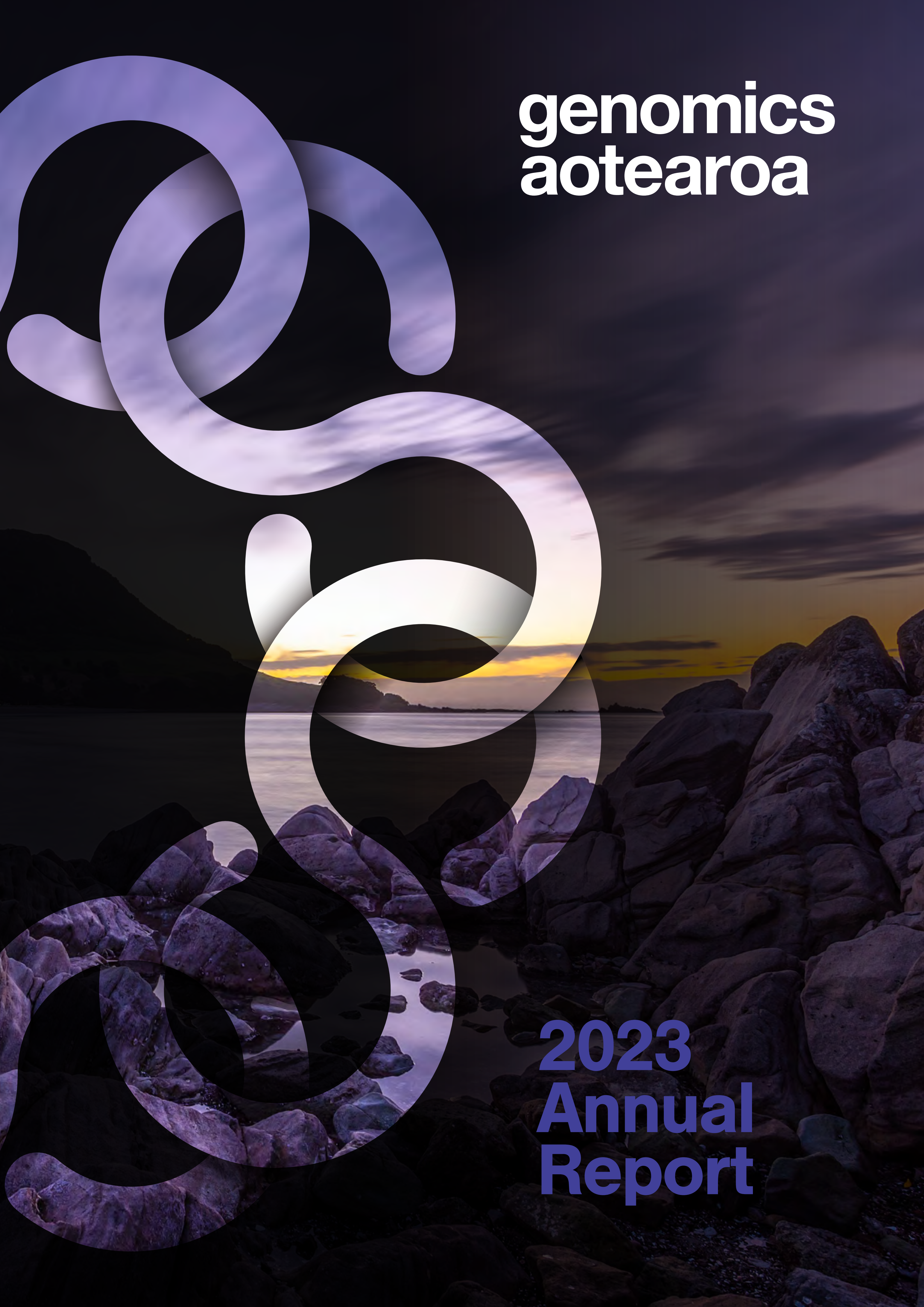

genomics
aotearoa

2023
Annual Report

“The human pangenome will be a key resource in the era of genomic medicine. Therefore, we need to co-develop tools and protocols that are designed to ensure the broadest benefit sharing and respect the rights of Indigenous data ownership”

Professor Karen Miga

Associate Director - University of California, Santa Cruz Genomics Institute, Human Pangenome Reference Consortium

Genomics Aotearoa Annual Report

Year 6

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Report from the Directors

It has been another strong year for Genomics Aotearoa. We have funded new projects, brought new investigators on board, extended our engagement into communities and end-user groups, and delivered new training workshops across the research sector. Business as usual, you might say, for a national genomics infrastructure. But as with any successful business, future planning is critical. In addition to continuing to fund excellent genomic science, we must also consider what genomics can do to mitigate the very real challenges that our nation, and indeed our entire planet, are already facing.

Current events can leave no-one in any doubt that climate change is real and is having impacts globally as well as here in Aotearoa. This comes on top of a biodiversity crisis which has seen the current extinction rate for vertebrates on earth at 100 times the rate it was before human impacts. Extinction rates for other groups of animals, plants and microbes are harder to determine, but suffice to say, our biosphere, that thin lens of life between our planet and space, is facing challenging times.

In human health and wellbeing, we face challenges on a similar scale. The impacts of overpopulation, globalisation and increasing inequity are international problems, but ones that we will need to develop unique solutions for in Aotearoa.

So how might we, as genomic scientists, help with the global impacts to biodiversity and human well-being that will accompany these planet-wide changes? Overseas, one approach is to ensure we understand biodiversity by sequencing the genomes of every known species on Earth. This is a huge undertaking, with massive implications for both the capacity of the science systems, but also the data generated: who owns it and where and how should it be curated and used? These questions are even more important as we, in Aotearoa, live in an ecosystem that has been less intensively studied. Do we know the full variety of life that lives here? Do we fully understand how our ecosystem functions?

What is clear is that genomics is a key tool to understanding the diversity of life around us, as well as improving our production systems, and ensuring our own health and wellbeing. It provides a means to monitor and understand biodiversity, boost and enhance food production, and deliver personalised healthcare. Many of the tools to achieve these goals are already built, but they need further development, and we also need to understand the implications for data sovereignty and ownership should these tools become ubiquitous.

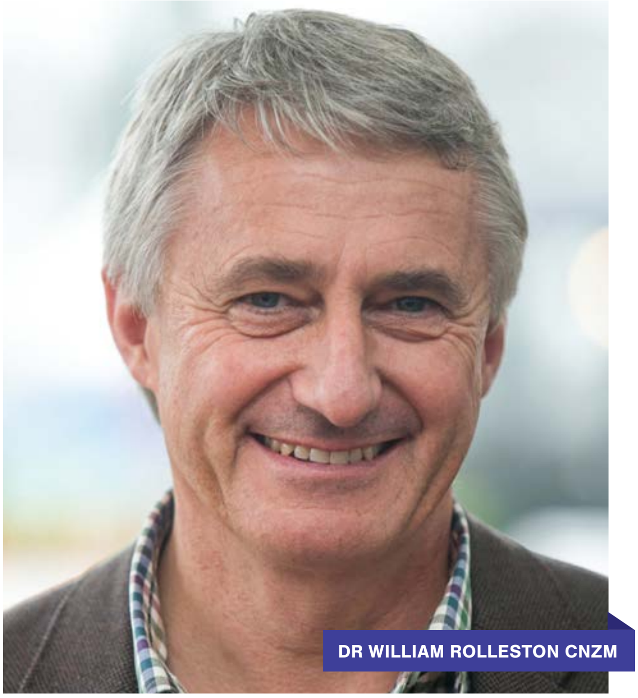
Genomic science will play a key role in how we manage human health, production systems and natural ecosystems in the future. Genomics Aotearoa stands ready for that challenge.

Prof Peter Dearden and Prof Mik Black
Directors of Genomics Aotearoa

PROFESSOR PETER DEARDEN

PROFESSOR MIK BLACK

Report from the Chair


DR WILLIAM ROLLESTON CNZM

Once again, this report highlights the diverse research and incredible achievements supported by Genomics Aotearoa. It is a reminder of the talent and capability we have at hand.

Genomics Aotearoa is approaching the end of its contract with MBIE, so we are giving careful thought to the future. In early 2022, a team of experts, led by Linda Faulkner (Ngāti Rangi) (Tūtaiao Ltd), supported by Andrew Gilbert (Bioplatforms Australia), Dr John Roche (Ministry for Primary Industries), Professor Ingrid Winship (University of Melbourne), and Professor Allen Rodrigo (University of Auckland), reviewed Genomics Aotearoa. The review was very positive and provided a set of recommendations for us to chart our future direction, and what needed change. We are grateful for the advice of this hugely experienced group.

One key outcome from the review was the need for a future-focused genomics strategy for our country and for Genomics Aotearoa. This work is well underway, and has involved extensive engagement with end-users, researchers, international advisors, and others to assess the strengths, challenges, opportunities, and threats faced by genomic research in Aotearoa.

Genomics is fast becoming a key underpinning tool in the biological sciences. The work cannot be undertaken in isolation from our communities of interest, which include researchers in the health, conservation, and primary production sectors, and in turn, their wide range of end-users. The research must involve hapori Māori in the spirit of Te Tiriti partnership and to promote equity. I would personally like to thank the teams of researchers and those communities who make up Genomics Aotearoa. Together, we are planning for the future and know where we are heading.

I particularly want to acknowledge those who took part in the strategy development. Their frank and insightful comments were vital. We expect to complete the strategy late in 2023 to support future decision-making around this science and its application.

Interviewing partners and end-users, gathering and synthesising data to inform a strategy takes a team of talented people. The Operations team, led by Jayashree Panjabi, have worked tirelessly to support the strategy effort, while supporting a wide range of day-to-day activities that underpin the work of Genomics Aotearoa researchers. The team's accelerated focus on outreach and education reflects two of the critical needs identified in the strategy work. I am very grateful for all their efforts.

I would also like to thank our colleagues at MBIE, Chris Miller and Heather Penny, for their advice and unwavering commitment to excellence.

My respect and thanks to our outstanding Board members, who have grappled expertly with both projects and strategies, and have continued to provide such excellent advice to Genomics Aotearoa.

Finally, I would like to thank, and introduce, our new Co-Director Professor Mik Black, who has come on board at exactly the right time as we look to the future.

Dr William Rolleston CNZM
Chair of the Genomics Aotearoa Governance Board

Genomics Aotearoa

A platform for collaboration

Since 2017 the Genomics Aotearoa collaborative platform has been leading, facilitating, and promoting genomics research and bioinformatics in Aotearoa. Genomics (the study of the complete genetic information in living things) and bioinformatics (the computational methods and software that enables analysis of genomic data) are our major research disciplines.

