AGRF REPORT

2021

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AGRF REPORT 2021

AGRF improves quality of life through exceptional life science.

This is the first report that I have been involved in since returning to the board of AGRF and I must say that it is a delight to be back and to be a part of the exciting events of the past couple of years.

The pandemic has not passed us without effect. As many of our clients are university-based researchers, these have been hard hit by the pandemic, its effect on the finance of universities and ultimately on the funds available to them from their university. This has had flow-on effects for us. However, having said this, we have had a very successful 2020-2021, including many fruitful collaborations.

Our role in the Autism Collaborative Research Centre has seen us sequence the whole genomes of over 1,500 individuals over a 5-month period.

We analysed around 2,700 samples for the Telethon Kids Institute from Perth to help rapidly accelerate the development of vaccines.

The Children's Cancer Institute in Sydney has welcomed us as its sequencing partner in a very ambitious and exciting program to sequence the whole genomes of children with cancer.

A collaboration with the University of Queensland (UQ) resulted in a grant that launched a long-read sequencing facility situated in our laboratories at UQ.

And post-June 2021 but before publication of this report, AGRF was appointed as the preferred provider of sequencing services for the Westmead Genomics Hub.

With significant improvements in workflows and capacity, we have reduced our turnaround times, allowing our clients to make faster decisions. This is particularly beneficial to our clinical and commercial clients, where a faster result can have a powerful impact.

A new three-year strategic plan was formulated with a major focus on clinical genomics. This has allowed a rethink on our direction and will challenge the organisation over the next couple of years.

- Professor Simon Foote

CHAIRMAN'S MESSAGE

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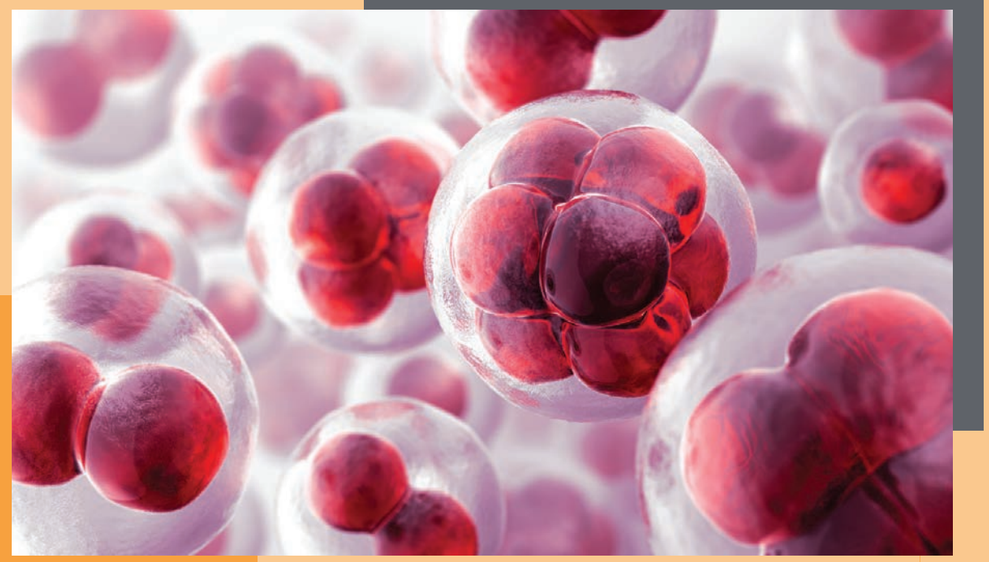

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A WORD FROM OUR OUTGOING CEO

DR KIRBY SIEMERING

The last two years have been a transformational period for AGRF, albeit forged in adversity. The COVID-19 pandemic changed the world abruptly, and with it our work and that of many of our partners and colleagues.

As an essential service to the Australian scientific community, AGRF has continued to operate in challenging circumstances throughout the pandemic, playing a vital role in Australia's response: from establishing accredited testing infrastructure, to supporting vaccine development efforts, to participating in research attempting to understand disease severity.

AGRF is proud to have also supported some of Australia's most important genomics projects throughout this period. In the space of just five months, we have sequenced the full genomes of more than 1,500 participants from the Australian Autism Biobank and genotyped around 2,700 individuals for the Human Immunology Consortium Project.

Building on these successes, AGRF has recently begun work as the genomics platform partner for the Zero Childhood Cancer Program – an amazing project that aims to extend genomic testing to every child, adolescent, and young Australian with cancer. We continue to work with a range of other clinical partners to deliver innovative solutions to clinicians and patients in the emerging era of precision medicine.

In the background, AGRF is working hard to continue building our capability and infrastructure capacity to ensure more projects of international scale and significance can be carried out in Australia. But AGRF is more than simply a provider of infrastructure – we work on a deep collaborative level with our partners in both academia and industry to deliver innovative technologies and solutions. We have forged new partnerships with the Westmead Research Hub and the South Australian Genomics Centre and have built on our existing partnership with the University of Queensland to deliver a new long-read sequencing capability.

Genomics is now making a significant impact in environmental science and AGRF is contributing to several exciting programs including as a partner in the CRC Transformations in Mining Economies, working together with researchers and mining companies on the ecological rehabilitation of mine sites. We are also a partner in a variety of important Bioplatforms Australia Framework Initiatives developing a genomic resource for Australia's unique flora and fauna, including Oz Mammals Genomics, Genomics for Australian Plants, and the Threatened Species Initiative.

AGRF has a long-standing history of working on livestock and crop improvement with a variety of partners in agricultural science. Building on our previous program of work with the aquaculture industry, we are a partner in the new ARC Research Hub for Supercharging Tropical Aquaculture Through Genetic Solutions which will make the Australian aquaculture industry globally competitive.

AGRF now stands on strong foundations and the opportunities that lie in front of us are exciting. We highly value the support of the genomics community and our funding partners, the National Collaborative Research Infrastructure Strategy and Bioplatforms Australia, and look forward to sharing the next chapter with you.

I would like to take the opportunity to thank our members and Board of Directors for their unwavering commitment in guiding the success of AGRF. Finally, it has been an exceptionally challenging period for our staff having to work through difficult circumstances and I am incredibly proud of the efforts of the whole AGRF family and how they have pulled together to achieve amazing results.

PROF. SIMON FOOTE

Chairman
BMedSci, MBBS, PhD, DSc, FFSc (RCPA), FAA, FAHMS, FTSE
Member of the Finance, Audit & HR Committee

Professor Foote is an Emeritus Professor at The John Curtin School of Medical Research at The Australian National University. He has been Director of the John Curtin School, Dean of the School of Medicine at Macquarie University, Director of the Menzies Research Institute at the University of Tasmania and Divisional Head at the Walter and Eliza Hall Institute, Melbourne. Professor Foote was elected to the Australian Academy of Science in 2016 and was a founding member of the Australian Academy of Health and Medical Sciences (AAHMS), after being elected to the Academy in 2014, and is also a Council Member of the AAHMS. In addition, he was elected as a Fellow of the Australian Academy of Technological Sciences and Engineering (ATSE) in 2009, and he was a postdoctoral fellow at the Whitehead Institute at the Massachusetts Institute of Technology.

PROF. NICK SAMARAS

Deputy Chairman
BSc (Hons), PhD, MBA, FAIM, FAICD, FWCLP
Member of the Finance, Audit & HR Committee

Professor Samaras has worked in senior positions of several global life sciences companies for over 30 years. Professor Samaras has served on the boards of several Australian-based technology companies and is currently the chairman of Genetic Signatures Ltd. He is also an Enterprise Professor at the University of Melbourne and Professor (Practice) at Monash University. He has extensive experience in the global life sciences and advises the AGRF Board on current technological advances, market trends and industry engagement.

DR JOHN BELL

BSc, MSc, PhD, FTSE, FRACI, Comp I.E. Aust, MAICD

Dr Bell is a Senior Associate with ACIL Allen. His previous experience includes working as a Senior Policy Adviser to the Australian Academy of Technological and Engineering (ATSE), Deputy Secretary and Chief Science Advisor in what is now the Department of Industry, Science, Energy and Resources. He was a member of the Industry R&D Board and the Australian Research Council and served short periods as Acting Director of ANSTO and AIMS. Dr Bell provides an invaluable service to the AGRF Board advising on government policy and assisting in liaison with government agencies.

PROF. GABRIELLE BELZ

BVBiol, BVSc, PhD, DVSc, FAHMS, MAICD

Professor Belz is NHMRC Senior Research Fellow. She joined the AGRF Board in May 2017 as a representative from Walter and Eliza Hall Institute of Medical Research and The University of Melbourne. With over 20 years of experience in medical research, Professor Belz provides significant knowledge and expertise to AGRF.

OUR GOVERNANCE

PROF. BENJAMIN KILE

BSc (Hons), LLB (Mon), PhD, FAHMS
Member of the Finance, Audit & HR Committee

Professor Kile commenced as Executive Dean, Faculty of Health and Medical Sciences, University of Adelaide in October 2019. Prior to this, he was Head of the Department of Anatomy and Developmental Biology, Monash University (2017-2019), and Head of the ACRF Chemical Biology Division at the Walter & Eliza Hall Institute of Medical Research (WEHI) (2014-2017). Ben conducted his PhD training in the Cancer and Hematology Division at WEHI, and postdoctoral research at Baylor College of Medicine in Houston, USA. He returned to WEHI in 2004 and established an independent research laboratory there in 2008. Professor Kile is a medical researcher internationally recognised for his work in blood cell development and survival, particularly in the context of cancer chemotherapy and its impact on the hematopoietic system.

MR ANDREW MACDONALD

BSc, BBus, CPA, MAICD.
Chairman of the Finance, Audit & HR Committee

Mr Macdonald has over 30 years of commercial experience, working across the biotechnology, technology and finance sectors in Australia, the US and the UK. In his role as Chair of the Finance, Audit & HR Committee, Mr Macdonald provides significant contributions in the areas of business and finance.

PROF. BRANDON WAINWRIGHT AM

BSc (Hons), PhD

Professor Wainwright is an Affiliate Professor and Group Leader at the Institute for Molecular Bioscience at The University of Queensland. As well as acting as a representative of The University of Queensland, he provides services to the AGRF Board as a liaison with the Queensland Government.

PROF. INGRID WINSHIP AO

MBChB, MD (Human Genetics), FRACP, FACD, FAICD

Professor Winship has a Doctorate of Medicine from the University of Cape Town (1986) and accreditation as a Clinical Geneticist from the Human Genetics Society of Australasia (1995). She is currently a Chair of the Australian Health Ethics Committee and a member of the NHMRC Council. She is a director of the Boards of Global Variome and the Human Genome Organisation (HUGO). Professor Winship was appointed Officer of the Order of Australia in 2020 for distinguished service to medicine, particularly to clinical genetics and research, to cancer prevention, and as a role model and mentor. She has more than 35 years' experience working in medicine and clinical genetics, bringing a wealth of knowledge and expertise to AGRF.

