 months developing in the pouch.

"We characterised the main components of the mother's milk – which is crucial for koala joeys," Professor Belov said. "We identified genes that allow the koala to fine-tune milk protein composition across the stages of lactation, to meet the changing needs of their young."

"Thanks to the high-quality genome, the team was able to analyse and discover koala-specific milk proteins that are critical for various stages of development. It also appears these proteins may have an antimicrobial role, showing activity against a range of bacterial and fungal species, including Chlamydia pecorum, the strain known to cause ocular and reproductive disease in koalas," Professor Belov said.

Chlamydia causes infertility and blindness and has severely impacted koala populations in New South Wales and Queensland. Using information gained from the koala genome, scientists hope to develop a vaccine to fight diseases like chlamydia.

One of the most threatening processes to koala survival is loss of habitat through land clearing and urbanisation, which results in a reduction of habitat connectivity, reduced genetic diversity and puts koalas at high risk of inbreeding. The results of inbreeding can be highly detrimental to the survival of those koala populations, as it leads to reduced genetic diversity.

"For the first time, using over 1000 genome linked markers, we are able to show that NSW and QLD populations show significant levels of genetic diversity and long-term connectivity across regions," Professor Johnson said.

"Ensuring this genetic diversity is conserved in concert with other conservation measures to protect habitat, reduce vehicle strikes, dog attacks and disease is the key to the long-term survival of the koala."

All of the sequence data generated by The Koala Genome Consortium has been deposited into public databases and made freely available to scientists around the world.

The story made headlines around the world, being picked up by CNN, National Geographic, New Scientist, the ABC and The Australian and positioned the Centre as the leading facility in Australia for the sequencing and assembly of *de novo*, mammalian genomes.

> "Because of its high quality, this genome is now a fundamental resource for all the other marsupial genomes which have yet to be generated and studied."

ASPREE Study: Analysis of 15,000 DNA samples from landmark aspirin trial

In June 2019, the Centre completed its largest single study to genotype 15,000 individuals, and one of the largest genetic screens of this type done in Australia to date. This analysis was part of Monash University's expansion on its landmark aspirin trial ASPREE (ASPirin in Reducing Events in the Elderly), whose participants had their health comprehensively tracked over a number of years.

The ASPREE study pioneers detailed research into genetic factors that contribute to good health or disease in the elderly and aims to gather as much genetic information about trial participants as possible to inform the trial and ultimately improve participants' outcomes.

The project utlised genome-wide single nucleotide polymorphism (SNP) analysis, using the Axiom Precision Medicine Diversity Array. Dr Paul Lacaze, Head of Public Health Genomics, Monash University, said the project could help identify genetic markers associated with a positive or negative response to therapies.

"It is possible that underlying genetic factors may explain why some people respond to drug therapies and others do not. If we can discover and understand these factors more clearly, it may help guide drug use in the future, maximising the benefits and minimising the risks for older people."

This partnership between the Ramaciotti Centre, Monash University and Bioplatforms Australia will complement extensive clinical and lifestyle information collected in the ASPREE trial and associated studies, creating a powerful resource for biomedical research.

Andrew Gilbert, General Manager of Bioplatforms Australia said "ASPREE is a significant international collaboration that we are extremely excited to see enhanced by Bioplatforms' genomics capability. This project will enable new research across a broad range of diseases and investigate the use of genetic data towards improved public health outcomes."

Monash University and Bioplatforms are funding the development of the $1 million genetic data resource, with analysis of this data expected to continue for several years.

Dr Lacaze said the new partnership was an example of leading Australian research groups coming together to generate new genetic research into a range of health problems affecting the elderly, such as cancer, diabetes, dementia and age-related macular degeneration.

"The project will generate much-needed evidence to help inform whether genetic factors may play a role in guiding more effective therapeutic and prevention strategies in years to come, bringing us closer to precision medicine."

Deputy Director and Centre Manager Dr Helen Speirs said "We are incredibly proud of this achievement. A huge thank you to the array team whose hard work and dedication saw the project completed within 8 months."

UNSW Launches the Cellular Genomics Futures Institute

In 2018, UNSW announced the investment of $200M in new and emerging areas of research which build on and link its existing strengths. Known as UNSW Futures, these initiatives present a bold new framework to address humanity's major challenges through innovative inter-disciplinary and cross-faculty research.