Genomics Aotearoa increasingly supports the development of resources by, with and for Māori, and enables greater participation of Māori researchers, communities, businesses, whānau and students in the fields of genomics and bioinformatics.

Genomics Aotearoa's ten partners represent the genomics expertise of the entire country. The partners include five universities – Auckland, Massey, Otago, Waikato, and Te Herenga Waka – Victoria University of Wellington – and five Crown Research Institutes – AgResearch, Environmental Science and Research (ESR), Manaaki Whenua – Landcare Research (MWLR), Plant & Food Research (PFR) and Scion.

Thirty-four associate organisations represent researchers and end-users of genomics and bioinformatics across all sectors in Aotearoa that intersect with the life sciences.

Our end-users include iwi and community groups, central and local government, health authorities, hospitals, clinicians, patients and their whānau, primary producers, and conservation practitioners. The connections between our consortium and end-users ensure that capabilities and discoveries translate to real-world benefits throughout Aotearoa.

The enabling infrastructure we develop underpins our research outcomes. Our bioinformatics and genomics capability resides in the 234 researchers directly involved in projects, and our close links to the wider community.

Since 2017, the platform has supported 43 world-class early-career scientists in Aotearoa. Postdoctoral fellows have gone on to become lecturers and lead their own research programmes. Doctoral candidates are involved in many projects, as are undergraduate and graduate students. Extensive genomics and bioinformatics training is available to students, scientists, and clinicians. Worldwide linkages capitalise on, and contribute to, international best practice.

Our cultural infrastructure is essential to our vision to honour our commitments to Te Tiriti o Waitangi. The *Aotearoa Genomic Data Repository* (AGDR) is essential to enable the appropriate curation and assignation of cultural rights to genomic data. This unique database is increasingly being used as a research data source. The *Te Nohonga Kaitiaki Guidelines* and additional resources being developed represent best-practice for inclusive research involving taonga natural resources.

Partnership with New Zealand eScience Infrastructure (NeSI) enables our research delivery. The AGDR and our bioinformatics platform, both hosted by NeSI, are free for all researchers in Aotearoa to use.

Our seven-year programme of research which ends in late 2024, is supported by funding from the Strategic Science Investment Fund, through the Ministry of Business, Innovation and Employment (MBIE). In the last year Genomics Aotearoa has been working towards a strategy for genomics in Aotearoa. We have engaged with a broad range of end-users to find out what they want from genomics and are grateful for those contributions. In late 2023 we plan to share these views, and our response to fulfilling these strategic needs to ensure our work continues to be relevant to solve real-world problems and achieve long-term social impacts for everyone in Aotearoa.

Strategic needs identified by the review included greater end-user participation in strategy design, planning for the development and permanence of resources such as the AGDR and bioinformatics platforms, more cohesive engagement to develop and promote the capability of Māori researchers, industry representatives and communities, as well as increasing the scope of benefits to include Pacific communities. Fulfilling these strategic needs will ensure our work continues to be relevant for solving real-world problems and achieving long-term social impacts for everyone in Aotearoa.

Our Partners

THE UNIVERSITY OF AUCKLAND
Te Whare Wānanga o Tāmaki Makaurau
NEW ZEALAND

agresearch
āta mātai, mātai whetū

VICTORIA UNIVERSITY OF WELLINGTON
TE HERENGA WAKA

UNIVERSITY of OTAGO
Te Whare Wānanga o Otāgo
NEW ZEALAND

genomics aotearoa

MASSEY UNIVERSITY
TE KUNENGA KI PŪREHUROA
UNIVERSITY OF NEW ZEALAND

Plant & Food™ Research
Rangahau Ahumāra Kai

scion™
FORESTS ▪ PRODUCTS ▪ INNOVATION

Manaaki Whenua
Landcare Research

E/S/R
Science for Communities

THE UNIVERSITY OF WAIKATO
Te Whare Wānanga o Waikato

Our Associates

- AbacusBio
- AngusPure
- Applied Molecular Solutions
- AsureQuality
- Auckland City Hospital
- Auckland University of Technology
- Beef + Lamb New Zealand Genetics
- Biomatters
- Bioprotection Aotearoa
- Bragato Research Institute
- Brain Research New Zealand
- Cardiac Inherited Disease Group
- Cawthron Institute
- Garvan Institute of Medical Research
- Gene Crypt Limited
- Growing Up in New Zealand
- Hill Laboratories
- LabPLUS
- Livestock Improvement Corporation
- Maurice Wilkins Centre for Molecular Biodiscovery
- Multi-Ethnic New Zealand Study of Acute Coronary Syndromes
- NIWA
- New Zealand eScience Infrastructure
- New Zealand Merino
- New Zealand's Biological Heritage National Science Challenge
- Orion Health
- Pastoral Genomics
- Prevar
- Real Time Genomics
- Royal College of Pathologists of Australasia
- Sophia Genetics
- Te Whatu Ora Waitematā
- University of Canterbury
- Wilderlab

Our Impact

TE AO MĀORI

6 Indigenous Genomics Platform projects

3 Funded in the Reporting Period

Summer Internships for Indigenous Genomics

18 interns

23 speakers

30 Data sets added to the Aotearoa Genomic Data Repository
25 species total

510 Māori variomes sequenced
+ 280 aligned and variant called

RESEARCH + RESEARCHERS

17 Ongoing projects

14 Completed projects

20 Projects this year

10 Alliance partners
led by University of Otago

34 Associate Organisations

234 Researchers and specialists
54 new this year from **34** institutions

43 Postdoctoral Researchers

28 PhDs, MSc, and BSc Hons students

90,000 hours of CPU time on NeSI over the past 12 months, across **17** different projects.

CAPABILITY BUILDING

Bioinformatics training provided to partner and associate organisation researchers **free of charge**.

Trained this year
248 participants in **14 events**
with attendees from 16 institutions

Metagenomics summer school attendees to date:
>80
from **15** institutions

Trained Researchers
1,521 to date from **23** institutions

73 Courses delivered

OUTPUTS

2221 Citations, with **1287** within the last 2 years

29 Publications this year

159 Peer-reviewed publications to date

13,734 Visits to the website this year

YouTube statistics **41,095** views

512 subscribers

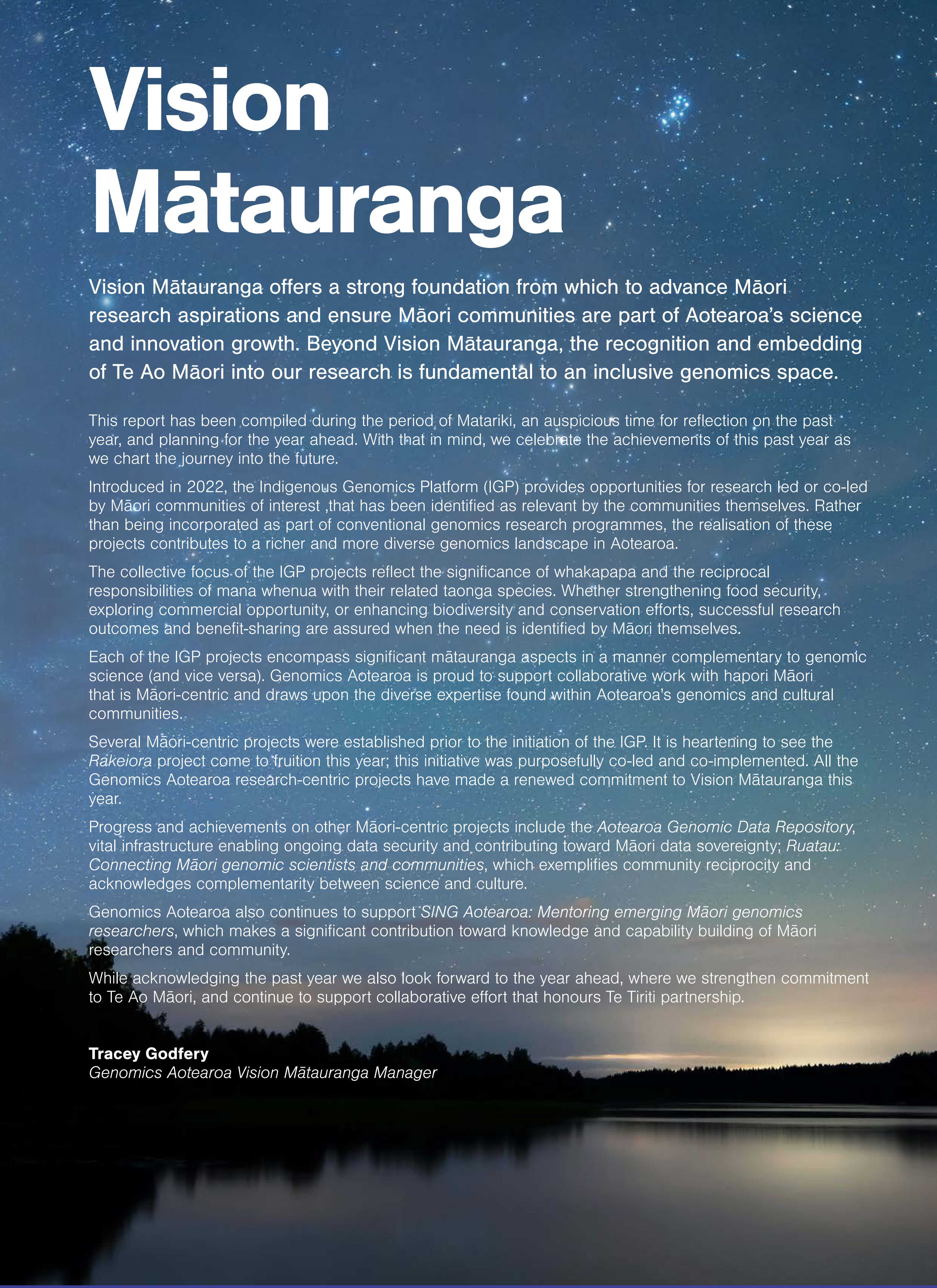
28 videos

675,775 impressions

35 Case studies demonstrating real-world impact

20 Genomics Aotearoa Friday Seminars this year

3 Asia Pacific Genetics Seminars this year



Vision Mātauranga

Vision Mātauranga offers a strong foundation from which to advance Māori research aspirations and ensure Māori communities are part of Aotearoa's science and innovation growth. Beyond Vision Mātauranga, the recognition and embedding of Te Ao Māori into our research is fundamental to an inclusive genomics space.

This report has been compiled during the period of Matariki, an auspicious time for reflection on the past year, and planning for the year ahead. With that in mind, we celebrate the achievements of this past year as we chart the journey into the future.