OUR EXECUTIVE TEAM

KIRBY SIEMERING, PHD

Chief Executive Officer

Dr Siemering has been with AGRF for almost two decades, starting his time as a scientist in Research & Technology and ultimately being appointed as AGRF CEO in 2019. Dr Siemering has more than 25 years' experience as an executive manager in life sciences and genomics in both academic and commercial settings. He has held senior positions in academia and ASX-listed and privately held biotechnology companies and is experienced in biotechnology industry analysis, strategy, and policy development at State Government level.

DAVID BENALLACK

Chief Operating Officer

David has over 20 years of experience within the industry with a further 13 years in the chartered environment where he was a director of Grant Thornton. He has worked across a variety of spheres including corporate travel, recruitment, auction services, Local Government and within the NFP sector. David's past roles have been predominantly ASPAC and C-suite, and he currently holds both COO and Company Secretary positions at AGRF, joining the company in 2018.

MARIA RICCI

Chief Business Officer

Maria has more than 25 years of extensive experience in new and emerging markets across the life sciences and in vitro diagnostics industries. Maria joined AGRF in 2017 and has held senior positions in several global biotechnology companies spanning commercial operations, marketing, business and market development, in addition to stakeholder engagement. Maria's experience and passion in working closely with clinicians and scientists to deliver personalised healthcare solutions in the molecular diagnostics market has positioned her to lead the Business Development teams at AGRF, with a special focus on clinical genomics.

MATTHEW TINNING

Head of Laboratory Operations

Matthew has more than 10 years' experience in the management of genomics technologies and services. Matthew has worked in immunology research at Westmead Hospital and as an application specialist with Roche Diagnostics. He joined AGRF in 2009 to manage delivery of NGS services, overseeing significant growth in sample throughput, and frequent NGS technology updates before being appointed Head of Laboratory Operations in 2020.
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CATH MOORE, PHD

Chief Scientific Officer

Cath holds a PhD in molecular genetics and is passionate about how genomic technologies can be used to advance the goals of researchers, clinicians and industry. Cath joined AGRF in February 2020 from QIAGEN where she led the ANZ genomics market development team tasked with engaging the research community to understand emerging needs, provide technical expertise and drive new partnerships.

KAREN JENKINS

Head of People & Culture

Karen spent her early career in Learning and Development roles before moving into the People & Culture sphere. With over 15 years of experience in Human Resources and People & Culture management roles in government, health and community organisations, Karen came to AGRF in 2017. Karen focuses on developing and implementing best practice people strategies and initiatives that enhance the attraction, retention, accountability, reward, recognition and development of our people as well as the review and development of HR systems.

OUR VISION

To help make the world a better and more sustainable place through the use of genomics.

OUR MISSION

AGRF's purpose is to partner with the genomics community to make profound improvements to people's lives by delivering a world-class, innovative, and integrated capability.

OUR VISION, MISSION & VALUES

INNOVATION

We embrace challenges with curiosity, novel approaches and world class technologies.

INTEGRITY

We always act in the best interests of our stakeholders and AGRF as a whole.

RESPECT

We honour individuality and appreciate our working relationships.

EXCELLENCE

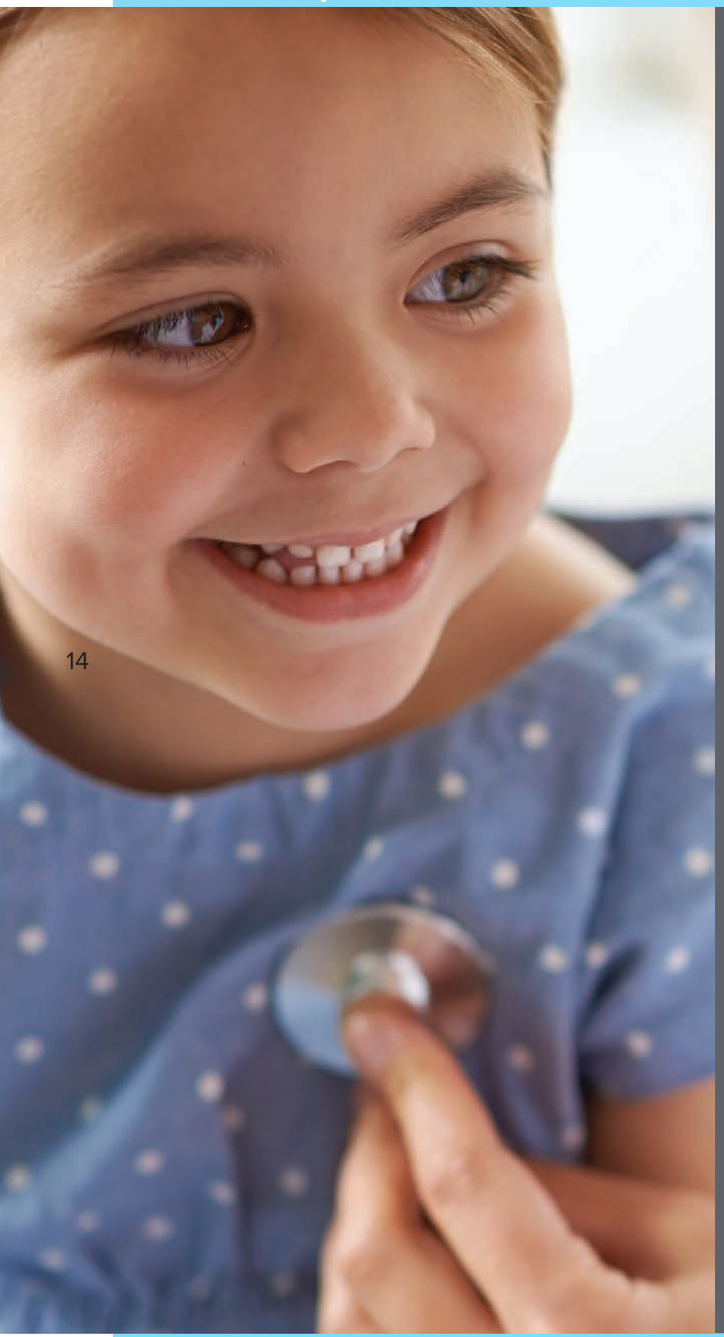
We are proud of our relentless quest to be outstanding in everything we do.

CLIENT-FOCUSED

We are committed to delivering to our client's expectations, all of the time.

COLLABORATION & TEAMWORK

We build inclusive and valued relationships in pursuit of shared goals.



TRANSFORMING CHILDHOOD CANCER

AGRF has become the preferred genomics partner for the Zero Childhood Cancer Precision Medicine Program at the Children's Cancer Institute (ZERO). A joint initiative of Children's Cancer Institute and Kids Cancer Centre, Sydney Children's Hospital, Randwick, ZERO is the most comprehensive and ambitious children's cancer research program ever undertaken in Australia and demonstrates unprecedented collaboration and utilisation of precision medicine.

In a huge step forward, ZERO will be expanding its translational comprehensive genomic testing platform to include all children and young adults in Australia with cancer by 2023, irrespective of cancer type, risk and stage.

A/Prof Vanessa Tyrrell, Program Leader of ZERO at Children's Cancer Institute said, "Today, we have over 700 children and young people enrolled on ZERO. As we prepare to expand ZERO to one day give every child in Australia diagnosed with cancer the very best chance of survival, we're excited to partner with AGRF to make this happen. Together, we will strive for a future where all children have the best chance of living their best lives."

From 2017 - 2021, ZERO was available across the full range of high-risk paediatric cancer subtypes. This integrated multi'omic pipeline has identified the molecular basis for the cancer in almost all cases (94%), with potentially actionable treatment recommendations being made for 72% of patients.[1] Over 700 children have been enrolled on the national clinical trial to date, and ZERO is expanding the program to be available to all Australian children and young people diagnosed with cancer by the end of 2023. ZERO's aim in this next phase is to expand this multi'omic program by an order of magnitude, to carry out whole genome sequencing, whole transcriptome sequencing and array-based methylation profiling of up to 1,000 children with cancer every year by the end of 2023. AGRF will deliver rapid, high-quality, multi-omics sequencing data to support ZERO and its partner network in the growing number of sequencing-centric research projects.

With AGRF taking on the role of genomics partner for this world leading program, this project will become transformational for us, cementing our place as a premier genomics facility within Australia and allowing us to build our capability and capacity out around this project.

AGRF CEO Dr Kirby Siemering said, "at AGRF, we are extremely proud to contribute our technology platforms and expertise in genomics to this incredible project. Working together with the talented team at Children's Cancer Institute and ZERO partners, we aim to make a meaningful impact on the lives of children living with cancer".

Dr. Siemering also pointed out that AGRF is ideally placed for these types of large-scale projects - and that taking on this one will position us to do more of these sorts of comprehensive projects in the future.

[1]Wong, et.al., Nature Medicine, 2020

"As we prepare to expand ZERO ... we're excited to partner with AGRF to make this happen."

DR MARTIN BREED

BUILDING HEALTHIER COMMUNITIES

Dr Breed is a Lecturer in Biology (Ecology/Organismal Biology) at Flinders University and he's been researching the impact that cities have on humans and the environment since 2015. He explains, "As cities expand, urban biodiversity decreases, and this has been shown to increase immune disorders and stress in people living in cities." After publishing a number of papers on how to restore microbial diversity in non-urban areas Dr Breed turned his focus to urban areas.

He explains, "we wanted to utilise genomic techniques to analyse the diversity of fungi in urban green spaces and determine how we can restore the microbial diversity. We know these diverse fungal communities are good for our health, so by restoring the diversity of fungi in urban green spaces we can restore our microbiomes – and improve our health." Dr Breed partnered with AGRF on this project and his results were published in *Revegetation of urban green space rewilds soil microbiotas with implications for human health and urban design*, Restoration Ecology, 12th April 2020.

After looking at the microbial diversity of green spaces, Dr Breed discovered that you can restore areas using simple approaches of replanting a native plant community which influences the soil biota, in this case the fungi. He believes the simple act of going out into biodiverse green spaces can significantly improve our mental health and our immune system, along with improving community wellbeing, which is really important right now.

Dr Breed says, "we used AGRF's Microbial Profiling Service bundled with Bioinformatics which allowed us to look at the environmental DNA in soil samples, collected across over 40 metropolitan green spaces, and interrogate those fungal communities using genomic methods. The aim was to determine whether the restoration intervention was creating a more biodiverse fungal community – and we found that it was, but there were significant differences between the fungal communities in areas like ovals than restored and more biodiverse areas. This is a problem because if we're living in cities where we have a disconnect with nature, we want to know how we can maximise our health.

"This is particularly important during COVID," Dr Breed says, "because viruses challenge our immune systems, so we need to make sure people have well-tuned immune systems and mental health. Getting out into nature - not just any nature, but biodiverse nature - is the most important thing you can do."