The first four of these Futures Institutes was announced by the President and Vice Chancellor Professor Ian Jacobs and the Deputy Vice Chancellor Research Professor Nicholas Fisk. The Ramaciotti Centre for Genomics is a partner in one of these initiatives, the UNSW Cellular Genomics Futures Institute, which is focused on precision medicine, specifically in the area of cellular genomics.

The UNSW Cellular Genomics Futures Institute brings together key researchers from the faculties of Medicine, Science and Engineering, including the Ramaciotti Centre for Genomics and the Kirby Institute. It leverages the scientific leadership of UNSW

"a bold new framework to address humanity's major challenges through innovative inter-disciplinary and cross-faculty research"

Sydney and the Garvan Institute of Medical Research in the field of single cell genomics, tackling the major challenges in single cell data acquisition and data analytics.

The Futures Institute aims to invent technologies that will allow it to decode the state of DNA, chromatin, and its entire output as RNA and protein in thousands of single cells, uncovering diagnostic cell origins, molecular targets, and treatments for human disease with unparalleled precision. This will have a major impact on the precision health management of patients, through early detection and personalised treatment of disease, and will reduce the costs of healthcare.

Professor Chris Goodnow, Executive Director of the Garvan Institute of Medical Research and Director of the UNSW Cellular Futures Institute said "Many diseases such as cancer and autoimmune disease arise from genetic changes in only one or a few cells in a large population. Until recently, technological limitations have meant we have had to rely on bulk methods where we analyse millions of cells often masking the individual variation that exists between one cell and the next, limiting the diagnosis and precision treatment of diseases."

"Rapid advances in single cell technology combined with computational informatics are allowing us for the first time to uncover the cellular origin and evolution of disease. The UNSW Cellular Genomics Futures Institute will invent technologies that will be used for precise diagnosis and treatment of human disease. Single cell genomics is the next great revolution in medicine."

Professor Marc Wilkins, Director of the Ramaciotti Centre and Executive team member of the Cellular Genomics Futures Institute said "We look forward to this exciting new initiative and the impact that it will have in single cell science."

Journal article acknowledgments

» **2018** 134

» **2019** 146

Publications

The Ramaciotti Centre supports a diverse range of research across a variety of organisations. This is evident in the peer reviewed publications that acknowledge the Centre as the genomic service provider. In 2018 and 2019, the Centre was acknowledged in 134 and 146 journal articles, respectively. We thank the authors for their acknowledgement of the Centre's contribution.

Examples of high impact publications that feature genomic data generated by the Ramaciotti Centre include:

Natural regulatory mutations elevate the fetal globin gene via disruption of BCL11A or ZBTB7A binding

Professor Merlin Crossley and team
Journal of Nature Genetics | 2018 | Vol 50, 498-503

Genomic Service: Chromatin Immunopreciptation (ChIP) sequencing
Background: β-hemoglobinopathies such as sickle cell disease and β-thalassemia result from mutations in the adult haemoglobin gene. Reactivating the developmentally silenced foetal haemoglobin genes is a therapeutic goal for treating these diseases. The authors used CRISPR gene editing to introduce beneficial natural mutations into blood cells to boost their production of foetal haemoglobin. The research also solves a 50-year-old mystery about how these mutations – which are naturally carried by a small percentage of people – operate and alter the expression of human genes.

Draft genome assembly of the invasive cane toad, Rhinella marina.

Professor Peter White and team
Gigascience | 2018 | Vol 7 (9), giy095

Genomic Service: PacBio Whole genome sequencing
Background: The cane toad (Rhinella marina formerly Bufo marinus) is a species native to Central and South America that has spread across many regions of the globe including Australia. Cane toads are known for their rapid adaptation and deleterious impacts on native fauna in invaded regions. However, despite an iconic status, there are major gaps in our understanding of cane toad genetics. The authors report a draft genome assembly for R. marina, the first of its kind for the Bufonidae family. The availability of the genome will help to close these gaps and accelerate cane toad research.

"The availability of the genome will help to close these gaps and accelerate cane toad research."

Beyond the panel: preconception screening in consanguineous couples using the TruSight One "clinical exome".

Professor Edwin Kirk and team
Genetics in Medicine | 2019 | Vol 21, 608-612

Genomic Service: Exome panel sequencing.
Background: The study aimed to provide proof of concept by broadening preconception screening beyond targeted testing to inform reproductive risk in consanguineous couples. The authors concluded that preconception screening of consanguineous couples for recessive and X-linked disorders using genomic sequencing is practicable and is likely to detect many more at-risk couples than a targeted panel could achieve.