Introduced in 2022, the Indigenous Genomics Platform (IGP) provides opportunities for research led or co-led by Māori communities of interest ,that has been identified as relevant by the communities themselves. Rather than being incorporated as part of conventional genomics research programmes, the realisation of these projects contributes to a richer and more diverse genomics landscape in Aotearoa.

The collective focus of the IGP projects reflect the significance of whakapapa and the reciprocal responsibilities of mana whenua with their related taonga species. Whether strengthening food security, exploring commercial opportunity, or enhancing biodiversity and conservation efforts, successful research outcomes and benefit-sharing are assured when the need is identified by Māori themselves.

Each of the IGP projects encompass significant mātauranga aspects in a manner complementary to genomic science (and vice versa). Genomics Aotearoa is proud to support collaborative work with hapori Māori that is Māori-centric and draws upon the diverse expertise found within Aotearoa's genomics and cultural communities.

Several Māori-centric projects were established prior to the initiation of the IGP. It is heartening to see the *Rakeiora* project come to fruition this year; this initiative was purposefully co-led and co-implemented. All the Genomics Aotearoa research-centric projects have made a renewed commitment to Vision Mātauranga this year.

Progress and achievements on other Māori-centric projects include the *Aotearoa Genomic Data Repository*, vital infrastructure enabling ongoing data security and contributing toward Māori data sovereignty; *Ruatau: Connecting Māori genomic scientists and communities*, which exemplifies community reciprocity and acknowledges complementarity between science and culture.

Genomics Aotearoa also continues to support *SING Aotearoa: Mentoring emerging Māori genomics researchers*, which makes a significant contribution toward knowledge and capability building of Māori researchers and community.

While acknowledging the past year we also look forward to the year ahead, where we strengthen commitment to Te Ao Māori, and continue to support collaborative effort that honours Te Tiriti partnership.

Tracey Godfery
Genomics Aotearoa Vision Mātauranga Manager

How We Work

With its diverse team, the Genomics Aotearoa consortium is in a prime position to lead Aotearoa's genomics research into the future. Genomics Aotearoa researchers have established national collaborations with end-users across key themes. These themes encapsulate the applied life sciences relevant to Aotearoa New Zealand's economic, environmental, and social wellbeing.

The High Quality Genomes and Population Genomics (HQG+PG) programme has several new sub-projects, including the Mānuka microbiome. These are split over the Environment, Primary Production, and Bioinformatics Infrastructure sections.

ENVIRONMENT

Genomics for biodiversity discovery, conservation, and natural resilience

Projects within Genomics Aotearoa's Environment theme use new approaches to solve some of the most complex genomics puzzles. The challenges include cryptic and mixed genomes from environmental samples, large, complex animal and plant genomes, and getting meaning from data for small populations. As a result, a significant number of new tools, technologies and pipelines continue to be tested, refined, and shared among the genomics community of Aotearoa.

Three projects are reported on this year, *Environmental Microbiomes*, *Tactical Genetic Control of Rats*, and *High Quality Genomes and Population Genomics (HQG+PG)*, with the latter made up of six environment focused sub-projects that increase our understanding of the genomics of a vast array of species, mostly taonga, and a few invasive species threats also. Sequencing the genome is only the beginning – the projects then use those and other resources to develop tools to better manage our natural environment.

HEALTH

Genomics for health equity and the well-being of all New Zealanders

Health is a cornerstone of wellbeing for Aotearoa. Development of, and equitable access to, the most up-to-date tools is key to harnessing the power of genomics for health. The success of health genomics relies on the committed involvement of iwi and community groups, whānau, and clinicians. The vision of health genomics is to enable personalised medicine that targets individual healthcare needs and increases equity in treatment outcomes.

Four projects are reported on this year: *Rakeiora, The Aotearoa New Zealand Variome, Extending Whole Genome Analysis into Healthcare*, and *Identifying the Drivers of Streptococcus pyogenes.*

PRIMARY PRODUCTION

Applying genomic solutions to increase productivity and investment value

Genomics Aotearoa's Primary Production theme aims to develop genomic tools, technologies and approaches for agriculture, horticulture, and aquaculture applications. The projects within this theme aim to better protect Aotearoa from invasive species and pests, improve livestock and crop production and develop new aquaculture initiatives. Many of the approaches taken within the Primary Production projects can be applied across Genomics Aotearoa themes.

Two projects are reported on this year, *Invasomics* and *High Quality Genomes and Population Genomics*. Seven primary production-focused sub-projects are being executed under the *High Quality Genomes and Population Genomics* project. These sub-projects encompass diverse new topics related to primary production, such as the microbiome of Mānuka, pasture genomics and aquaculture, as well as those that build on previous achievements.

TE AO MĀORI

Platform-wide commitment to equity and mātauranga

Genomics Aotearoa infrastructure is not just about the technical platform used by researchers. Cultural infrastructure demonstrates Genomics Aotearoa's commitment to a framework that supports Māori participants, whether they are researchers, students, partners, or end-users.

Genomics Aotearoa continues to support important genomics research in the environment, health, and primary production, and targeted support for Māori aspirations also continues to grow.

Vision Mātauranga provides a foundation for advancing Māori research and ensuring Māori communities are part of Aotearoa's science and innovation growth. Beyond Vision Mātauranga, embedding Te Ao Māori is vital to creating an inclusive and equitable genomics research environment.

BIOINFORMATICS INFRASTRUCTURE

Supporting research and building technical capability

High quality computing infrastructure and support enables the growth of genomics research in Aotearoa, for Aotearoa. World-class researcher bioinformatics skills, secure state-of-the-art computing facilities and data curation; coupled with mentoring future bioinformatics researchers are all essential needs.

Genomics Aotearoa has established a strong foundation for on-going support and capability building in bioinformatics. These foundations include a strong leadership team of experts from several institutions, working groups that review and progress roadmaps for bioinformatics, computing and data management, a full-time training coordinator, and a key partnership with New Zealand eScience Infrastructure (NeSI).

Project Highlights

Genomics Aotearoa projects are all at different stages in their journey towards making an impact.

This section highlights five projects representing each of our themes, that demonstrate the impact generated through supporting high-quality research.

Kākāpō (*Strigops habroptilus*)

ENVIRONMENT:

Genomic disease diagnostics solve the riddle of sudden kākāpō deaths

Back in 2019, in the middle of what had been heralded as a record breeding year, Department of Conservation (DoC) staff became concerned that a disease was sweeping through the kākāpō population breeding on Whenua Hou, near Stewart Island, and called on Genomics Aotearoa researchers to help understand the cause.

Autopsies of dead birds revealed that they were suffering from aspergillosis, a lung disease that can also affect people and other animals. The same disease can be caused by the black mould found in leaky homes.

Genome-wide testing of the *Aspergillus* fungus isolated from the birds allowed researchers to study the origin and dynamics of the disease outbreak in much greater detail than other forms of testing could.

The testing found a single strain of *Aspergillus* from the many samples taken from dead kākāpō, their nests, feeding areas, boats, clothing, beaches, people, and equipment. This was unusual – normally even in a single patient there would be multiple strains of the fungus.

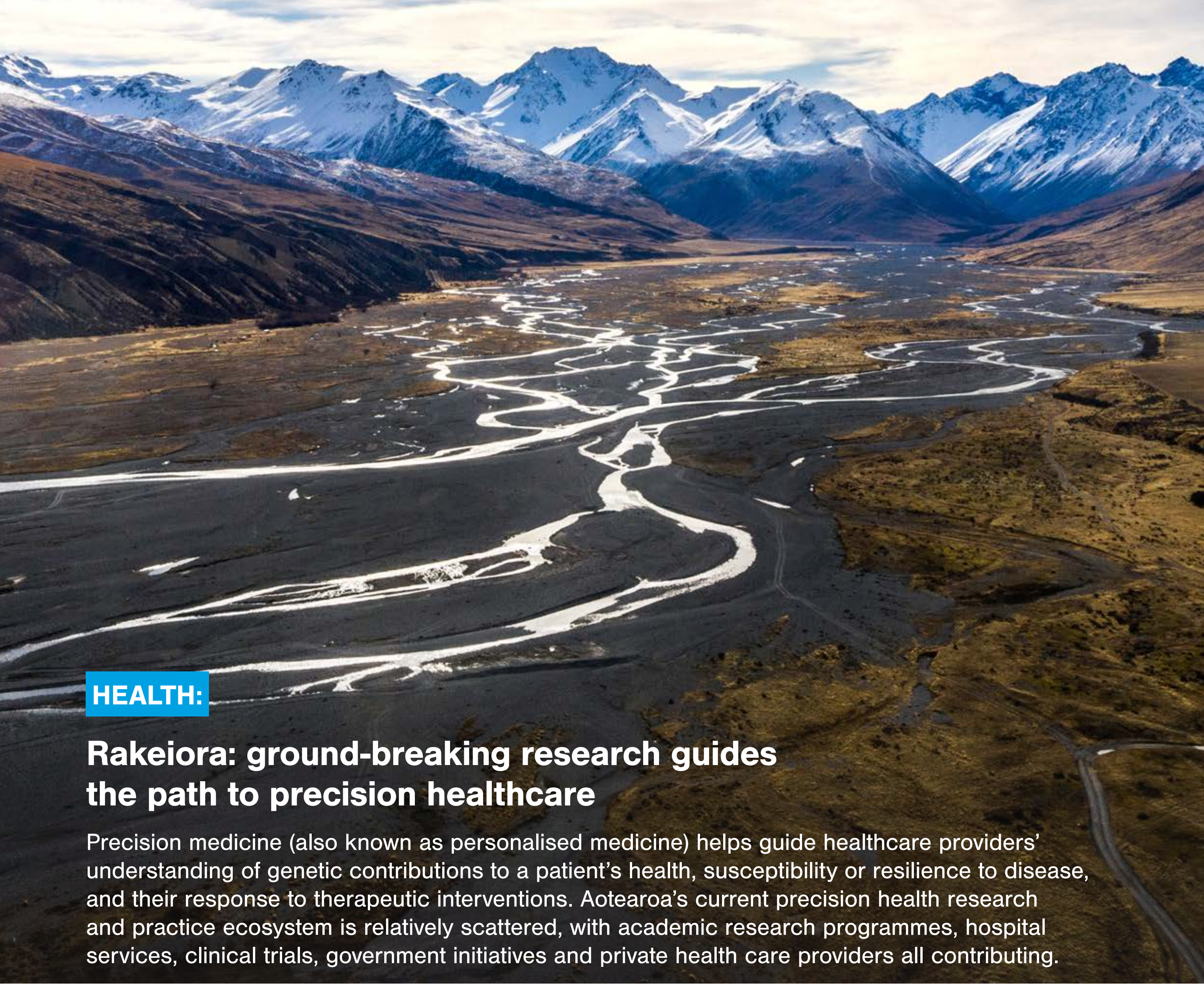
“Because it was kākāpō dying, and because finding only one strain was so unusual, when someone sent out a tweet about it, we got scientists, doctors, geneticists, and immunologists from Imperial College London, Manchester University, University of California, the University of British Columbia, and hospitals in the Netherlands and across Aotearoa contributing their time and expertise to the study”, said project leader Peter Dearden.