Dr Breed has worked with AGRF for many years and is currently partnering with us on a few projects. He explains, "as someone who is not a trained molecular biologist, I understand the concepts and building blocks but I don't run a sequencing lab with AGRF's level of molecular biology. Forming long-lasting partnerships with AGRF has been extremely fruitful because it makes this big problem of molecular biology and sequencing go away. One of the biggest benefits for me is being able to ring people during work hours that are similar to mine, and it's easy to have conversations with your bioinformaticians, for example, to work on different data analysis approaches so the data comes back to me in a state where I can work with it. In addition to that, the service is second to none from my experience – I haven't had any reasons to go elsewhere."

"AGRF's service is second to none."

"SMRTER" CONSERVATION DECISIONS

DR CAROLYN HOGG

Dr Carolyn Hogg, Senior Research Manager for the Australian Wildlife Genomics Group (University of Sydney) has been working on the conservation of threatened species for over 25 years. For the past 10 years she has worked as a geneticist, and has spent the past four years utilising genomics to help conservation managers make smarter decisions about threatened species. Recently she sent some very special threatened species samples to AGRF and they became the first samples to be sequenced via our new AGRF-UQ PacBio Service, on the Sequel II, using four SMRT (Single Molecule, Real-Time) cells.

After working with endangered species, including the Koala and the Tasmanian Devil, Dr Hogg turned her focus to the Woylie and the Bilby, sequencing their genomes as part of a large project in collaboration with Bioplatforms Australia - the Threatened Species Initiative. The Bilby and the Woylie were chosen because they are both managed species – Bilbies are listed as vulnerable in Australia and their population has decreased by around 80%, while Woylies (Burrowing Betongs that live in Western Australia) had a significant population crash (95%) in the early 2000's.

Dr Hogg explains the importance of the reference genomes in helping conservation managers make timely decisions for threatened species. "Having a reference genome lets us call our variants against the genome and it allows us to differentiate between populations that have been through a number of genetic bottlenecks historically and have very low genetic diversity. The more genomic data we have, the greater the resolution, which allows us to make smarter conservation decisions," she explains.

“The reference genomes are crucial - without them, it would be like giving conservation managers a thousand pieces of a five-thousand-piece jigsaw puzzle. However, if I gave them the puzzle box lid it would make it much easier to know what each piece is and where it belongs – and that’s really what the reference genomes are - they are the puzzle box lids.”

Dr Hogg tells us the easiest part of the process was the sequencing and assembly, but getting the samples was much more complex. She explains, “the Bilby reference animal was an old female that was euthanised for health and welfare reasons at Perth Zoo, but the Woylie genome is from a roadkill animal found on the side of the road by a PhD student.”

Once Dr Hogg sent her samples to AGRF however, she was thrilled with how quick the sequencing and assembly was, saying, "AGRF sent the data on Friday night and we received it and ran some tests late on Friday, then sent it for assembly on Monday night thinking it would take a few days, but on Tuesday morning it was complete.” Not only was the service quick, but the quality of the genome is high, as is the quality of the data provided. “Interestingly, the Woylie genome is a better-quality genome than the Bilby even though the sample is a roadkill sample, so it's impressive that we managed to get a genome of such high quality from something that was dead on the side of the road," she says.

When asked why she chose AGRF as a sequencing partner, Dr Hogg said, "as a researcher I'm interested in the technology that will be the most efficient and help us achieve our goal and AGRF was the first organisation to provide a Sequel II service in Australia. Working with AGRF means I don’t have to send my samples overseas for sequencing which makes the process so much easier, particularly when the animals are CITES (Convention on International Trade in Endangered Species) listed or critically endangered. There’s no need to worry about issues with permits, or whether something is going to happen to your DNA in transit – and being able to communicate with somebody in the same country makes it so much easier than communicating with someone overseas."

"I'm interested in the technology that will be the most efficient and help us achieve our goals"

WORKING TOWARDS COVID-19 VACCINES

AP KEITH CHAPPELL

The Rapid Response Vaccine Team based at the University of Queensland are always trying to make a difference in the world and cure disease – and the past two years have seen them face one of the biggest challenges of their careers. Working together to tackle the COVID-19 crisis, the team were attempting to create a critical vaccine that would stop SARS-COVID-19 from crippling countries across the world.

The team, lead by Associate Professor Keith Chappell, Associate Professor Daniel Watterson and Professor Paul Young, received funding from the Coalition for Epidemic Preparedness Innovations (CEPI) in 2019 to establish a rapid response vaccine pipeline. They began the crucial work towards finding an answer for an exponentially spreading virus – racing alongside a number of teams around the world, including ultimate frontrunners Pfizer, Moderna, and the Oxford AstraZeneca program. While the team at UQ were around six months behind the frontrunners, they kept working regardless – finding the vaccine was an ‘all hands on deck’ effort, and Assoc. Prof. Chappell points out that some wonderful collaborations and teamwork came out of it.

“There’s been a lot of communication, it’s a really great time for science and collaboration...it’s very integrated, what people are doing and how we work together now," he says.

Assoc. Prof. Chappell, Assoc. Prof. Watterson and Prof. Young are the inventors of the molecular clamp platform, a technology that would become essential to their research and trials – the clamp is an effective way for presenting the structure that's on the surface of the virus because it functions as a stabilising motif.

While early results were promising with the vaccine presenting a strong neutralising immune result in phase 1 clinical trials, their efforts ultimately encountered a roadblock: their vaccine candidate was found to generate false positive HIV results in some tests. Trial participants who were given the vaccine and not a placebo generated a low level immune response to HIV because the molecular clamp that held the viral protein together was derived from a fragment of one HIV protein called Gp41. While there was no risk of participants contracting HIV, it meant that when the fragment was recognised by the body’s immune system, it generated some antibodies – the same antibodies that are used in screening for HIV. No good workaround could be found and as a result, UQ’s initial trial had to be terminated 11 months into their work.

But that wasn’t the end of the story. Assoc. Professor Chappell fills us in on what has changed since then and what they’ve been working on: the team have reset, and are back on track towards developing what might be a newly-necessary vaccine for new strains of the virus.

“We went back to the drawing board, trying to solve this issue, and over the last year have been reworking the technology to find a solution – so that we can have molecular clamp 2.0 that works just as well, but doesn’t have the issue, and doesn’t use any HIV sequences. So, that is looking really positive, and it’s just in time for this Omicron [variant] that looks like it does need a whole new vaccine. I guess it’s round two, and the race for a vaccine starts all over again.”

While round one may not have made it to fruition, it demonstrated an important proof of principle finding for the molecular clamp technology – and, with funders that remained supportive, the team have been able to completely rework their technology to create a new system.

“In science, things don’t always go right,” were Assoc. Prof. Chappell’s words at the time, and they resonate at this stage, two years into the pandemic. And while things may have taken a detour for his team, others got there in the end when our world most needed it. “The fact that science as a whole has achieved that [a vaccine] is such a triumph for people in my field, and all the other vaccine developers around the world.”

Assoc. Professor Chappell has worked very closely with AGRF over the years and these last two years have been no exception, as he says AGRF were instrumental in the early stages of the project. He explains, "AGRF played a huge role in our work at the very early stages – at the very start of this outbreak we began working on this the day the sequences were released from China, way back in January 2020.”

And it’s an ongoing relationship, he says. “We use AGRF every week. Our sequencing for the new clamp development has been done through AGRF. When we make a new vaccine, everything is confirmed through AGRF. The turnaround time is really important to us, it’s great that we can get sequences back quickly and move quicker into producing the vaccine.”

"without AGRF providing results in a day or two ... we'd still be scratching our heads"

MELINDA ZIINO
GENOTYPING SECTION MANAGER

Melinda Ziino began working at AGRF as an Honours Student with The University of Melbourne in 1999. After she completed her studies the following year she joined AGRF, and has become an integral member of the team. Naturally her role has changed considerably over the last 20 years. She began as a Genotyping Analyst and now manages the whole Genotyping Section.

Working with Illumina array technologies is a major part of her job, and she explains how these technologies benefit our clients: "from small, to large-scale projects, our AGRF Illumina array service can fit our clients' needs. With arrays of varying densities, from humans, to plants and animals, there is usually an array available to suit our clients' research goals."

Melinda informs us that Illumina arrays, and the genomic information they target can be beneficial to a vast number of applications – from disease association, to clinical and precision medicine, to pharmacology and direct-to-consumer applications such as nutrition and agriculture.

At AGRF, Illumina arrays are widely used in genome-wide association studies (GWAS) for identifying disease associations across the whole genome – but the list of applications goes on.

As her role changed over the years, so too did the technologies and methods used. Melinda explains, "when I first started at AGRF over 20 years ago, all GWAS were performed using microsatellites. The process was time-consuming and manual. As array technology emerged, with the discovery of SNPs, AGRF was at the forefront of adopters, and our early array service was using the Affymetrix array system. When Illumina launched the Infinium array, AGRF transitioned to this technology and Illumina arrays continue to be the leading technology in this field."

With AGRF's clinical offering continuing to expand, Melinda says the primary clinical application AGRF is involved with is the use of arrays for cytogenetics. "Arrays are now the first recommended test in paediatrics, for determining if there are genetic causes for intellectual disability, developmental delay, and behavioural conditions such as autism spectrum disorders," Melinda explains.

So what is it about AGRF that has kept Melinda staying for over two decades? She points to two reasons: "the staff, which are like a second family, and the variety of projects. Each day talking to researchers about their different research goals and the best technology fit, means every work week is different".

MELINDA ZIINO

Melinda is our Genotyping Section Manager and has been with AGRF for more than 20 years.

DR DAVID MARTINO

UNDERSTANDING NEWBORN IMMUNITY

Neonatal death due to infections and sepsis affects millions of newborns each year, mostly in the developing world – and the majority of those babies die within the first few weeks of life. The Human Immunology Project Consortium (HIPC) seeks to target this, aiming to understand newborn immunity and how vaccines can be optimised to prevent neonatal death. Undertaking research that applies systems biology and vaccinology, HIPC is working with vaccines to map out the pathways from vaccination to immune protection.

When Telethon Kids Institute were asked to be the epigenetics lead for the project, the Institute jumped on board – with AGRF joining as their service partner.

Dr David Martino, Team Leader of Clinical Epigenetics at Telethon Kids, speaks on what this project and its results could mean for newborn mortality.

“Vaccines are really the most effective and safe way to prevent this burden of neonatal mortality. However, the current vaccine designs are ill-suited to newborns, because they have a very unique immune system. We really don't quite understand the newborn immune system in the same way that we do the immune system of young children, where most vaccines are targeted to work. What this project is about is trying to understand the best strategies to protect newborns against infection and sepsis.”

To build detailed maps of the biological pathways leading from vaccination to immune protection, the study focused on two vaccines in particular: the BCG vaccine (used to protect against tuberculosis), and the Hepatitis B vaccine, with the BCG in particular demonstrating some interesting attributes.