Specific Bacteria and Metabolites Associated with Response to Faecal Microbiota Transplantation in Patients with Ulcerative Colitis.

Dr Nadeem Kaakoush and team
Gastroenterology | 2019 | Vol 157 (4) 1165-116

Genomic Service: 16S amplicon sequencing and shotgun metagenomic sequencing.
Background: Faecal microbiota transplantation (FMT) can induce remission in patients with ulcerative colitis (UC). In a randomized controlled trial of FMT in patients with active UC, the authors identified bacterial taxonomic and functional factors associated with response to therapy. These findings may be used in design of microbe-targeting therapies for ulcerative colitis.

Operational Excellence

"we couldn't be prouder of our hardworking team of professional staff who achieved this in such a short time"

Laboratory Accreditation: Ensuring quality and performance

The Centre was accredited to ISO/IEC 17025 by the National Association of Testing Authorities (NATA) - Australia's national laboratory accreditation authority on the 29th April 2019. Accredited services include next generation sequencing of client prepared libraries and genotyping by microarray. Producing test results that are valid and trusted is at the heart of the Centre's laboratory activities. Accreditation to ISO/IEC 17025:2017 means that we have met the management and technical requirements of the standard and are deemed technically competent to produce valid and reliable results.

The Centre's Quality Manager Keren Natalia said "The journey towards ISO/IEC 17025 accreditation started in early 2018 and we couldn't be prouder of our hardworking team of professional staff who achieved this in such a short time, and with such ease and grace, on top of their already large workload."

Getting to an audit-ready state was no mean feat. In speaking to our team, it is no surprise that many found it daunting and challenging, especially at the beginning. The process was certainly arduous and really pushed everyone's capacity to the limit – juggling project commitments and day-to-day business demands whilst trying to document, implement, and improve processes to meet the requirements of the Standard.

Looking back, the team all agree that as difficult a task it was, it was worthwhile. It helps us analyse our practices in a systematic manner, provides structure and framework to ensure the quality of our ever-growing business, and gave us even more confidence in our processes and the services we provide to those who use our services.

It is our mission to deliver internationally competitive genomic services by providing our clients with access to technology and services of the highest quality. This accreditation is yet another demonstration of our commitment and a testament to the dedication of our team which has worked very hard in ensuring our success. Well done, team!

NATA
WORLD RECOGNISED ACCREDITATION

Our Quality Policy

We are committed

to providing internationally competitive, high quality genomic services.

We do this through:

- Consistently providing quality analytical services to our clients through data that meets or exceeds manufacturer's specifications.
- Ensuring that all personnel are competent and qualified for the tasks they perform.
- Developing, maintaining and applying a quality management system according to the ISO/IEC17025:2017 Standard.

Our Quality Objectives

To ensure:

- All personnel are familiar with and implement the quality system documentation and policies required to professionally and effectively perform procedures that produce accurate and precise results.
- The Ramaciotti Centre's standard of service and specifications are defined, documented, managed and maintained.
- The quality system is maintained and continually improving.

At the Leading Edge: Illumina and MGI sequencing platforms added to portfolio

Providing access to leading edge enabling technology is core to what we do. We monitor trends and work closely with technology companies to remain at the forefront of change.

In 2018, we expanded our Illumina sequencing capacity to include the newly released iSeq and another MiSeq. The iSeq is the smallest sequencer manufactured by Illumina, with an output of up to 1.2 Gb / 4 million reads. It has proved to be a useful tool for the quality control of library pools prior to sequencing on larger more expensive runs and for projects that need a small amount of data. MiSeq sequencers are workhorses of the Centre, running 24/7 throughout the year, with an average of 295 runs per annum. The addition of the third MiSeq has reduced the turnaround time for one of our high sample volume services, the amplicon sequencing service.

MGI, a division of BGI, emerged as a new and developing player in the short-read sequencing instrument market in 2016 and began selling into the Asia Pacific region in early 2018. MGI technology, an alternative to Illumina short read sequencing, uses rolling circle replication to amplify small fragments of genomic DNA into DNA nanoballs. To evaluate the technology, specifically its application to single cell sequencing, the Centre acquired an MGI DNABSEQ G400 in December 2019. The instrument has an output of up to 720 Gb / 1.8 billion reads making it a good fit for single cell libraries or for small numbers of genomes or transcriptomes. Evaluation of the data quality and reliability of the instrument will be conducted throughout 2020.