Around four percent of the world’s entire kākāpō population was wiped out in this aspergillosis outbreak – something that was surprising given only one kākāpō had died of the disease in previous decades.

The sequencing and intense detective work ultimately revealed a sad truth; the strain was related to the birds’ supplementary grain feed, which humans had carried onto the island and spread.

“Researching the cause of the 2019 kākāpō aspergillosis outbreak has provided information which may prove vital for kākāpō conservation. The findings are already helping us to fine-tune management and care practices to minimise the risk to future breeding seasons, and to the population in general”, said Andrew Digby, Science Advisor Kākāpō/Takahē of DoC.

Image Credit: **Kimberley Collins** (CC by 2.0)



HEALTH:

Rakeiora: ground-breaking research guides the path to precision healthcare

Precision medicine (also known as personalised medicine) helps guide healthcare providers' understanding of genetic contributions to a patient's health, susceptibility or resilience to disease, and their response to therapeutic interventions. Aotearoa's current precision health research and practice ecosystem is relatively scattered, with academic research programmes, hospital services, clinical trials, government initiatives and private health care providers all contributing.

The nationally scalable prototype computational infrastructure developed through the *Rakeiora* project links together whole genome sequence data and healthcare information for individuals and whānau. It is flexible enough to allow different types of federated storage for genomic data from different groups. A powerful but secure "walled garden" environment is used by researchers to analyse the data, while upholding kaitiakitanga (guardianship) and data sovereignty principles consistent with Te Tiriti o Waitangi. The *Rakeiora* prototype also aspires to also incorporate whakapapa (genealogy) alongside genomic and healthcare information.

This nationally scalable prototype provides an evidence base for a new model of healthcare and opens many opportunities for research. Genomically informed precision health in Aotearoa ultimately will lead to improved healthcare for all New Zealanders.

One of the most significant outcomes is that the project's approach ensures Māori have the greatest opportunity to derive value from precision health. This has been through deliberate co-design, co-governance and co-innovation with Māori, respecting Māori data sovereignty, CARE (Collective benefit, Authority to control, Responsibility, Ethics) and FAIR (Findable, Accessible, Interoperable, Reusable) principles and the principle of tino rangatiratanga over Māori genomic resources and Māori hauora data.

Stephen Robertson, one of the *Rakeiora* co-leads, said the equity-driven hard wiring of indigenous values into a nationally scalable genomic research infrastructure is, as a far as we know, unique and strengthens the *Rakeiora* infrastructure for Māori and non-Māori alike.

"The inclusion of mātauranga by Māori to the project generates a far better infrastructure than would otherwise be possible."

The *Rakeiora* infrastructure includes specific procedures for data security and privacy, and aims to ensure that individuals, whānau, iwi, mātāwaka (urban Māori communities and organisations) as well as non-Māori communities can maintain control and sovereignty over their genomic information.

Effective collaboration promoted the inter-weaving of tikanga Māori and western science into the project's processes and protocols.

PRIMARY PRODUCTION:

Enhancing opportunities for blueberry production in Aotearoa and internationally

Blueberries are a delicious economically important premium crop. But they are also a challenge to genomic analysis, as they carry multiple sets of chromosomes. While most animals, like us, have two sets of chromosomes, blueberries and many other plants have more. The various species of blueberry can have two, four or six sets; this phenomenon is known as polyploidy.

Blueberry (*Vaccinium sect. Cyanococcus*)

Genomics Aotearoa and Plant & Food Research have been involved in an international project to improve fruit quality of blueberry and cranberry, led by North Carolina State University, as part of a United States Department of Agriculture National Institute of Food and Agriculture Specialty Crop Research Initiative.

Known as the VacciniumCAP project, the goal is to create new genetic and genomic resources to support the selective breeding of blueberry cultivars with improved fruit quality. The aim is to understand the factors that contribute to quality, and what will lead to greater economic value.

The team at Plant & Food Research has created the first full genome sequence of this polyploid species. But genome analysis in polyploid species is complex, and sequencing the genome is just the start.

The team's analysis of the assembly identified a set of candidate genes and their variants that control the phytochemical composition of blueberry, specifically the compounds responsible for pigmentation of the fruit skin, which mediate antioxidant properties. These characteristics are prime targets for fruit quality improvement.

David Chagné of Plant & Food Research said polyploid genomes have been notoriously difficult to analyse. "Our new methods enable us to crack complex genomes and shed new light on the genetic control of key traits in such species."

The blueberry genome work builds on the team's Genomics Aotearoa supported project to sequence the world's first bilberry genome, with which blueberry shares a common ancestor, in 2021. The close relationship between bilberry and blueberry genomes helps track down the genes responsible for key traits.

Sara Montanari, Plant & Food Research blueberry molecular breeder, said the genome resource is now helping to identify which variants contribute to higher antioxidant content. "We can now develop a DNA marker for selecting new blueberry cultivars that have high antioxidant properties."

As well as creating a new solution and methods that can be implemented in other polyploid species, this high-quality blueberry genome resource could be a game changer for producers in Aotearoa and beyond.

Haku (*Seriola lalandi*)

TE AO MĀORI:

Genomic resources help Ngā Iwi i Te Rohe o Te Waiariki develop aquaculture for haku

Kaimoana are taonga that are interwoven with Māori whakapapa as direct descendants of Tangaroa. Ngā Iwi i Te Rohe o Te Waiariki have developed the Smart Māori Aquaculture initiative, recognising that wildstock fisheries are in decline while demand for protein is growing. The iwi collective is critically investigating alternative ways to supply high value kaimoana for whānau and for global food markets.

Genomics Aotearoa researchers are honoured to be part of this exciting partnership that is responding to a specific need within an iwi-led initiative.

Ngā Iwi i Te Rohe o Te Waiariki want to develop haku (kingfish), among other finfish species, aquaculture in their rohe in Toi-te-Huatahi (the Bay of Plenty) and need to generate genomic knowledge and resources to do this.

Understanding the genomic basis for desirable traits is fundamental to help select the most suitable wild individuals to form a broodstock for the region, and to develop breeding resources to select for best performing individuals from the selected haku genotypes.

Chris Insley (Te Arawa Fisheries), who leads Ngā Iwi i Te Rohe o Te Waiariki, Maren Wellenreuther and David Chagné (Plant & Food Research) and Peter Ritchie (Te Herenga Waka - Victoria University of Wellington) have been working together on this Indigenous Genomics Platform project since 2022.

Having scientists and iwi partners working side-by-side ensures the project plan is fit for purpose.

Haku samples for genomic analyses have already been sourced by Te Arawa Fisheries from multiple locations around Toi-te-Huatahi.

Naturally, Ngā Iwi i Te Rohe o Te Waiariki, as kaitiaki of haku from the Bay of Plenty, will have ownership and control over genetic data and its access, and all results and interpretation will be shared among the research partners.

“Together we are beginning a journey towards genomic selection that will enable innovation of a taonga for aquaculture, supporting a ground-up approach for generating new capability and supporting iwi to exercise rangatiratanga and kaitiakitanga over haku aquaculture in Aotearoa,” Maren said.

BIOINFORMATICS INFRASTRUCTURE:

Metagenomics Summer School builds capability across the motu

Metagenomics is the study of the genetic material recovered from environmental samples – a mixture of DNA from multiple organisms. The technique has a wide range of practical applications. Metagenomics is already used in a variety of contexts, including the influence of the gut microbiome on human health, diagnostics of unknown diseases in animals for production and conservation, and biogeochemical cycling in the environment.

Genomics Aotearoa is continuing to build capability to make full use of this emerging practice. That means investing in training, something the Genomics Aotearoa *Environmental Microbiomes* team at the University of Auckland has successfully addressed through its annual Metagenomics Summer School.

The goal of the summer school is to increase exposure to open-source bioinformatics methods used in environmental metagenomics, and the Summer School is raising awareness of what metagenomics can deliver, and providing networking opportunities.

Lead researcher Kim Handley (University of Auckland) said that the impact of the school is demonstrated by its popularity. “We’re pleased with the results – we are now seeing enhanced bioinformatics skills in researchers, across teams and their organisations. That capability ultimately creates more opportunities for researchers.”

The week-long in-person workshop is both practical and theoretical, focusing on current best practices in metagenomics, and guides learners step-by-step through a metagenomics workflow with hands-on analysis in a mini-project.

NeSI provides computational resources, troubleshooting assistance, and training in command-line fundamentals.

The joint team has created an open-source workflow resource. Genomics Aotearoa Trainer Jian Sheng Boey said this is providing a base for learners from universities, CRIs and the private sector to start applying metagenomics in their research.

“The workflow we have developed for this workshop is highly flexible. It can be a jumping-off point for tailoring workflows better suited to a group’s research questions or needs. And the beginner-friendly structure and open-source materials helps build capability within a research group new to metagenomics,” he said.

One person who has attended and is now using their new-found skills is Priscila Salloum of the Department of Zoology, University of Otago.

“As well as working on a pilot project in our lab to use metagenomic data and the potential to train students in the near future to incorporate the approach into their projects, I have been writing a Marsden Fast Start application that is going to require metagenomics analysis, which was greatly inspired by the knowledge acquired in this workshop.”

Environment

This year saw the *Environmental Microbiomes* project continuing to build capability nationally in metagenomics and reveal ever more insight into the role of viruses in microbial ecology.

In addition, the six *High Quality Genomes and Population Genomics* sub-projects are increasing our understanding of the genomic nature and function of a vast array of species.

The *Tactical Genetic Control of Rats* project, is attempting to develop novel rat control methods.

PROJECT LEAD:

Dr Kim Handley (University of Auckland)

Environmental Microbiomes

The Influence of Viruses on Microbial Dynamics of Aquatic Systems

Metagenomics is rapidly transitioning into a mainstream tool in microbial ecology and is now routinely applied to understand communities of prokaryotic organisms. The *Environmental Microbiomes* project extends the scope to include the DNA viruses that infect these prokaryotic organisms.

In the last year, the research has been supported by new facilities to generate complete bacterial culture isolate genome sequences from long-read sequencing. These genome sequences allow the analysis of integrated viruses and estimation of their prevalence among taxonomic groups. Analysis of environmental data enables the description of a range of characteristics of natural microbial communities.