“The BCG vaccine protects against tuberculosis but also has this wonderful ability to protect against other pathogens in what we call a ‘pathogen agnostic’ way,” explains Dr Martino. “So, it seems to provide protection against all cause-mortality, and against respiratory infection and hospitalisation later on in life – and the way it does this is through the innate immune system. This vaccine seems to be really good at boosting innate immunity through epigenetic changes.”

Dr Martino points out that although the initial results are promising, the data at this stage is only in pilot analysis form – while promising, it’s merely at first-pass stage, and there’s much more work to be done.

“We’ll be looking at what the various ‘omics signatures are between Hep B as a more conventional vaccine, versus BCG as a novel innate immune adjuvant vaccine,” he explains.

And, in a very relevant divergence from the neonatal focus, studying the use of the BCG vaccine and its efficacy against respiratory viruses has led to it now being trialled for potential defence against COVID-19. “This could potentially be another tool in the arsenal that could be deployed rapidly in pandemic situations,” explains Dr Martino.

He also talks about how AGRF worked with the Telethon Kids Institute, to help forge the way through this large-scale project.

“We worked with AGRF to be able to offer the methylation array service and the human global screening array service, at a really competitive rate that was able to compete internationally. Over the second half of 2020 and into 2021, we processed somewhere in the order of 2,000 samples for the consortium,” says Dr Martino.

And it was a smooth and satisfying partnership – with Dr Martino pointing out that the locality of AGRF combined with our level of service made the decision to come to us an easy one. “We have a great relationship with AGRF that probably goes back a good 5 years at least. It helps a lot having someone local being a service provider and that's what we were really looking for - a local partnership that would be able to communicate effectively and turn around work rapidly, and that's what I think really differentiated AGRF.”

Being able to support local business as well as build on existing relationships was a priority for Telethon Kids in this project, Dr Martino says. “We wanted to support local business as well, and AGRF did a fantastic job at working with us to be able to offer the service at the level and price point that we needed to be able to bring it to the consortium.”

"AGRF did a fantastic job at working with us to be able to offer the service at the level and price point that we needed."

INFORMED CLINICAL DECISIONS

MR EMANOUIL SIGALAS

Emanouil Sigalas is the Molecular Pathology Senior Medical Scientist at The Royal Melbourne Hospital where whole exome sequencing and variant calling is used to report on complex diseases. Mr Sigalas was drawn to clinical pathology because he was fascinated by the concept of an ‘immortal cell’ as seen in various cancers, and he was especially drawn to using such cell lines as a means for investigating disease by inserting engineered pieces of cDNA into these cells and characterising the structure and function of the protein produced.

Mr Sigalas recalls, “I eventually became interested in multi-gene pathways leading to disease, then an opportunity arose to develop diagnostic assays to identify genetic variants that predispose individuals to Hereditary Breast and Ovarian Cancer (HBOC), Li Fraumeni Syndrome, Multiple Endocrine Neoplasias as well as other non-cancer related genetic diseases. I particularly enjoyed this experience because I was able to apply my experience from structure versus function experiments to curate genetic variants for diagnostic clinical use. It has been a privilege to work as part of a collaborative effort with local and international research and diagnostic laboratories, and service providers," he says.

Whole exome sequencing and variant calling play a vital role in Molecular Pathology's clinical studies, as Mr Sigalas explains "our collaboration with AGRF allows us to access high quality next generation sequencing data and a best practice bioinformatics pipeline that is NATA accredited against the international standard ISO15189 for medical testing. This feature is important in our diagnostic setting because consistently high-quality data is needed for clinical reporting of complex diseases. This reduces the need to investigate further potential artefacts or variants of poor quality.

"Our workflow and resources immensely benefit from reliable variant calling files that are ingested by our variant curation platform to provide a streamlined process for variant prioritisation, curation and reporting. This enables us to focus on triaging cases for curation and delivering diagnostic reports within a clinically relevant timeline. The whole exome approach provides an opportunity to investigate a panel of genes based on a given phenotype, and then have the same data re-analysed when new clinical information is available. Overall, the burden on the patient is reduced and the level of care is greatly enhanced," Mr Sigalas says.

In terms of the outcomes they are able to achieve with these technologies, Mr Sigalas is confident that "the services provided by AGRF are helping to transform the delivery of healthcare at The Royal Melbourne Hospital. We achieve this goal by rapidly reporting clinically important genetic information, relevant to an individual’s care, to multidisciplinary clinical teams as well as the Genetic & Genomic Medicine team. Often this data affects other family members or assists in answering clinical questions depending on the family history and personal presentation, and helps patients understand the clinical advice received in relation to their disease, its progression, and therapeutic options. It’s worth noting, a collaborative effort often involving other institutions is required to distil accurate information. We’re hoping over time, robust data sharing systems will aid further development of local and government policy to enable genomics into mainstream disciplines with improved funding models."

"The strength of our working relationship proved to be effective when technical issues required clear communication and a like-minded approach to patient care."

INFORMED CLINICAL DECISIONS

"Another significant goal is the use of genomic tools to unlock aspects of personalised medicine by providing precise evidence for early diagnosis, improved chances of successful treatment, intervention and prevention by detecting disease before symptoms (often serious) present in patients. For example, Malignant Hyperthermia is a rare life-threatening condition usually triggered by exposure to volatile anaesthetic drugs used in general anaesthesia. These patients and their family members routinely benefit from our combined multidisciplinary approach by better preparing for surgery, and the family members shown to harbour the disease-causing genetic variant are also counselled to wear warning bracelets in the event of unforeseen emergencies requiring urgent surgical treatment," he adds.

Mr Sigalas stresses the importance of genomics in helping to achieve outcomes for patients, saying, "genomics has driven and accelerated fast, low-cost, large-scale generation of sequencing data that allow scientists and clinicians to identify variants that influence disease, drug response or overall health of a patient. Rare diseases that would have taken years to diagnose using conventional methods are now being investigated with precision by tailoring a gene-specific panel for every individual, depending on the clinical presentation, personal and family history. However, commercially available chemistries are subject to limitations so it's imperative the correct product and platform is selected to interrogate a given condition.

"In our adult setting, we use whole exome sequencing as a screening tool, rather than a complete test, to analyse a patient's DNA sample because certain regions cannot be assessed adequately. For example, we cannot detect repeat expansions, copy number variants or chromosomal rearrangements. Additionally, variants in regions of low coverage may not be detected, and there's limited sensitivity for insertion/deletion variants, as well as reduced sensitivity and specificity of variant detection in genes with pseudogenes, paralog genes and genes with regions of low complexity.

"For these reasons, complementary or supplementary testing is required for effective clinical management. Therefore, advances in sequencing technology, whole exome and whole genome chemistries, bioinformatic platforms, and most importantly, development of diagnostic-grade curated variant databases are vital in advancing clinical genomics. Significant effort in education and training in variant curation will be instrumental to improving the bottleneck of variant prioritisation, classification of variants and speed of reporting. Out of all the 'omics', I am keen to see genomics propel pharmacogenomics even closer to precise personalised medicine by identifying variants in multiple genes that are known to be associated with variable drug responses. This way an effective targeted therapy can be delivered based on an individual's genomic profile," Mr Sigalas explains.

With the choice of different service providers, Mr Sigalas cites the affordable cost of Sanger sequencing as the reason Molecular Pathology turned to AGRF for assistance with high throughput services. He elaborates, "we found our collaboration at that time provided a reliable and timely product to our clients. The strength of our working relationship proved to be effective when technical issues required clear communication and a like-minded approach to patient care. This mindset led to the beginning of a 'centre of excellence' attitude to clinical diagnostic genetic testing.

"Naturally, AGRF was the service of choice to help us meet our NATA accredited clinical whole exome screening strategy at The Royal Melbourne Hospital. The next generation sequencing data provided for our analysis is consistently of very high quality and always delivered within our expected turnaround time. Communication remains effective with swift resolution to problems, which is critical to our success. We feel safe in AGRF's support, meaning we can plan and advise the clinical team when diagnostic reports will be available for effective patient clinical management," Mr Sigalas concludes.

AGRF-UQ PACBIO SERVICE

In 2020, in collaboration with Bioplatforms Australia, AGRF and The University of Queensland (UQ) collaborated to become a joint Certified Service Provider (CSP), providing Long Read Sequencing using the PacBio Sequel II. The AGRF-UQ PacBio Service expands both AGRF's and UQ's capabilities in long-read PacBio sequencing for generating high-quality genome assemblies, comprehensive transcriptome analyses, and epigenetic information.

As a Certified Service Provider for Single Molecule, Real-Time (SMRT) Sequencing, this collaboration offers the Sequel II System for affordable, high-throughput studies of microbes, plants and animals, or humans.

One notable project utilising the AGRF-UQ PacBio Service was sequencing of four Māori genomes for the Aotearoa New Zealand Variome project, part of Genomics Aotearoa. The aim of this project is to build a catalogue of the unique variation of the New Zealand population in order to better understand it and its health consequences, and to ensure accurate diagnoses and the effective use of genomics in healthcare for the Aotearoa's unique and increasingly diverse population.

In late 2020, we partnered with PacBio to run an Australia / New Zealand SMRT Grant Competition, which was won by Professor Glenn King's group at the Institute of Molecular Bioscience at the University of Queensland, with his project '*Deadly Genes: Exploring the Genome of the lethal Australian Box Jellyfish*,' and he plans to use HiFi sequencing in an integrated multi-omics investigation into venom of the Australian box jellyfish *Chironex fleckeri*.

The aim of the project is to integrate genome, transcriptome and proteome data to systematically investigate the function, evolution, and ecology of *C. fleckeri* venom. Prof. King says "gene duplication and alternatively spliced transcripts are known to create protein diversity in venom of animals, therefore high-quality long-read sequence data are critical for resolving repetitive elements in the *C. fleckeri* genome, whereas deep sequencing of full-length transcripts enables comprehensive recovery of alternative splice-forms that explain protein diversity. Thus, PacBio HiFi combined with Sequel II system is the perfect, cutting-edge solution for us".

Since launching the AGRF-UQ PacBio service in May 2020, we have:

- processed 35 SMRT cells
- sequenced 14 new genomes
- completed 920 full length 16S microbial diversity profiles
- completed 11 reference genome annotation projects

INNOVATION & DEVELOPMENT

LONG READS

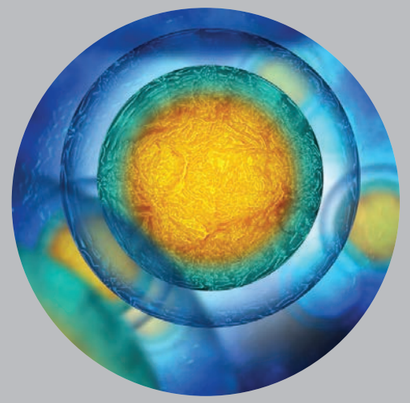
As an early adopter of new technologies, AGRF was the first organisation in Australia to implement both the Oxford Nanopore Technologies PromethION and the PacBio Sequel II long read sequencing platforms.