Acquisition of these platforms, along with that of the Illumina NovaSeq 6000 and PacBio Sequel in 2017, has positioned the Centre as the largest and most comprehensive genomics Centre at any Australian university and as the technological and technical hub for genomics in the state of NSW.

Improving our Client Experience: Client interaction and engagement

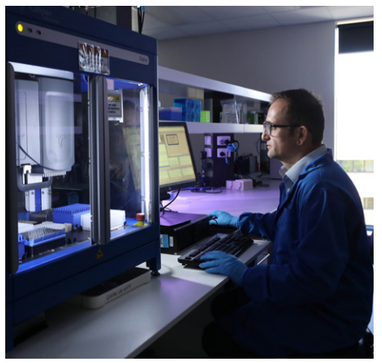
Our Research Managers and Research Officers spend a considerable part of their day communicating with clients via email, providing project planning and design advice, quotes, sample submission guidance and regular project updates. To provide clients with a better experience throughout this process, the Centre has implemented a Customer Relationship Management (CRM) software package, Microsoft Dynamics 365. The CRM brings together all the communication from a client in one view giving a complete overview of the background to an enquiry or project. This improves response times and ensures that we understand the client's requirements. Key staff have been working with the UNSW CRM team to bring about further improvements such as integrated quotes, automated replies, standardised response emails and knowledge articles for clients. Integration with the UNSW finance system is planned for 2020.

A dynamic new Centre website was launched in April 2019. Users of the Centre were involved in the design process and helped to guide the overall layout. Feedback indicated that the website is where researchers go for information on how to submit samples, find out what is new and see what genomic events are on in their local area. The new design features many improvements in the accessibility of this information.

"Ramaciotti Centre staff are passionate about the environment and have an ongoing commitment to reducing the environmental footprint of the Centre."

Automation of Laboratory Processes

The Centre continues to automate laboratory processes to increase throughput and reduce the cost per analyses. A successful application to the UNSW Research Infrastructure Scheme in 2018 funded the acquisition of Sciclone G3 NGSx iQ and Zephyr G3 384 liquid handling instruments from Perkin Elmer. The systems were installed late 2019 and will be used for microarray assays and library preparation for next generation sequencing. This brings the total number of liquid handling system in the Centre to five with further automation purchases planned for 2020.



Embracing the e-Notebook

As part of our ongoing commitment to maintain records of the highest standard, we have replaced all laboratory notebooks and paper documents with e-notebook records using LabArchives software. We use LabArchives to store all metadata generated by a project including quotes, sample receipt and sample and intra-assay quality control data. The e-notebook complements our Laboratory Information Management System (LIMS) that is used to track samples and reagents throughout a process. All staff have tablets, making data entry tasks in the laboratory straightforward. This ensures that we maintain records of the highest standard. LabArchives also allows us to easily share metadata with our clients when additional data is required for publication or with suppliers when troubleshooting instrument or reagent issues.

Green Impact Award — Reducing our environmental footprint

The Ramaciotti Greenomics team placed second at the inaugural UNSW Green Impact awards. These awards celebrate the incredible efforts of students and staff to make office and residential spaces more sustainable. In total 200 individuals across 35 teams completed 768 actions to embed sustainability within the workplace.

The awards presented on the 8th November 2019 were created by the UNSW SMaRT Centre and in keeping with the sustainability theme were made from waste plastic, fabric and glass.

Ramaciotti Centre staff are passionate about the environment and have an ongoing commitment to reducing the environmental footprint of the Centre. Specific actions implemented by the Ramaciotti Greenomics team included implementing the use of recyclable tip boxes that reduce plastic waste by 50%, successfully campaigning for polystyrene eskies to be recycled by UNSW, powering down computers and equipment at the end of the day, becoming almost completely paper free in the laboratory and office, using keep cups and supplying reusable cutlery and crockery at all meetings and events. The impact on the environment will continue to be a consideration on all consumable, reagent and instrument purchases.

"What the Green Impact initiative has shown is that among students and staff, and at every level of our university, we are prepared to lead on environmental sustainability," said Professor Anne Simmons, UNSW Provost.