The team also reconstructed 19,239 unique partial or full putative viral genomes (647 high quality) along a freshwater to marine gradient. Few of these genomes clustered closely with reference viral sequences, reflecting how little of the vast degree of viral diversity we are aware of.

The research contributed to capability development through a comprehensive viral genomics workflow for processing environmental whole genome sequence data

for the study of DNA viruses. Several steps from this workflow have also been incorporated into the annual Metagenomics Summer School workshop.

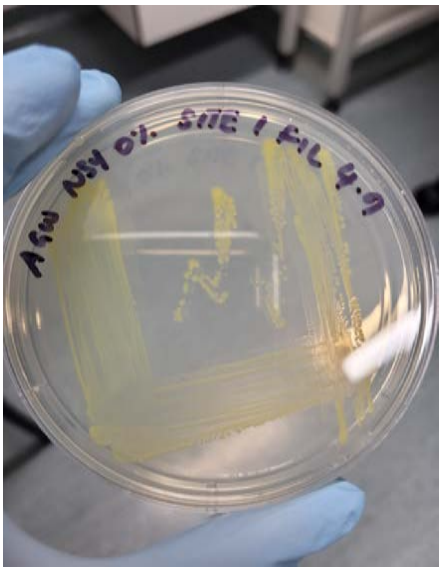
The project has also contributed significantly to early career researcher (ECR) development, with the completion of six PhD theses during the year (either directly or indirectly facilitated by Genomics Aotearoa).

Genomics Aotearoa funded PhD student Hwee Sze Tee also co-developed a course at the University of Auckland, *Practical Approaches in Genomics*, based on her research. The course has now trained over 70 graduate students, and has received contributions from several Genomics Aotearoa-funded researchers, with course computing needs supported by NeSI's Dinindu Senanayake in labs conducted using the Jupyter hub.

The project has been promoted through five domestic and international events. Several peer-reviewed articles have been published to date, either with direct or indirect Genomics Aotearoa support.

In the coming year the team is looking to engage further with Māori, including working with Genomics Aotearoa's Vision Māturanga Manager Tracey Godfrey to translate and communicate research outcomes back to iwi.

Waiwera River



PROJECT LEADS:

Dr Björn Oback (AgResearch)

Dr Catherine Collins (University of Otago)

Assoc Prof Maren Wellenreuther (Plant & Food Research)

Assoc Prof Ocean Mercier and Dr Sara Belcher (Te Herenga Waka – Victoria University of Wellington)

Tactical Genetic Control of Rats

Funded by Predator Free 2050 Limited

Advancements in genetic technologies have opened up new possibilities for pest control, and the *Tactical Genetic Control of Rats* project in Aotearoa New Zealand is leading the way in this field. This project aims to develop novel rat control methods to potentially contribute to Aotearoa's ambitious Predator Free 2050 goal, making it possible for a future where native ecosystems can thrive without the devastating impact of these invasive pests.


Image Credit: **Ouwesok** (CC BY-NC 2.0)

Norwegian Rat (*Rattus norvegicus*)

Two species of rat are the focus of this work: the Norwegian rat (*Rattus norvegicus*) and the ship rat (*Rattus rattus*). Norwegian rats (also known as brown rats), arrived in the late 1700s on whaling ships and with European settlers, adapting to urban and rural environments. They are strong swimmers that can cross waterways up to 2.2km. These rats pose a significant threat to ground-dwelling native species, particularly ground-nesting birds near braided rivers and beaches. Ship rats (also known as black rats), share a similar introduction but did not establish themselves until after the 1860s. They prefer forested areas and are skilled climbers and nest invaders, endangering native bird species. They also eat the fruit and seeds of native plants, as well as wētā, spiders, and beetles. The Predator Free 2050 Limited program aims to manage and eradicate these invasive rats to protect native plants and wildlife.

Since February 2022, Genomics Aotearoa has actively contributed to the Predator Free 2050 Limited funded *Tactical Genetic Control of Rats* project, employing a comprehensive three-pronged approach through strategic partnerships with AgResearch, the University of Otago, Plant & Food Research, and Te Herenga Waka - Victoria University of Wellington.

AgResearch has spearheaded the first aim of the project by pioneering the development of a transgenic Norwegian rat control system within secure containment facilities. This endeavour involves the integration of a genetic construct into the Norwegian rat's genome, ultimately resulting in sex-specific mortality in offspring.

The University of Otago and Plant & Food Research are using population data to understand the differences between the Norwegian rat and the ship rat, facilitating a deeper understanding of the gene flow dynamics of these species. Understanding rat population interactions on a genomic basis helps inform on the consequences of any potential future introduction of transgenic rats to existing populations.

Te Herenga Waka - Victoria University of Wellington has taken the lead in the project's final objective, which entails the assessment of attitudes toward the single-sex offspring control approach. A dedicated focus on understanding mātauranga Māori knowledge with regards to predator control and the application of transgenic technologies such as this enables proactive identification and potential mitigation of concerns. A better understanding of public opinion will help guide any ongoing development of this technology.

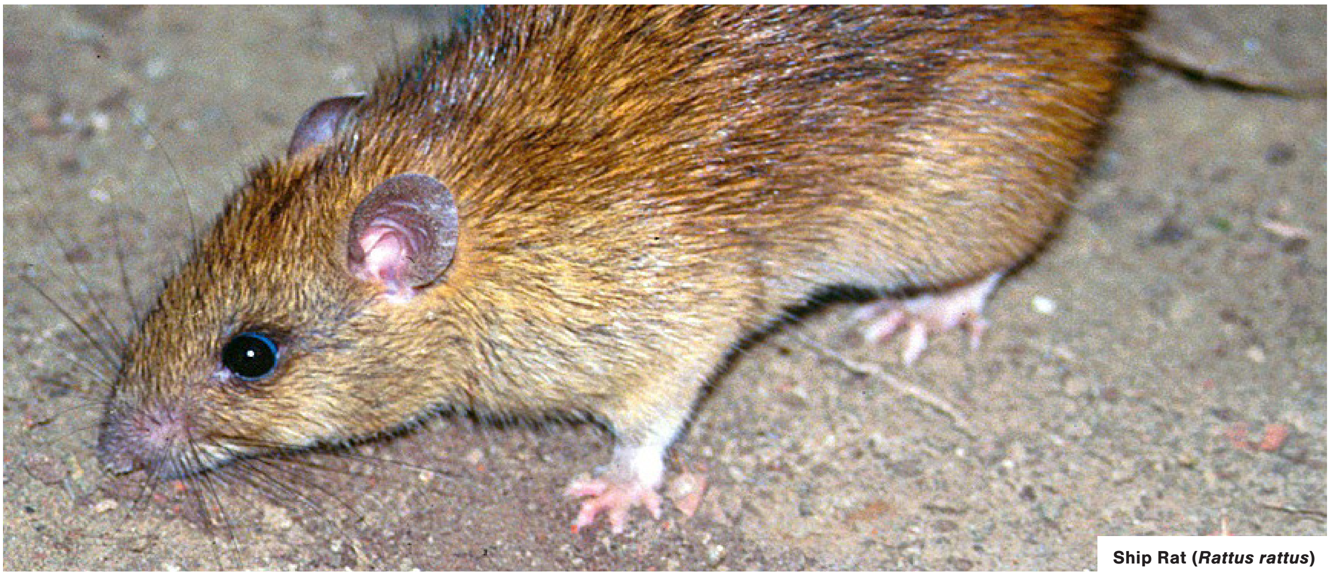


Image Credit: **CSIRO** (CC by 3.0)

Ship Rat (*Rattus rattus*)

PROJECT LEADS:

Assoc Prof Thomas Buckley (Manaaki Whenua – Landcare Research)
Dr Shannon Clarke (AgResearch)
Dr David Chagné (Plant & Food Research)
Assoc Prof Anna Santure (University of Auckland)

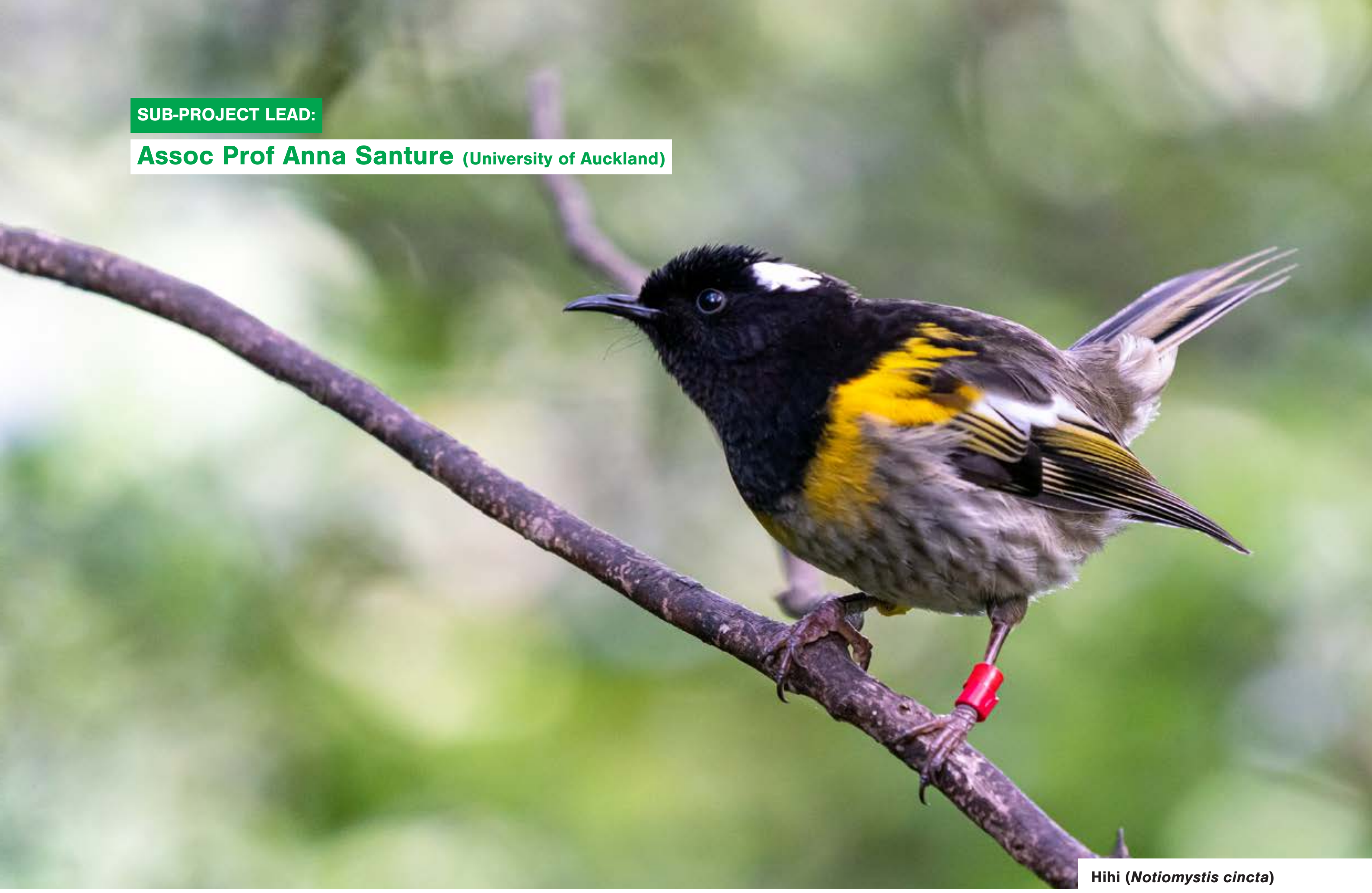
High Quality Genomes and Population Genomics

Kākāpō (*Strigops habroptilus*) - "Hugh"

High quality genomes are increasingly being used as tools to help threatened species recovery. Potential adaptive resilience to environmental instability, such as climate change, requires knowledge of the genomes of entire populations of a species (the pangenome), and their variation (the variome). Increasingly, population genomics tools are within reach of conservation managers. Honouring Te Tiriti o Waitangi is critical to the success of research on taonga, and conservation outcomes.