After being the first supplier to purchase and install a PacBio Sequel II in Australia, our workflows have now been fully implemented into our service offerings, providing the Australian research community, and beyond, with the tools required for generating high-quality genome assemblies, comprehensive transcriptome analyses, epigenetic information and characterisation of large structural variants.

PRAWN GENOME

Working in partnership with the ARC Industrial Transformation Research Hub (ITRH) for Advanced Prawn Breeding, our scientists have applied state-of-the-art long read sequencing to enhance our understanding of the Black Tiger Prawn transcriptome.

This work involved a close collaboration between ITRH partners and AGRF Innovation & Development scientists and bioinformaticians, and culminated in a publication in Nature's Scientific Reports. This work will enable the Australian prawn industry to benefit from advanced genomic-informed breeding programs.

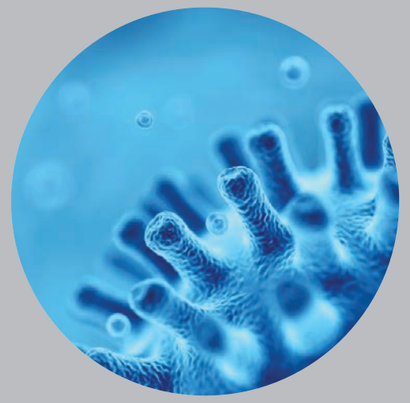


SINGLE CELL

Developing new technologies to meet emerging needs in the research community is a key focus of AGRF's Innovation & Development Team.

Single cell genomics is a major growth area in medical research enabling insight at the cellular level.

Through collaborations with researchers throughout the Parkville biomedical research precinct, we have co-authored a publication in this area: *Lukowski SW, Lo CY, Sharov AA, et al. A single-cell transcriptome atlas of the adult human retina. EMBO J. 2019 38(18):e100811.*



COVID-19

The outbreak of the global COVID-19 pandemic in 2020 presented massive challenges to scientific and medical communities alike.

AGRF has joined the effort to overcome some of those challenges. We immediately started working towards establishing an ARTIC v3 protocol and applied for NATA accreditation to sequence COVID patient samples, and we were able to quickly establish workflows for diagnostic testing, waste water testing, viral genome sequencing and confirmation sequencing for vaccine development.

EVENTS

JUL 2019

- **UQ Winter School in Computational Mathematics & Computational Biology (QLD).** We provided Travel Sponsorships for $500 and Dr Chris Noune, our Microbial Profiling & Custom Amplicon NGS Laboratory Supervisor, gave a presentation on our Microbiome services.
- **Oz Single Cell Meeting (VIC).** AGRF was a Silver Trade Sponsor and our Senior Scientist Dr Jafar Jabbari presented on our Single Cell work.

AUG 2019

- **HGSA Annual Scientific Meeting (New Zealand).** Our New Zealand Account Manager Dr Glenn D'Cunha had a trade booth at this international conference for the Human Genetics Society of Australasia, and our Operations Team members Melanie O'Keefe and Matthew Tinning also attended the event.
- **Translational Research Symposium (QLD).** We were a Bronze Sponsor of this symposium and our QLD Account Manager Saurabh Shrivastava was on our AGRF trade booth.
- **QUT IHBI Inspires Conference (QLD).** AGRF had a trade display at this event run by the Institute of Health and Biomedical Innovation (IHBI) at the Queensland University of Technology (QUT). Our QLD Account Manager Saurabh Shrivastava attended this state event.
- **Bio21 Institute Trade Show (VIC).** We had a trade table at this event and our VIC Account Managers Dr Glenn D'Cunha and Anna Ikonomopoulos manned the table on the day.
- **West Coast Microbiome Network Global Microbiome Meeting (WA).** We had a trade display at this meeting and our WA Account Manager, Dr David Chandler, attended the event.

SEP 2019

- **Sanger September (AUS).** We held 15 events across Brisbane, Melbourne, Perth, Sydney and Adelaide, where clients were able to meet AGRF staff and tour our laboratories.
- **AGRF NGS & Bioinformatics Workshop (VIC).** We ran a workshop and morning tea at Monash University.
- **VCCC Conference (VIC).** Our National NGS Manager Matthew Tinning presented a talk.

OCT 2019

- **Australasian Genomic Technologies Association Conference (VIC).** We had a trade exhibition at this conference, manned by our VIC Account Manager Dr Glenn D'Cunha.
- **AGRF Spring Into Agrigenomics Seminar (SA).** We ran a seminar on agrigenomics featuring talks from Dr Kristy Sumby (ARC Wine Centre), Dr Adam Croxford (University of Adelaide's School of Agriculture, Food & Wine), Dr Korjent van Dijk and Dr Iain Searle (both of University of Adelaide's School of Biological Sciences).
- **Monash ECR Symposium (VIC).** VIC Account Manager, Anna Ikonomopoulos, presented a talk at this event.

NOV 2019

- **International Tropical Agriculture Conference (QLD).** We had an exhibition booth at this conference and our QLD Account Manager Saurabh Shrivastava and National Research Partnerships Manager Dr John Lai attended the event.
- **Epigenetics Consortium of South Australia Meeting (SA).** We sponsored the People's Choice Award and SA Account Manager Dr Leanne McGrath presented the award.
- **GeneMappers Conference (NSW).** Our NSW Account Manager Dr Oliver Distler attended this event as a delegate.
- **Melbourne HIV Exchange - World Aids Day (VIC).** We sponsored this event and our VIC Account Manager Dr Glenn D'Cunha attended.

DEC 2019

- **AGRF Introduction to Cloud Computing Workshop (VIC & NT).** Our Bioinformatician Naga Kasinadhuni presented bioinformatics workshops on Cloud Computing in Melbourne and Darwin.

JAN 2020

- **AGRF Summer Oncology Seminar (VIC).** We ran a seminar on genotyping which featured talks from AGRF CEO Dr Kirby Siemering, Genotyping Manager Melinda Ziino, Dr Tunguyen (Monash University), Dr Christine White (Hudson Institute), Dr Elizabeth Christie (Peter Mac) and Dr Edward Chew (WEHI).

FEB 2020

- **Lorne Genome Conference (VIC).** We had an exhibition booth at this international conference and AGRF posters were presented by our Bioinformaticians Naga Kasinadhuni and Alexis Lucattini; Lab Technician Justin Crockett, National NGS Manager Dr Chris Noune, Bioinformatics Manager Dr Kenneth Chan and Senior Scientist Dr Jafar Jabbari.
- **CSIRO Cutting-Edge Symposium (VIC).** Our VIC Account Managers Dr Glenn D'Cunha and Anna Ikonomopoulos attended this symposium at CSIRO.

APR 2020

- **Human Genome Meeting (WA).** Our WA Account Manager Dr David Chandler attended this event.

MAY 2020

- **Bio21 Trade Show (VIC).** We were a trade sponsor of this show and our VIC Account Managers Dr Glenn D'Cunha and Anna Ikonomopoulos were at our trade table on the day.

EVENTS

JUN 2020

- **International Conference of Quantitative Genetics (Online)**. We sponsored the ISQG6 Conference, which was changed to a virtual event due to COVID.

AUG 2020

- **PacBio virtual webinar (Online).** We ran a successful 'PacBio HiFi Sequencing: A New Era Of Genomic Discovery, hosted and moderated by BRIS Node/SNP Platform Manager Dr David Hawkes - the recording also went up on our YouTube channel post-event.

SEP 2020

- **OzSingleCell 2020 Episode (Online).** We sponsored the Oz Single Cell episode 'Oz Single Cell - Cell type, cell state, or cell artefact? Finding the biology in your single cell data' which went ahead online, as part of their series of virtual seminars.
- **Virtual Nanopore Day (Online).** Senior Scientist Dr Jafar Jabbari presented a talk at this virtual day that was themed "Cancer research with nanopore sequencing".

OCT 2020

- **Single Cell Webinar (Online).** We held a webinar on Single Cell RNA Sequencing - both our Senior Scientist Dr Jafar Jabbari and our NT/QLD Account Manager Saurabh Shrivastava presented talks.
- **TQEH Research Expo (SA).** Our SA Account Manager Dr Leanne McGrath attended The Queen Elizabeth Hospital (TQEH) Research Expo in person.
- **GENE Awards (BRIS).** QLD Account Manager Saurabh Shrivastava attended the Globally Engaging Networking Event (GENE) Awards luncheon.

NOV 2020

- **ASMR WA Symposium (Online).** AGRF supported the Australian Society for Medical Research (ASMR) as a trade sponsor at their Symposium, which ran as part of the ASMR Medical Research Week. Originally planned as an in-person event, it was changed to a virtual event due to COVID.
- **ICQG6 Conference (Online).** We sponsored the virtual International Conference in Quantitative Genetics (ICQG6) - QLD Account Manager Saurabh Shrivastava and Head of Science and Tech Dr Cath Moore attended + ran our virtual booth.
- **SCMB Symposium (Online).** AGRF sponsored the University of Queensland's School of Chemistry and Molecular Biosciences (SCMB) Research Symposium, an online event this year due to COVID.
- **HGSA Virtual Conference (Online)** We had a virtual exhibition booth at the Human Genetics Society of Australasia (GGSA) Virtual Conference. VIC + NSW Account Managers Dr Glenn D'Cunha and Dr Oliver Distler, Quality Manager Melanie O'Keefe and Head of Lab Operations Matthew Tinning dropped in to chat to attendees.

DEC 2020

- **Complex Diseases Webinars (Online).** We ran the first of our free Complex Diseases Seminars, with three instalments featuring speakers Prof Stephen Harrap (University of Melbourne), Prof Simon Laws (Edith Cowan University), and Dr Cristina Fortuno (Fitgenes).

JAN 2021

- **Complex Diseases Webinars (Online).** We ran the final event in our series of free Complex Diseases Seminars. We heard from A. Prof Michael Hildebrand (University of Melbourne), on 'Somatic Mutation: The Hidden Genetics of Epilepsy".

MAR 2021

- **UQ PEF Molecular Engineering Workshop presentation (online).** QLD Lab Tech Kimberley Rogl gave a presentation on Sanger Sequencing + Troubleshooting at the University of Queensland's PEF Molecular Engineering Workshop.
- **Future Labs Conference (Online).** Head of Lab Operations Matthew Tinning gave a presentation at the virtual Future Labs Conference, on "Streamlining Large-Scale Data and Cutting-Edge Automated Processes within Laboratory Operations".
- **AGRF Lab Tour (MELB).** Our National NGS Manager Dr Chris Noune hosted a virtual site tour of our Melbourne laboratory, filmed and live-streamed by Sales + Marketing Manager Desley Pitcher.
- **AGRF On Demand - Carolyn Hogg webinar (online).** Dr Carolyn Hogg (University of Sydney) presented a webinar for our first AGRF Live & On Demand event - "Conservation in the Age of Genomics", with our PacBio Service Manager Dr David Hawkes moderating.