"A place like UNSW that holds such rich knowledge of climate science and renewable energy, and which has a true commitment to having a positive impact on society, must lead by example."

In line with UNSW's sustainability policy, the Centre is committed to continuing to find ways to reduce its environmental footprint. Sustainability is a standing item on the agenda of our weekly meetings where we discuss ideas on how we as a team can contribute to change. Ramaciotti Greenomics is here to stay.

Operational Report

Genomic Technology

The Centre has a comprehensive technology portfolio that supports a wide range of genomic services. It is the largest and most comprehensive genomics Centre at any Australian university. The Centre's technology suite includes long- and short-read next-generation sequencing, single-cell analysis, microarrays and capillary sequencing.

Short Read Next Generation Sequencing
- Illumina NovaSeq 6000
- Illumina NextSeq 500
- Illumina MiSeq
- Illumina iSeq 100
- MGI DNBSEQ-G400

Long Read Next Generation Sequencing
- PacBio Sequel

Sanger Sequencing
- AB3730

Gene Expression & Genotyping
- Life Technologies GeneTitan
- Fluidigm BioMark
- NanoString nCounter

Single Cell Genomics
- 10X Genomics Chromium
- Fluidigm C1 single cell

Services Offered

The Ramaciotti Centre offers a wide range of genomic, transcriptomic and epigenomic services. All genomic analyses performed by the Ramaciotti Centre are carried out in our ISO/IEC 17025 accredited laboratories. Services offered to the researched community include:

Next Generation Sequencing
Genome
Exome, targeted and panel
Epigenome
RNA sequencing
Single cell sequencing
Microbiome and amplicon
Metagenome

Sanger Sequencing
Core and user prep
Fragment analysis
Custom design & validation

Microarray
Gene expression
Transcriptome
Genotyping

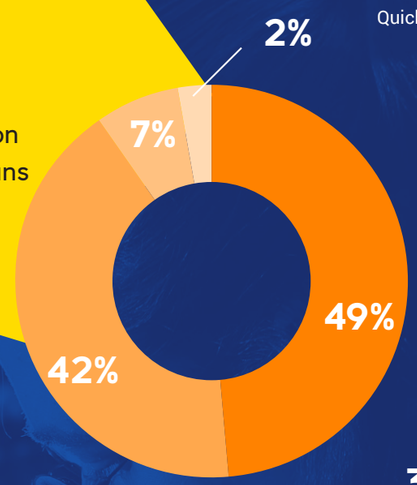
Other Services
Gene expression by qPCR or NanoString
Genotyping by qPCR

Services Provided

The Centre has an outstanding track record in service provision, consistently delivering high quality genomic and transcriptomic services to the research community and to industry.
Quick stats by year:

2018

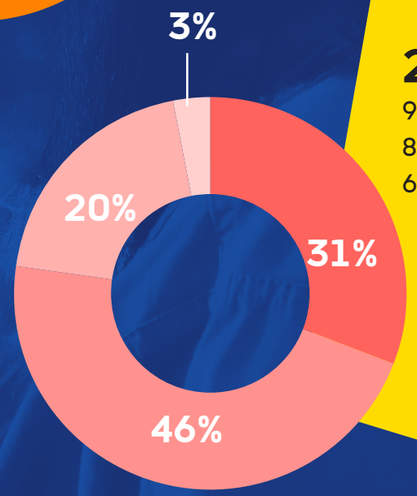
1,135 projects
68,041 samples
533 next generation sequencing runs



33,179 samples sequenced by Sanger sequencing
28,378 samples sequenced by next generation sequencing
4,720 samples analysed by microarray
1,764 samples analysed using other genomic technologies