The *High Quality Genomes and Population Genomics* (*HQG+PG*) project includes sub-projects within the Environment and Primary Production themes, and some have an equal focus on Bioinformatics Infrastructure so are presented in those later sections.

Image Credit: **Jake Osborne**


SUB-PROJECT LEAD:

Assoc Prof Anna Santure (University of Auckland)

Hihi (*Notiomystis cincta*)

Understanding Hihi Genomics
to maximise their conservation management systems

Hihi are a threatened bird found only in predator-free sanctuaries. Hihi are actively managed by the Department of Conservation (DoC) under advice from the DoC Hihi Recovery Group, which includes DoC staff, research scientists, conservation practitioners and Ngāti Manuhiri, who are kaitiaki for hihi.

The hihi genomics sub-project investigates the potential of hihi to adapt to environmental change in the face of their low genetic diversity and has developed a 'genetic health check' for hihi to inform conservation management of the species.

In the past year the project leveraged the hihi genome assembly to assess genetic data of 400 birds from the Tiritiri Matangi population. Individuals with high levels of inbreeding suffer higher mortality as juveniles, and inbreeding at a specific part of the genome is predictive of low lifetime reproductive success (fitness).

The integration of genomic and long-term demographic data from Tiritiri Matangi suggests that active management for hihi is only somewhat successful in buffering the impacts of inbreeding and low genetic diversity of the population. However, active management interventions that increase genetic diversity, such as translocating individuals between populations, are likely to be beneficial for species survival.

The team recently welcomed a new PhD student; Hui Zhen Tan will be exploring the impact of inbreeding on hihi across several populations.

Genomics Aotearoa postdoctoral fellow Annabel Whibley joined the Bragato Research Institute as a Bioinformatician in late 2022 and PhD student Laura Duntsch is now a postdoctoral scientist at Livestock Improvement Corporation. It is encouraging to know that these early career researchers have positions with Genomics Aotearoa associates, and that such talented people are retained within Aotearoa.

The project team acknowledges Ngāti Manuhiri as kaitiaki for Te Hauturu-o-Toi and for its taonga species hihi.

The work has been presented at four domestic and international conferences and has generated five peer-reviewed articles, as well as receiving considerable media attention.

Image Credit: **Judi Lapsley Miller** (CC by 4.0)

SUB-PROJECT LEAD:

Prof Neil Gemmell (University of Otago)

PROFESSOR NEIL GEMMELL, DR ALLISON MILLER, AND DR ALANA ALEXANDER (UNIVERSITY OF OTAGO)

Genomics of Taonga Species

deriving genomic information from imperfect samples

The Genomics of taonga species sub-project goal is to provide high quality genome assemblies, and analysis for several taonga to enhance species recovery and management plants.

To date the team has assembled, polished, and analysed the genomes of kanakana (New Zealand lamprey), takahē and peka peka (short-tail bat) in partnership with iwi, DoC, and other stakeholders.

The focus continues to be on the generation of genomes from samples of mixed quality (old, low quality, poorly stored) using a variety of size selection and genome sequencing approaches.

The team has completed de novo transcriptome based and comparative annotation together with fast and reliable SNP calling, to empower population genomic and comparative genomic analyses for peka peka and kanakana (lamprey). These annotated assemblies and SNP variant sets are, and will, form the basis of multiple publications looking at the evolutionary history, and population genomics of these taonga, empowering better conservation management.

In the last year, capability-building has included the graduation of Allison Miller, whose PhD on lamprey population genomics was graded as "exceptional". In addition, one of the papers from her thesis won the American Genetics Association student authored paper of the year. Keeping capability onshore, former Genomics Aotearoa postdoctoral fellow Alana Alexander (University of Otago) and Allison's co-supervisor is now a Rutherford fellow and lecturer at the University of Otago. Alana also co-leads the *Ruatau* project, which you can read more about in the Te Ao Māori section.

For each project the team have built strong working relationships and partnerships with Māori. Jane Kitson (Ngāi Tahu and Ngāti Mamoe me Waitaha) is our collaborator for kanakana work. Takahē iwi relationships are managed via the DoC Takahē Recovery Group through Tāne Davis (Ngāi Tahu). Ngāti Rangi are the team's iwi contacts for peka peka.

The project has generated six peer reviewed publications in the reporting year.

SUB-PROJECT LEADS:

Assoc Prof Thomas Buckley (Manaaki Whenua – Landcare Research)

Prof Tammy Steeves (University of Canterbury)

Genomic Solutions

for threatened species and those with large and complex genomes

Giant Wētā (*Deinacrida heteracantha*) - "Dave"

This sub-project is designed to support the management of threatened species and build knowledge of our biodiversity.

End-users are key to the success of the project and include mana whenua who are kaitiaki for the species the team studies and the Department of Conservation (DoC). The current species the team is assisting with are kuaka Whenua Hou (Whenua Hou diving petrel), wētāpunga, huhu, rātā Moehau, stick insect and native pepeketua (*Leiopelma* frogs).

The kuaka Whenua Hou genome is now being used to call SNPs from re-sequenced population data. Preliminary analyses show a lack of significant gene flow between the common diving petrel and the highly endangered kuaka Whenua Hou, which has practical implications for conservation of kuaka Whenua Hou.

Wētāpunga are subject to captive rearing for translocation, but until now this has been without information on the impacts of this on genetic diversity. The team has generated PacBio data for the species and are in the final stages of collecting Hi-C data to complete the genome assembly. Our genomic insights will help understand the impacts of captive rearing and translocation on genetic diversity.

The huhu genome has been analysed together with bacterial metabarcoding data, which show a difference in microbiome between different native tree hosts. This feature aligns with mātauranga understanding, which demonstrates how these different disciplines can complement each other.

The team has also generated PacBio data from pepeketua and are collecting Hi-C data to complete the genome assembly.

Postdoctoral fellow Natalie Forsdick has contributed to a huge range of capability-building this year, including developing teaching materials, helping deliver the Otago Bioinformatics Spring School, delivering a workshop for ECRs with Jana Wold at the University of Canterbury, a seminar for the MWLR Molecular Group, helping with the GA/NeSI outlier analysis workshop led by Kat Stuart, and the R for genomics workshop, as well as contributing to outreach.

Engagement with mana whenua is naturally key to this collaborative project. In the reporting year the team has worked with Ngāi Tahu and Ngāti Whatua o Kaipara on high quality genome assembly, data management and knowledge sharing.

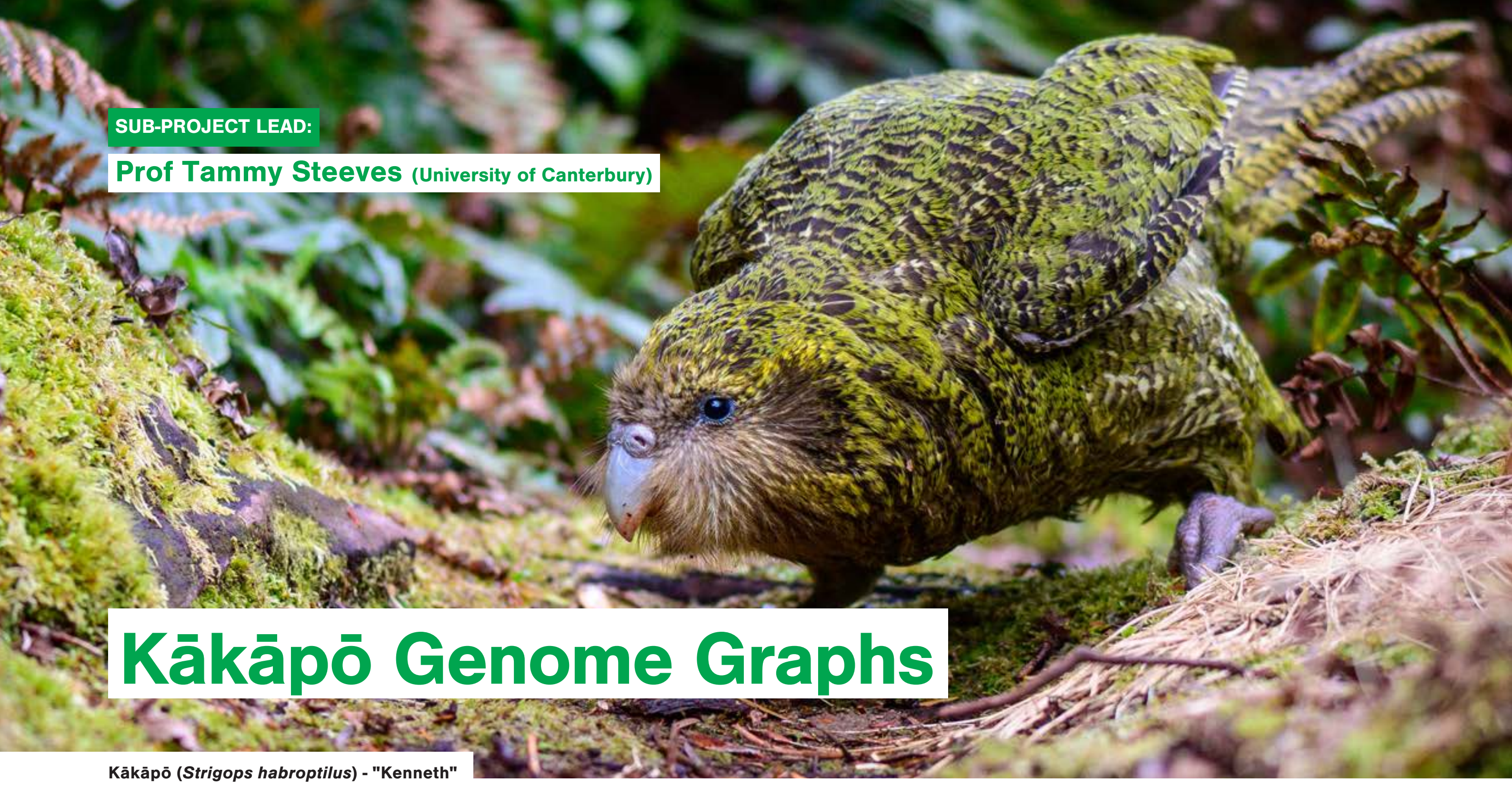
The team is also working with Ngāti Kuri on taonga genomic data management (rātā Moehau and stick insect) and knowledge sharing with Te Awahohonu Forest Trust on huhu beetle.