MAY 2021

- **In-person lab tour (MEL).** The NCRIS Roadmap Expert Committee meeting took place at VCCC, and the committee members took a tour of our lab. AGRF CEO Dr Kirby Siemering attended the tour.
- **Precision Medicine Conference (NSW).** NSW Account Manager Dr Oliver Distler attended the Westmead Precision Medicine Conference in person, setting up with a trade table.
- **Science on the Swan (WA).** We were a bronze sponsor of Perth's Science on the Swan Conference, with WA Account Manager/Node Head Dr David Chandler attending in person.

JUNE 2021

- **Illumina Webinar (Online).** A joint AGRF x Illumina webinar was presented by our National NGS Manager Dr Chris Noune, titled "Illumina and the Post-TruSeq World".
- **ASMR WA Symposium (Online).** The Australian Society for Medical Research (ASMR) ran their annual Medical Research Week event and we took part in the virtual symposium. Our WA Account Manager/ Node Head Dr David Chandler attended and our NGS Manager Dr Chris Noune pre-recorded a presentation to play.
- **GeneMappers 2021 (BRIS).** The GeneMappers Conference went virtual for this year, but they held a "Local Hub" meeting in Brisbane which we sponsored, and our QLD Account Manager Saurabh Shrivastava attended in person.

POWERING AUTISM RESEARCH

Autism CRC's Australian Autism Biobank was launched in December 2018 and contains Australia's largest collection of biological, behavioural, environmental and medical information of children on the autism spectrum and their families. The large-scale collaborative project recruited almost 2,900 autistic and non-autistic children and adults from four sites across Australia, contributing 4,500 biological samples, as well as behavioural and questionnaire data.

Having samples and detailed information from many study participants can accelerate the pace of research, as recruiting participants and collecting these samples and information takes time and money. A biobank safely stores this data in one place for approved research access. However, biological samples can also degrade and become less usable over time.

In 2020 - 2021, Autism CRC took on an internationally significant project, commissioning the generation of whole genome sequencing (WGS) data for more than 1,500 participants. Sequencing was carried out on DNA samples from 437 families (children on the spectrum, siblings not on the spectrum, and parents) who participated in the Australian Autism Biobank between 2014 and 2018.

The project used the expertise and innovation of Australian-based service providers, working within the Commonwealth-funded National Collaborative Research Infrastructure Strategy (NCRIS). The bioinformatics pipeline utilised the resources of Bioplatforms Australia, with sequencing undertaken by AGRF and bioinformatics being carried out by the Garvan Institute of Medical Research.

Autism CRC CEO, Andrew Davis, says AGRF was chosen for a number of reasons.

"Beyond their technical platform and capability, AGRF's reputation as an effective NCRIS collaborator was a key part in our partnering decision. This proved to be very valuable, particularly given the unforeseen challenges all had to manage."

The entire project took place during the COVID-19 pandemic with the project team - which included representatives from each service provider and Autism CRC project staff - meeting virtually each week.

"We started this large and ambitious project in the initial stages of the global pandemic and, while there was a lot of upheaval during 2020, the AGRF team has worked seamlessly to support the delivery of this project. Despite travel restrictions and lockdowns, AGRF provided us with continuity of service and worked collaboratively and flexibly with us and our other service providers to deliver the project," says Dr Felicity Rose, then Autism CRC Project Coordinator.

The success of the Biobank to date has resulted in enormous interest from the scientific community. This resource is among a small number of autism resources globally which encompass varying levels of biological and phenotypic information.

The addition of the sequencing data will make the dataset an even more valuable and lasting asset for national and international research.

Over 25 projects are already underway, across different research areas relating to autism, including better and earlier

"the experience of working with AGRF ... has been amazing"

POWERING AUTISM RESEARCH

autism diagnosis, sensory subtyping, behavioural characteristics of females on the spectrum, and a range of co-occurring conditions and health issues such as anxiety, gastrointestinal disorders, sleep disorders and oral health.

Through an understanding of how biology and environment interact, and their linkages to autism sub-types, we may help connect individuals and families to more appropriate and effective supports for health and wellbeing.

One of the first studies approved to use the new WGS data will investigate genetics underlying motor difficulties in children on the autism spectrum. Motor impairment is not currently included in the diagnostic criteria or evaluation of autism but there is interest in understanding the emergence of motor impairment and its value as an early prognostic marker.

“Our research team feel very fortunate to have access to Australian Autism Biobank to gain further insight into motor impairment. While most of our work up to this point has concentrated on the phenotypic data through the Australian Autism Biobank, we will soon be using whole genome sequencing to examine autistic children with and without motor impairments and evaluate specific genes of interest,” says lead researcher, Melissa Licari, Telethon Kids Institute.

The Australian Autism Biobank and Autism CRC are committed to creating end-user driven research, and this includes use of the data from WGS. Oversight of the applications to use any Biobank data is by the Autism CRC Access Committee. This committee consists of at least six people appointed by Autism CRC, including at least one member of the autistic or broader autism community. All approved projects must have Human Research Ethics Committee approval, and demonstrate a clearly identified potential benefit to the autistic and autism communities.

Autism CRC has also established the Australian Autism Research Council (AARC) to identify end-user driven autism research priorities in Australia. Initially, the Biobank will look to communicate the outcomes of the AARC to applicants with a view to encouraging applications guided by community priorities.

Autism CRC will be seeking further funding to complete sequencing of other data samples, adding to the depth of information available and opportunity to further increase the value of the Biobank as an international asset.

DR MELONY SELLARS

FARMING BETTER PRAWNS

Dr Melony Sellars is a global shrimp expert and Co-Founder of Genics Pty Ltd, which aims to solve real world problems through developing and applying novel biotech solutions. She has 20 years of experience in aquaculture, with an in-depth background in shrimp aquaculture, and extensive expertise in the application of novel biotech solutions for industry, genetics and breeding programs. Her goal is to revolutionise shrimp farming practices to deliver global food security for the future, and she's taken a huge leap towards achieving that goal thanks to the Genics Shrimp Multipath Panel which enables early detection of pathogens and empowers farmers to make smarter management decisions.

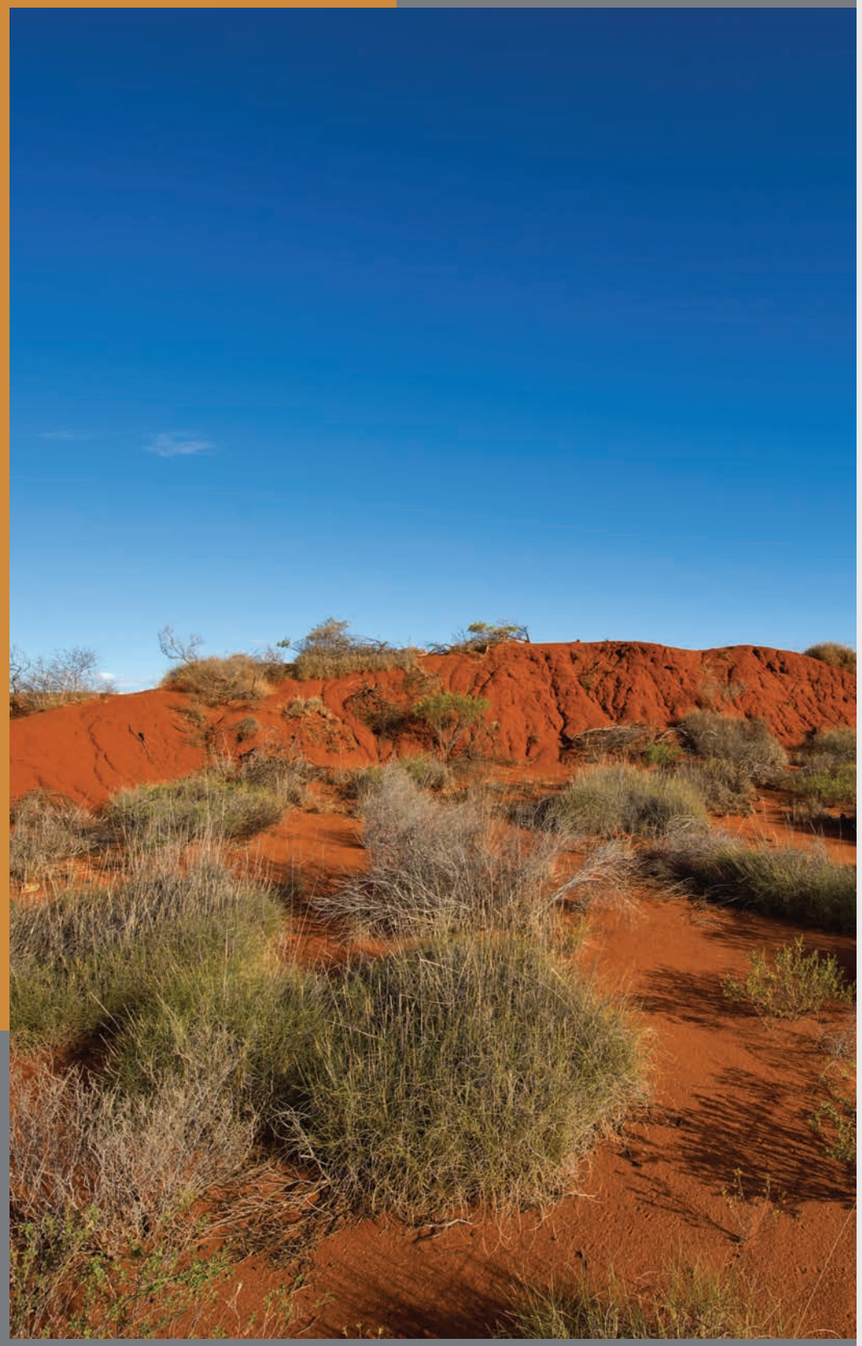
Dr Sellars explains how the PCR-based platform was conceived, "when working closely with the domestic and international prawn industries I could see a clear problem that needed solving: they had multiple pathogens at any one time and no cost-effective accurate mechanism of detecting them early enough to mitigate stock losses. At the laboratory in my role as a CSIRO Research Scientist I uncovered a range of techniques and chemistries that, when combined, formed the foundation of Shrimp MultiPath.

"It's all about early detection and early mitigation before clinical signs are seen. Multipath gives farmers up to three weeks early warning that a problem is arising and puts them in a position to make smart management decisions. We identify them before clinical signs are detected, which means sick prawns can be identified before they reject their feed. Importantly, early detection empowers farmers to implement management choices that, as in the case of one pathogen (IHHNV,) can increase production output by AUD67,000 per hectare simply by screening postlarval or juvenile shrimp on MultiPath," Dr Sellars explains.

The implications for the commercial aquaculture industry are simple: healthier, faster growing shrimp that survive. In Australia, this directly improves the profitability of farms and increases crop consistency. In farms overseas, it reduces the use of chemicals and antibiotics during production.