2019

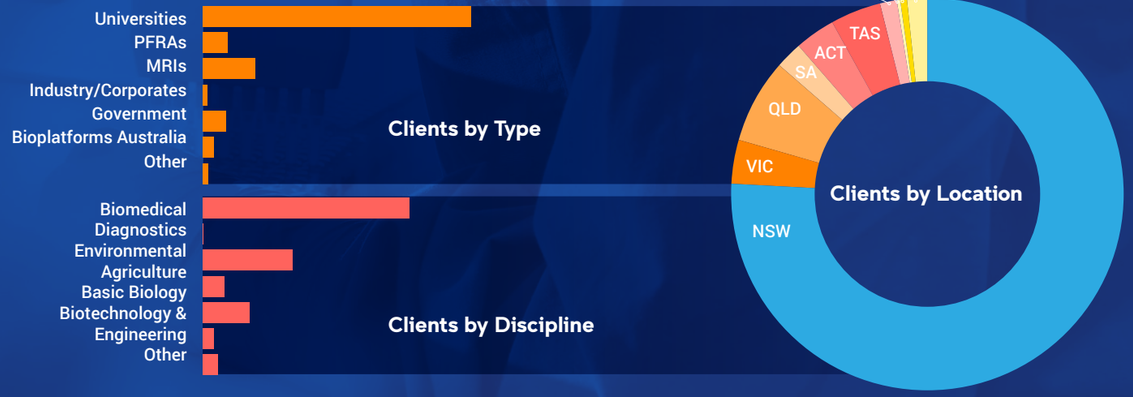
965 projects
81,087 samples
603 next generation sequencing runs

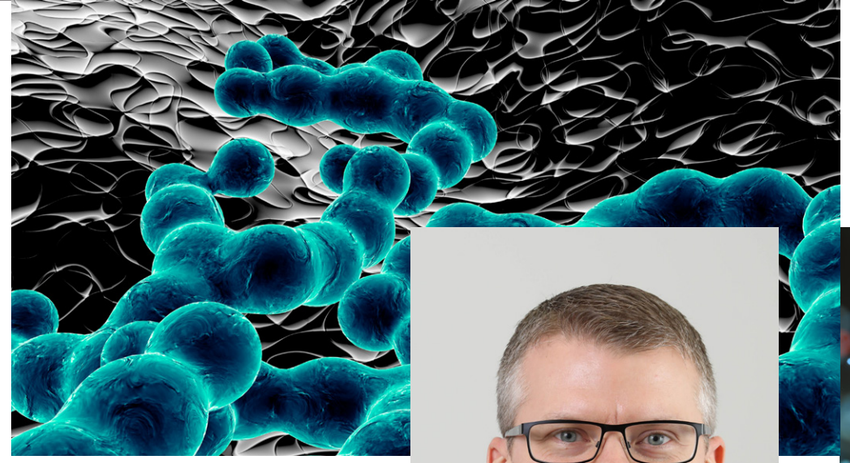


25,068 samples sequenced by Sanger sequencing
37,634 samples sequenced by next generation sequencing
16,006 samples analysed by microarray
2,379 samples analysed using other genomic technologies

Client Profile

The Centre has a client base of over 500 research groups representing over 1,000 individual researchers from academia and industry. We serve all the leading universities and research institutes in Australia and support a wide range of research covering many disciplines.





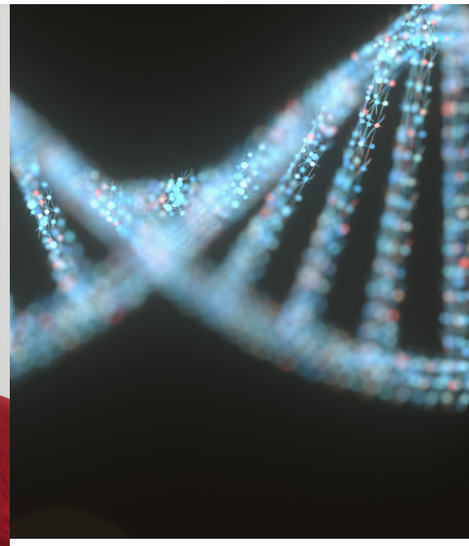
Services in the Spotlight

A Service with Many Names

It is referred to as microbiome analysis, microbial or taxonomic profiling or amplicon sequencing. Call it what you will, this genomic service crosses more disciplines than any other service and routinely handles a large volume of samples. It has also perhaps sequenced some of the most interesting samples including, seal guts, whale snot, wild wine ferments and a whole lot of poo.

Jason Koval who heads up the amplicon sequencing team said "The fact that the technique is amenable to so many different sample types keeps the job interesting. Hearing from researchers on the application of amplicon data to projects such as soil remediation in fragile environments like Antarctica, conservation of the Great Barrier Reef, or its use by forensic teams to identify the geographical origin of soil contamination at a crime scene makes it worthwhile. A challenge moving forward will be keeping up with the demand for this type of analysis, since our Illumina MiSeq's are running 24/7."