Outputs from the project have include three new bioinformatics pipelines, eight presentations at events, and one DoC technical report.

SUB-PROJECT LEAD:

Prof Tammy Steeves (University of Canterbury)

Kākāpō Genome Graphs



Kākāpō (*Strigops habroptilus*) - "Kenneth"

Kākāpō, Aotearoa's charismatic but critically endangered night parrots need all the help they can get, and genomics tools are assisting their conservation in many ways.

Genetic and genomic diversity are an important component of adaptation. Single nucleotide polymorphisms (SNPs) are routinely used to measure genome-wide diversity. However, structural variants (SVs, chromosomal rearrangements ≥ 50 base pairs) represent a higher proportion of genomic variation, intersect more often with genes and gene regions and, like SNPs, are associated with adaptive and maladaptive traits.

This kākāpō genome graph sub-project presents a case for the routine characterisation of SVs in threatened species.

To demonstrate how this could work in practice, the team began sequencing low coverage Oxford Nanopore (ONT) long reads for 10 highly represented individuals in the extant population. They are now sequencing high coverage ONT long reads for primary cell lines of two kākāpō generated in an aligned sub-project and assembling the first genome graph for kākāpō.

While the real-world impact of our research will be demonstrated in our partner sub-project, the underpinning kākāpō genome graph is critically important to delivering that impact.

The foundational resources generated will be used to characterise SV diversity in kākāpō at the population level. In addition to shedding early light on SVs and the small population paradigm, as previously envisaged, the team will leverage the kākāpō genome graph to extend recently developed tools to identify the genomic basis of fitness-related traits like early hatching failure.

The team works closely with other kākāpō research teams, as well as the Kākāpō Recovery Group of DoC, including Tāne Davis, the Ngāi Tahu representative. Tāne is the kaitiaki for the kākāpō data produced for this sub-project and cared for tin the AGDR.

Both lead researcher Tammy Steeves, and researcher Jana Wold have been invited to speak about the work at international conferences.

Lead researcher Tammy Steeves was awarded a Regional Distinguished Service Award 2022 (Highly Commended) by the Society for Conservation Biology Oceania for her genomics work and the team's work was nominated for the Molecular Ecology Prize 2022 by the *Molecular Ecology* journal.

Kākāpō (*Strigops habroptilus*) - "Stefan"

SUB-PROJECT LEAD:

Prof Peter Dearden (University of Otago)

From Charisma to Enigma

New tools for kākāpō and invertebrate genomics

Kākāpō (*Strigops habroptilus*) - "Makorea and Willans"

Genomics Aotearoa has another kākāpō sub-project to develop genomic tools to assist DoC with managing the critically endangered kākāpō.

The extensive kākāpō genomics research programme is spread across several research activities, and collaboration is a key factor in the success of this programme, which can be seen from the papers published this year alone.

DoC is intimately involved in the research programme. The sub-project provides them with breeding and management advice and tools they can use to support decision making. They have already made breeding and intervention decisions based on this genomics research. The research has changed the way DoC manages kākāpō and provides lessons for the management of conservation species worldwide.

This year the team has sequenced the genomes of the next generation of kākāpō chicks, which provides a dataset to validate previous predictions.

As well as working on the charismatic kākāpō, the team is developing genomics resources for invertebrates. This year the genome of the enigmatic native velvet worm was completed. The work required a new method for producing high-quality insect genomes rapidly and cheaply. The velvet worm genomics research was featured by New Zealand Geographic and Radio New Zealand.

Sub-project lead Peter Dearden has been involved in several outreach activities, including University of the Third Age (U3A) public lectures, and postdoctoral researcher Joseph Guhlin has assisted with training in a series of workshops on genome assembly.

The team also works on other invertebrates, including honeybees.

Little Barrier Island / Te Hauturu-o-Toi Island Rangers (*Homo sapien*) - "Richard and Leigh" | Kākāpō (*Strigops habroptilus*) - "Flossie" (in carrier case)

All Images Credit: **Jake Osborne**

SUB-PROJECT LEAD:

Assoc Prof Libby Liggins (Massey University)

Whole Genome Sequencing
of the Australian long-spined urchin and kelp

Australian Long-Spined Urchin (*Centrostephanus rodgersii*)

The goal of the sub-project is to generate high quality genome sequences for two marine species that are of major ecological significance and economic potential for primary production and fisheries – the Australian long-spined urchin (*Centrostephanus rodgersii*) and kelp (*Ecklonia radiata*).

The genomic resources created will provide information for comparative studies, be used as a reference for population genomic studies, and will inform the maintenance of strains and selective breeding.

Importantly, Te Whānau ā Apanui's own aspirations motivate the work on kelp. They want to undertake selective breeding of algae but are also very aware of the potential for genetic pollution of wild populations. Members of Te Whānau ā Apanui were directly involved in the sample collection. The choice of locations and collection activity followed their tikanga.

The completed genomes and collected biological material will have Biocultural Labels attached to them by Te Whānau ā Apanui. These Labels ensure that any future use of these genetic resources requires their consent as kaitiaki and will provide the iwi an opportunity for benefit-sharing.

The kelp genome will be used to inform selective breeding of kelps for favourable traits and for population genomic assessments of genomic diversity in wild populations of kelp in the rohe of Te Whānau ā Apanui and neighbouring rohe. This baseline understanding of diversity will inform the selection of individuals for cultures, and monitoring the region for genetic pollution once the open water farming is established.

The Australian long-spined urchin genome will provide a reference for existing population genomic data. These data will help assess whether the populations in the rohe of Te Whānau ā Apanui are the result of mixing between two distinct populations, heightened genomic diversity, or whether the level of genomic diversity is typical of this species (which has the tendency to become a pest).

The sub-project has experienced several technical challenges. The high heterozygosity of the urchin genome is proving challenging to assemble. To date several draft assemblies have been completed with a variety of assemblers and flags. While the high heterozygosity creates unique challenges in the assembly stage, it may also signify interesting biological aspects that help this sea urchin expand its range in Aotearoa. Algal species, particularly kelp, have unique characteristics and protective mechanisms that make DNA extraction difficult. Much time has been invested in perfecting a DNA extraction method so that genome sequencing can begin.

The sub-project is contributing to capability-building. A PhD student, Melissa Nehmens, joined the team in February and has made immense progress.

CASE STUDY:

Genomic insights assist conservation of kuaka Whenua Hou

With only 200 kuaka Whenua Hou (Whenua Hou diving petrel) breeding in a single 1.8-hectare colony on Whenua Hou (Codfish Island), the entire species is at risk if a disaster impacts breeding at that site.

Kuaka Whenua Hou / Whenua Hou Diving Petrel (*Pelecanoides whenuahouensis*)

Image Credit: **Jake Osborne** (CC BY-NC-SA 2.0)

Common diving petrels (kuaka) are closely related to kuaka Whenua Hou and breed within the same dune system. As the name suggests, Common diving petrels are much more numerous than kuaka Whenua Hou and have a wider breeding distribution throughout the Southern Ocean.

Hybridisation between these two species has been observed, and one aim of the project is to assess genomic impacts of this hybridisation.

Future actions could include translocations of kuaka Whenua Hou to a new site to minimise hybridisation and create an “insurance policy” for the population. Understanding of the genetic diversity of the population can help select individuals for translocation, as well as help predict the impact of translocation on future genetic diversity of the species.

Genomics Aotearoa *HQG+PG* researchers from MWLR are working with DoC, Ngāi Tahu, and Papatipu Rūnaka (including Whenua Hou Komiti and Kaitiaki Rōpū) to establish baseline genomic data that can be used to inform conservation management.

The team’s draft reference genome for kuaka Whenua Hou is complemented by low coverage whole genome sequencing of 69 diving petrels, including both kuaka Whenua Hou and kuaka, to compare genetic diversity, and to assess potential impacts of hybridisation.

Preliminary population genomic analyses indicate differentiation between kuaka and Common diving petrels. Work is continuing to quantify genetic diversity and assess the extent of inbreeding.

Project researcher Natalie Forsdick said that we now have a depth of knowledge that we didn’t have before. “This basic biological understanding is the foundation for targeted management for the kuaka Whenua Hou, but at the same time it opens doors for us to use the expertise and processes more widely in conservation.”