Dr Sellars explains why she chose AGRF as a sequencing partner for this project, saying, "I chose AGRF because of the people and culture. As a CSIRO scientist, I was privileged to meet AGRF CEO (at the time) Sue Forrest thanks to one of your wonderful Account Managers. Sue was such a visionary for AGRF and together we sparked the beginning of a very large prawn genome project. From there my interactions with the Brisbane Team continued to grow, as did interactions with your new CEO Kirby Siemering. Then when I was ready to start Genics, AGRF was wonderfully supportive at every step of the way."

"I chose AGRF because of the people and the culture."



TRANSFORMING MINING ECONOMIES

We were delighted to announce in March 2020 that AGRF became a partner in the CRC for Transformations in Mining Economies (CRC-TiME), which aims to ensure a sustainable future for our mining towns in rural and regional Australia. The new CRC was designed to help mining communities broaden their local economies, and provide the tools for towns and regions to navigate opportunities to create new jobs and development that keep communities strong.

AGRF CEO Dr Kirby Siemering said, "CRC-TiME has now been formed with AGRF working as part of a Biodiversity working group to develop a Foundation Project which will drive biodiversity and ecogenomics as a future research theme."

AGRF Head of Science and Technology Dr Cath Moore expanded on this saying, "AGRF is a Supporting Partner in the newly-funded and formed 10-year CRC-TiME. The CRC is grappling with the challenges of mine closures in regional communities around Australia – and trying to determine how to successfully transform the land and community from mining to a sustainable post-mining environment.

"AGRF will play a key role in bringing genomic tools to the forefront of these challenges, particularly around biodiversity, environmental sustainability, climate change resilience and soil remediation. This will provide us with the opportunity to develop new technologies which will be relevant to various environmental and agricultural applications, as well as opening the mining sector up as a new market for our genomic services, Dr Moore explained."

The successful outcome was announced by the Hon Karen Andrews MP and Hon. Keith Pitt MP in a joint statement on Friday 13th March, 2020. In the statement, they explained that this new CRC will work to ensure a sustainable future for our mining towns in rural and regional Australia.

The Hon. Karen Andrews MP and Hon Keith Pitt MP stated, “Australia’s world-class mining expertise is undeniable, but being a global resources powerhouse means ensuring a sustainable future for our mining towns in rural and regional Australia.

“The Government and the resources industry recognise more needs to be done to diversify the economy of some local communities which are highly dependent on individual mining projects. The work of CRC-TiME will help mining communities broaden their local economies so they can continue to grow and stay strong.”

CRC-TiME

NAGA KASINADHUNI
SENIOR BIOINFORMATICIAN

Naga joined the Brisbane team in June 2012. Working as a bioinformatician, Naga's role was primarily aimed at developing, maintaining and supporting various bioinformatics pipelines including Illumina, GS-FLX and Ion-Torrent platforms. He was also responsible for designing primers, maintaining the Sanger pipeline to ensure high uptime and reliability, and he also developed and maintained identity databases.

Naga explains the evolution of his role, "in later years I worked on assembly projects, developed diversity profiling pipeline on GS-FLX – and when it became obsolete, I continued development on the Illumina platforms. Currently I'm responsible for metagenomics, genome and transcriptome assembly projects, developing various data processing and analysis pipelines for PacBio, and I supervise diversity profiling, Genotyping and Sanger projects.

When asked why microbial diversity profiling is so important Naga replied, "for most of the earth's 4.5 billion-year history, microbes were the only living organisms in existence. Microbes evolved on Earth approximately 3.5 billion years ago and eventually occupied every habitable environment in the planet's biosphere. Although microorganisms are known to be responsible for key functions on Earth, such as carbon and nutrient cycling, and determining the health and disease state of the planet's plant and animal inhabitants, greater than 99% of the trillions of microbes thought to exist have yet to be discovered. In addition, high microbial diversity has made it difficult to study specific functions carried out by complex microbial communities in microbiomes.

"In the 1960s, Carl Woese began to analyse 16S ribosomal RNA (16S rRNA) genes, which encode a component of the 30S small subunit of ribosomes in prokaryotes. These genes have both highly conserved as well as variable regions, and therefore can be used to accurately determine microbial phylogeny. Woese profiled 16S rRNA genes from many different bacteria, changing the field of microbiology as we know it, and creating the field of molecular phylogenetics. This paved the way for the idea of metagenomics, or the genomics of communities, by direct extraction and cloning of DNA to capture the yet uncultured microorganisms present among us and provide insight into their function within the community," he concluded.

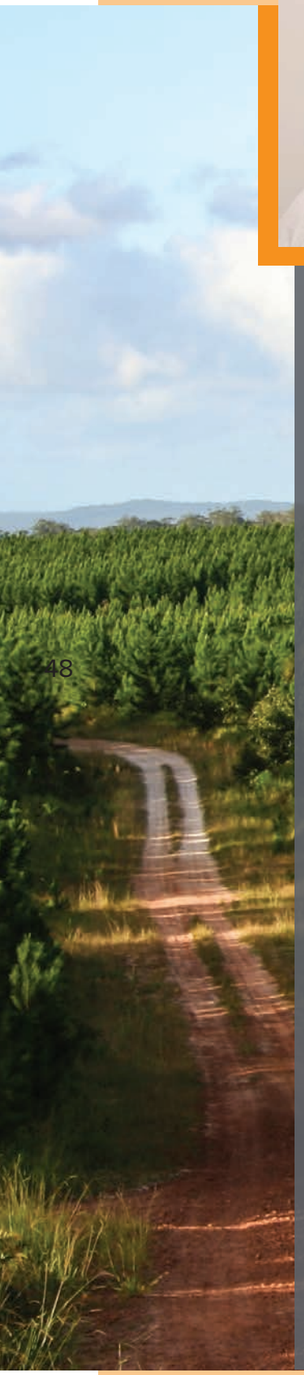
Naga has published papers on using microbiome analyses to treat parasite infections and he explains why it is a particularly useful tool. "Parasitism is a successful lifestyle that has evolved in virtually every clade of multicellular organisms. Parasitology seeks to develop the means to prevent, limit, or cure infections by parasites for the benefit of humans, agriculture, and wildlife and to understand how parasitism and parasitic disease impact not only the host but also host communities and ecosystem health. This is a challenging task, considering the diversity and complex nature of host–parasite interactions. We have limited insight into the nature and importance of these interactions for parasite ecology and evolution, and not a single parasite species has its entire microbiome fully characterised. Microbiome analysis plays a key role in getting more insights in understanding these organisms."

High-throughput sequencing platforms have revolutionised the detection and characterisation of microbial species, and Naga believes that the impressive increases in the speed and throughput of nucleic acid sequencing technologies are at the forefront of advances in microbiome research. "In particular," Naga says, "there has been a revolution in high-throughput sequencing platforms as they have surpassed the traditional Sanger sequencing methods that dominated the field for nearly three decades (from 1977 to 2005). Sequencing a single bacterial genome using the Sanger dideoxynucleotide-based chain-termination approach previously was a major endeavor that took years to complete. The first bacterial genome to be completely sequenced using the Sanger approach was *Haemophilus influenza* in 1995 (with *Escherichia coli* completed in 1997). Currently, because of the availability of rapid and inexpensive sequencing platforms, it is now possible to sequence complete bacterial genomes in a matter of hours."

When asked what Naga enjoys most about working at AGRF, he replied, “AGRF is synonymous to genomics and sequencing. Genomics is changing at a rapid pace, at AGRF we always get hands-on experience with the latest technology, be it sequencing or analysing data. This is very satisfactory and an immense pleasure.”

NAGA
KASINADHUNI

Naga is one of our Senior Bioinformaticians, specialising in Microbial Diversity Profiling and Metagenomics



ELIMINATING ILLEGAL LOGGING

DR ARIF MALIK

Wildlife and forestry crime is the fourth largest illegal trade in the world – and Dr Arif Malik is one of the team behind the prevention of illegal timber logging.

Dr Malik, of The University of Adelaide's Advanced DNA, Identification and Forensic Facility (ADIFF) applies genetic techniques to help eliminate illegally logged timber in international supply chains. In his role at ADIFF, he tests DNA to identify the species and origin of timber, verifying the timber to contribute to the screening that happens on a global level for supply chains. This screening is used to identify and eliminate illegally logged timber, acting as a watchdog for an enormous industry.

"At the end of my PhD in 2019, I applied for an internship with the Defence Science and Technology Group," says Dr Malik, on how he entered the field. "I got it, and it involved me doing some soil metagenomic work for forensic purposes. I was placed at ADIFF, and they had all these fantastic toys and beautiful labs...I instantly fell in love with it."

Professor Andy Lowe, who was heading the timber tracking work, then made Dr Malik a job offer upon the completion of his PhD.

"I've always dreamed of having a job where I could apply all the molecular techniques that I've learned to something that could actually make a difference in the world, and now I've gotten very lucky," says Dr Malik. "I've got my dream job through a combination of hard work and luck."

The DNA identification testing for timber species is crucial in the process of gauging whether claims of species and origin of wood are genuine or not – and often, people who are illegally logging will have forged documentation that supports their false claims. This is where the testing comes in.

"Documents can lie, but DNA doesn't lie," says Dr Malik, explaining how a higher rate of consumer peace of mind can also result. "That's why these tests are important. Not only can this kind of DNA identification testing be used to punish those doing the wrong thing, but it also ensures the consumers that they can have confidence that what they're purchasing is genuine and sustainably sourced."

The work has become a means to monitor and protect global supply chains from becoming laundering routes, as well as reduce the likelihood of illegal activity. In a recent case in the US, bigleaf maple logs were identified by their DNA being matched up upon the timber being taken to the original stumps in the forest. The result, upon presenting the evidence, was a guilty plea.

"This screening can assist in not only catching people doing the wrong thing, but it can also act as a deterrent: if people know that this testing is being increasingly used, then they also know that their chances of being caught are also increasing," says Dr Malik, as he describes recent work undertaken by ADIFF that showed up to 40% of timber being imported into Australia is incorrectly labelled.

In addition to screening timber imports coming into Australia, Dr Malik has been working with a range of national and international partners, but the application of techniques are most closely developed with Singaporean biotech company DoubleHelix Tracking Technologies. This testing is not just creating deterrence for illegal loggers – work by DoubleHelix has also developed a scientifically verified standard, Natures Barcode. This uses DNA and other tests to let consumers know that what they're purchasing is being sustainably sourced. "This could start a shift in the way people think: big companies will feel more responsible," explains Dr Malik. "If consumers can go somewhere where they know their product is sustainably sourced, they will."

Dr Malik concludes by talking about why he chose to come to AGRF as a sequencing partner. "The thing I like most about working with AGRF is how accommodating you've been with some of the ridiculous projects I've had short time frames to get done. There was one instance this year (2020) where a client gave me a week to go from having blocks of timber sitting in my lab to having data presented on a screen – not the easiest this year to get DNA sent to Brisbane quickly during COVID – but I went from having bits of timber to having a report written within 2-3 days."