The team is involved in many national collaborative projects generating data for researchers using a range of pre-optimised gene region targets for the 16S, 18S and ITS genes. They also assist with the design and delivery of custom amplicon sequencing projects. The team's reputation in this area is something that the Centre is very proud of.



RNA Sequencing in All its Forms

The RNA sequencing service manages some of the most challenging project design requests and handles a variety of difficult sample types. Our RNA services include, mRNA sequencing, total RNA sequencing, low input RNA and miRNA sequencing and targeted RNA sequencing, with all services being compatible with RNA extracted from many different sample types.

Dr Helena Mangs who leads the RNA team said "Working with RNA from so many sources such as tissue, blood or serum, exosomes or sorted cells from a vast array of species for example plants, bacteria or RNA of environmental origin, makes our job interesting, as different approaches are needed for each. The team have enjoyed the challenge of working on several large plant and mammalian RNA sequencing projects, as well as on the intricacies of smaller custom-designed low input and miRNA sequencing projects. We have also been involved in several studies complementary to RNA sequencing, such as chromatin immunoprecipitation (ChIP) sequencing and methylation sequencing for both mammalian and plant samples. One day in our service is never the same as the next!"

The RNA sequencing team's breadth of knowledge and ability to make projects that are technically challenging possible have earned them a reputation for project delivery in this area that is world class.

New Services

The Centre has a reputation for staying ahead of the curve by delivering genomic services using newly released kits or techniques that are technically challenging. We do our very best to support the research community with new and emerging techniques and enjoy the challenge of working on bespoke projects that challenge our technical skills. Some of those that were tested and implemented during the period include: Dovetail – for genome scaffolding, TraDIS – sequencing of transposon introduced mutations, and single nucleotide variation design and validation using Sanger sequencing.

"a reputation for staying ahead of the curve by delivering genomic services using newly released kits or techniques that are technically challenging."

4 seminars hosted
3 workshops organised
9 educational tours
13 conferences supported
4 trade shows

Events and Engagement

As part of our commitment to the Australian genomics community, we organise educational workshops, special interest group meetings, seminars and support scientific meetings and conferences.

Engaging the broader community in genomics research and its impact on society is part of what we do, by providing facility tours for indigenous students from around Australia attending the UNSW Nura Gilli Winter school, work experience students from high schools in the Sydney area, and high school teachers seeking to remain up to date with developments in this area.

Activities in 2018 and 2019 included:
Number of seminars hosted 4
Number of workshops organised 3
Number of educational tours 9
Number of conferences supported 13
Number trade shows 4

Grants and Funding

National Collaborative Research Infrastructure Scheme Funding Renewed

For almost 10 years, the Centre has been fortunate to receive funding from National Collaborative Research Infrastructure Scheme (NCRIS) to support its activities, albeit on a year by year basis. Following on from a Federal Government consultation, review and roadmap process, a further 4-year tranche of funding was released to administrators such as Bioplatforms Australia. The Centre, regarded as an exemplar of collaboration and research support during the review process, was awarded continued funding to support operations from July 2019 – June 2023.

ARC Linkage, Infrastructure and Equipment Funding

Towards the close of 2019 the Ramaciotti Centre consortium was awarded its 17th ARC Linkage, Infrastructure and Equipment Funding (LIEF) grant. The grant, led by Professor Brett Neilan, University Newcastle, will support the acquisition of equipment at the University of Newcastle, Macquarie University, University of Technology, Sydney and at UNSW. The Centre's funding allocation will support the acquisition of an Oxford Nanopore GridION long read sequencer in 2020.

UNSW Research Infrastructure Scheme

The Centre successfully applied to the UNSW Research Infrastructure Scheme in 2018 and 2019 for equipment support. The scheme along with ARC LIEF is an important source of infrastructure funding for the Centre, supporting the purchase of instruments that are the backbone of genomic service provision. A total of $314,000 from both rounds will support the purchase of much needed liquid handling platforms to increase throughput and reduce hands on time.

BIOPLATFORMS AUSTRALIA

NCRIS National Research Infrastructure for Australia
An Australian Government Initiative

Australian Government
Australian Research Council

Acknowledgements

The work that we do would not be possible without the support from our partners. The Ramaciotti Centre gratefully acknowledges the support it receives from NCRIS and the co-contributions made by the University of New South Wales

UNSW SYDNEY

UNSW Ramaciotti Centre for Genomics