"It's not just that you do good work - you help me troubleshoot. It's when I've said 'this client needs work done immediately, can you help me out?' - you guys have delivered."

"it's not just you do good work - you help me troubleshoot."

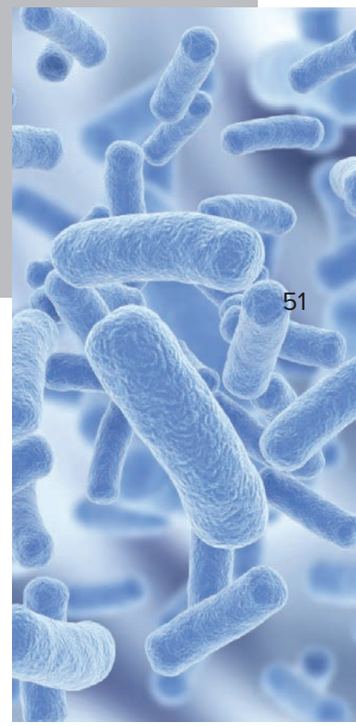
FUNDING PARTNERS

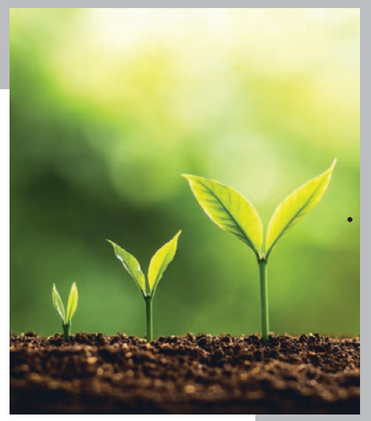
Bioplatforms Australia

Bioplatforms Australia (BPA) provides access to ‘omics technologies for a wide range of academic and commercial users. BPA is enabled by the Australian Government's National Collaborative Research Infrastructure Strategy (NCRIS) and is AGRF’s largest funding partner.

We work closely with BPA on their Framework Initiative projects, which are large national projects comprising researchers from many different institutes, universities, museums, and conservation and wildlife organisations. These projects are of great cultural and economic importance as they provide a genomics resource on many native species, some of which are on threatened species lists. AGRF is an integral partner to BPA’s Framework Initiatives by providing a wide range of innovative genomics platforms to ensure these projects are successfully completed. In addition, our National Next Generation Sequencing Manager, Dr Chris Noune, and Brisbane Node and Platform Manager, Dr David Hawkes, serve on BPA’s Framework Initiative Steering Committees which further solidifies our partnership.

The four major Framework Initiatives that AGRF has collaborated with BPA on in recent years are Genomics for Australian Plants, Oz Mammals Genomics, Threatened Species Initiative, and Australian Microbiomes. Other notable BPA projects that AGRF has worked on include the Stem Cells project, the Australian Research Data Commons Cross-NCRIS OzBarley project, and the Commonwealth Scientific and Industrial Research Organisation’s (CSIRO) Future Science Platforms projects.





The work we do is made possible with the support of our funding partners.
We thank the Commonwealth Government infrastructure funding schemes, administered through NCRIS and BPA.

PARTNERSHIPS

Innovative Wine Production
AGRF continues to support the Industrial Transformation Training Centre for Innovative Wine Production, led by Vladimir Jiranek from the University of Adelaide. This is a five year partnership where AGRF is helping researchers profile the genomes and transcriptomes of microbes and plants that are beneficial to improving wine production.
AGRF investigator: Dr John Lai.

Novel Genetic Tools for Tracking the Origins and Spread of Plasmodium Vivax
Led by Sarah Auburn from the Menzies-School of Health Research, AGRF is collaborating on an NHMRC Ideas grant project: Novel genetic tools for tracking the origins and spread of Plasmodium vivax.
AGRF investigator: Dr Chris Noune.

Understanding Adaptation to Improve Conservation of Australian Flora
Under the leadership of Andrew Lowe (University of Adelaide), AGRF completed a five year ARC Discovery project in December 2020 to understand adaptation to improve conservation of Australian flora.
AGRF investigator: Dr John Stephen.

Integrating Climate Adaptation into Rainforest Restoration Plantings
AGRF also collaborated with Associate Professor Lucas Cernusak from James Cook University who was awarded an ARC Linkage grant in May 2020 to integrate climate adaptation into rainforest restoration plantings. In this collaboration, AGRF's Dr Kenneth Chan will take part in genomics experimental design processes and bioinformatics support, and Dr Chris Noune will develop and improve laboratory and in silico methods in microbiome analyses.
AGRF investigators: Dr Chris Noune & Dr Kenneth Chan.

OzBarley: from Genome to Phenome and Back Again
We collaborated with the Australian Plant Phenomics Factory for the Cross-NCRIS National Data Assets Program's OzBarley project: a barley data and germplasm asset for the Australian research and breeding community. The aim of the OzBarley project is to develop a publicly available Genotype-to-Phenotype (G2P) data asset meeting FAIR principles that is specifically designed by, and for, Australian researchers and breeders focusing on barley as an economically important model crop. The OzBarley G2P data asset will provide the basis for the discovery of important genes and will reduce the barrier to entry for future barley funding applications.
AGRF contributions: cash and various in-kind contributions.

ARC Research Hub for Advanced Prawn Breeding
We concluded a five-year collaboration with Professor Dean Jerry from James Cook University in December 2020 on an ARC Industrial Transformation Research Hub project that was designed to transform prawn aquaculture via advanced breeding. AGRF is continuing this collaboration with Professor Jerry by co-submitting another ARC Industrial Transformation Research Hub grant to supercharge northern Australian aquaculture through genetic solutions.
AGRF investigators: Dr Kirby Siemering, Mr Matthew Tinning.

Using Genetics to Recover Australia's Lost History
AGRF wrapped up its collaboration with Alan Cooper in December 2020 on an ARC Linkage study that used genetics to recover Australia's lost history. This project aimed to use historic hair samples collected by anthropological expeditions in the early 20th Century to generate the first genetic map of Aboriginal Australia to reconstruct Australia's pre-European genetic and cultural past.
AGRF investigator: Dr John Stephen.

Optimising Seed Sourcing for Effective Ecological Restoration
We collaborated with Dr Martin Breed from Flinders University to be awarded an ARC Linkage grant in May 2020 to optimise seed sourcing for effective ecological restoration. AGRF's National Next Generation Sequencing Manager Dr Chris Noune is a partner investigator on this grant where he will be leading the microbial profiling program, which includes developing and testing improved laboratory and in silico methods in microbiome analysis.
AGRF investigator: Dr Chris Noune.

Cooperative Research Centre on Transformations in Mining Economies (CRC TiME)
This multi-institute CRC has approximately $135.4M of contributions from commercial, academic, and government entities. Dr Guy Boggs spearheads this CRC which aims to ensure the local community benefits after closure of mining sites. This encompasses both economic benefits to the community, as well as restoration of both flora and fauna to ensure the land is fit for purpose. AGRF will serve as a technological enabler through novel uses of genomics platforms within different CRC programs.
AGRF contributions: cash and various in-kind contributions.

Seagrass Adaptation and Acclimation Responses to Extreme Climatic Events
Led by Gary Kendrick from the University of Western Australia, AGRF completed an ARC Discovery project in December 2020 that studied seagrass adaptation and acclimation responses to extreme climatic events. AGRF provided the genomics expertise and bioinformatics support to ensure this project met its milestones.
AGRF investigator: Dr John Lai.

Integrating Australian Phage Biobanking and Therapeutic Networks and Delivering Solutions for Antimicrobial Resistance
AGRF has partnered with Professor Jonathan Iredell from the Westmead Institute of Medical Research and the University of Sydney on developing novel therapeutics using viruses to treat antibiotic-resistant bacterial infections.
In-kind contribution: National Research Partnerships Manager, and an NGS technician.

Supercharging Tropical Aquaculture Through Genetic Solutions
AGRF continues its successful collaboration with Professor Dean Jerry who will lead this research hub, which is an extension and successor of the recently concluded Advanced Prawn Breeding Research Hub. This new research hub will begin in 2022 and will build on the advances made by the Advanced Prawn Breeding Research Hub.
Partner Investigators: Dr Chris Noune, Dr Dhanya Sooraj.

Integrated Multimodal Precision Liquid biopsy to enhance Melanoma and NSCLC Treatment
Beginning in 2022, this MRFF project will be directed by Prof. Helen Rizos from the Macquarie University. This clinical genomics project aims to develop liquid biopsy blood biomarkers to inform of disease progression, guide treatment regimes, and to monitor treatment response for various cancers.
Associate investigator: Dr John Lai.

CO-AUTHORED PUBLICATIONS

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As part of the Australian genomics community, we work with a wide variety of organisations throughout Australia. This includes partnerships with our funding bodies, host institutes, and the greater research community. These partnerships and connections allow us to uphold our mission of being a key enabler of genomic research in Australia.

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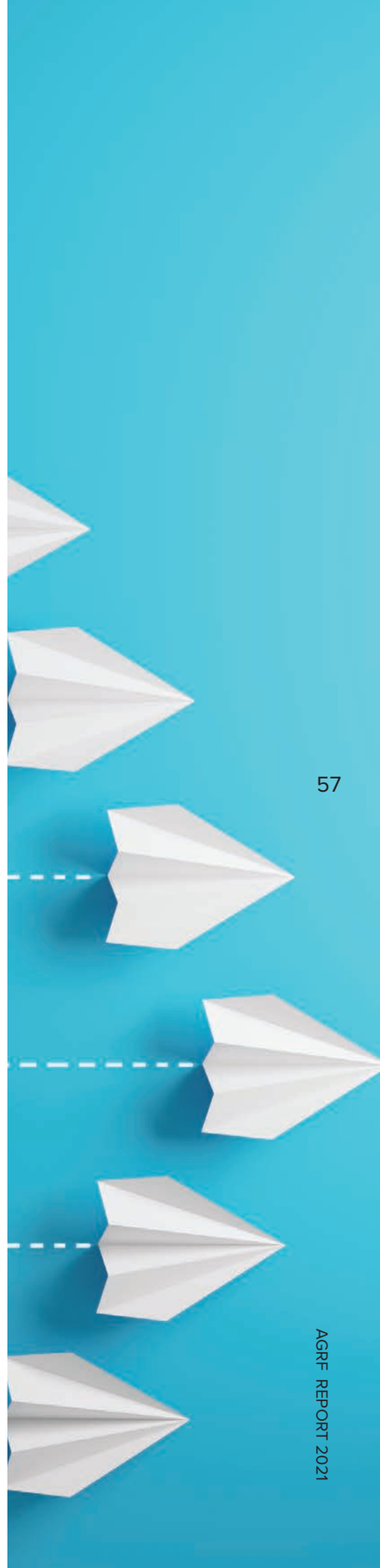

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