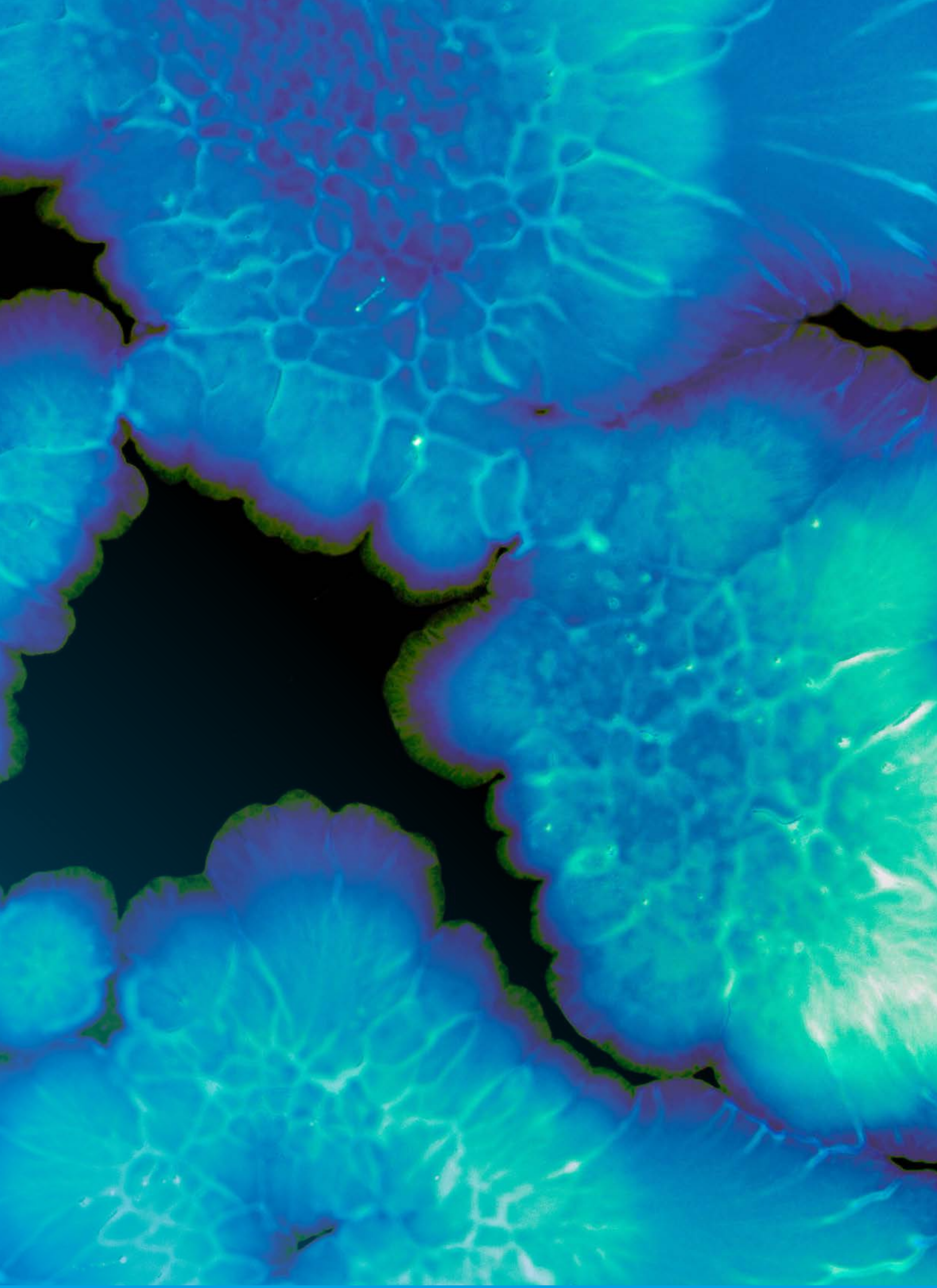
“The kuaka Whenua Hou genome was generated using PacBio HiFi and Dovetail Omni-C sequencing technologies, both of which are relatively new in the Aotearoa genomics space, so we are building capability with these technologies.”

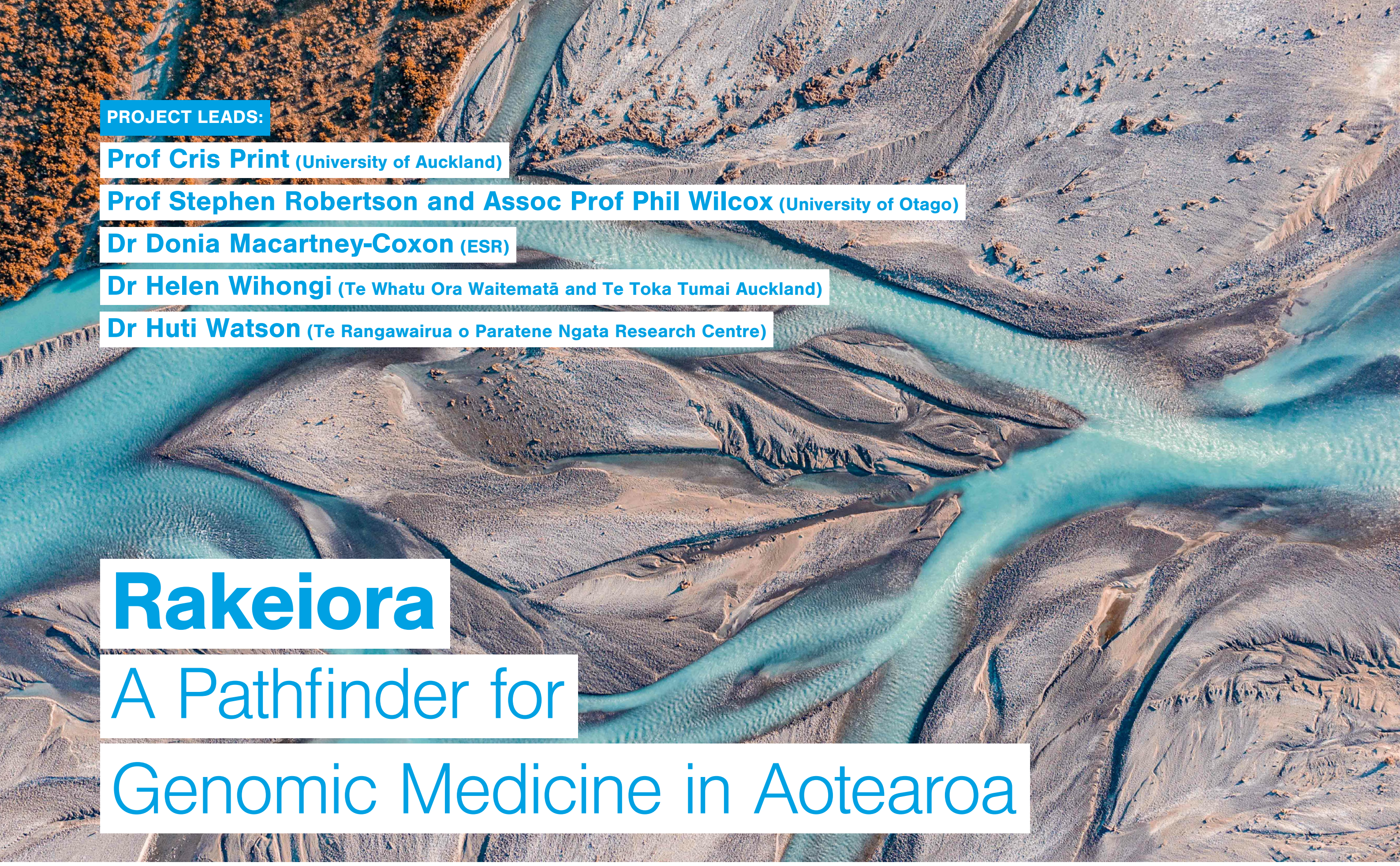
Health

This year saw the completion of the ground-breaking *Rakeiora* project, with a prototype demonstrating how precision health could work for Aotearoa.

The *Aotearoa Variome* project, which is complementary to, and essential to the realisation of the *Rakeiora* vision, was awarded additional Genomics Aotearoa funding to continue to work toward its goals.

The *Whole Genome Analysis* and *Streptococcus pyogenes* projects have continued to deliver outcomes, while highlighting some of the challenges facing health genomics research in Aotearoa.





PROJECT LEADS:

Prof Cris Print (University of Auckland)
Prof Stephen Robertson and Assoc Prof Phil Wilcox (University of Otago)
Dr Donia Macartney-Coxon (ESR)
Dr Helen Wihongi (Te Whatu Ora Waitematā and Te Toka Tumai Auckland)
Dr Huti Watson (Te Rangawairua o Paratene Ngata Research Centre)

Rakeiora

A Pathfinder for Genomic Medicine in Aotearoa

Rakeiora is Aotearoa's flagship project for precision medicine, aiming to transform Aotearoa's translational genomic health research infrastructure.

Historically we have had high quality but siloed projects with little potential for collaboration and inconsistent inclusion of Te Tiriti o Waitangi rights and Māori & Pacific health needs. We need an effective, safe, adaptable, and equitable Aotearoa-wide genomic research infrastructure. That has been *Rakeiora's* vision.

The project developed research themes that require further investigation, to scale up and implement genomically informed precision medicine as a clinical reality for all New Zealanders.

These research themes include:

1. The development of a national-scale informatics research environment and infrastructure
2. The exploration of regulatory and ethical underpinnings so that the legal basis for its implementation is secure
3. The development of conceptualisation of definitions of benefit in an economic sense from the establishment of this resource
4. The formalisation of kawa and tikanga so that all people undertaking genomics research and healthcare with Māori and Pacific People have a reference framework.

Aspects of the latter are embodied in a report by a rōpū formed during the early stages of the *Rakeiora* Project – Ira Tātai Whakaheke.

The major achievement this year has been the successful completion of the project prototype and the delivery of a comprehensive report to MBIE. The report described the project's adoption of agile adaptive research methods, co-design and creation of a research environment, a set of protocols informed by Te Tiriti-informed Principles, which enable efficient, safe, and powerful research projects to be performed for the benefit of all New Zealanders. This was all achieved in close and extensive collaboration with a team at NeSI led by Jun Huh and Claire Rye. A Proof of Principle pharmacogenetic exemplar was presented and was demonstrated to interested parties over a zoom seminar in March 2023.

The project has fostered considerable capability development in both computational infrastructure and in Te Ao Māori engagement to partner organisations NeSI and Cancer Trials New Zealand. The *Rakeiora* team has also contributed to *SING Aotearoa* along with numerous workshops, educational and scientific presentations at events this year, including invited presentations at international conferences and to international indigenous researchers.

Several bioinformatic pipelines have been deployed in the *Rakeiora* computational environment including genome sequence alignment and variant calling pipelines as well as workflows to demonstrate co-analysis of pharmacogenetic genotypes, prescription, and dispensing records and a tumour-normal variant calling pipeline for cancer patients.

Image Credit: **Amy Dowdle - Genetics Otago**

CASE STUDY:

Meet postdoctoral fellow Ben Halliday

Ben Halliday is a key member of the *Variome* and *Rakeiora* project teams. Ben developed an interest in bioinformatics and using genome sequencing to explore rare disease genetics during his BSc (Hons) project - identifying the genetic drivers of childhood kidney cancers.

His doctoral research at the University of Otago focused on the genomic investigation of patients with brain malformations, identifying disease-causing genetic changes and improving understanding of the mechanisms critical for brain development.

Now a postdoctoral fellow with Stephen Robertson, Ben's work focuses on assessing the appropriateness of genomic diagnostic tools in Aotearoa and investigating how to improve their performance for Māori and Pasifika populations.

Ben has transitioned from the student learning environment to participation in Genomics Aotearoa training, then to demonstrating in workshops, and now to data carpentry workshops and training people in bioinformatics. "I love the bioinformatics side of this, I can't see myself doing anything else."

A key aspect of the learning process for Ben is transferring skills back into the team.

"I enjoy the aspect of learning from the bioinformatics community, then developing and applying code within the lab. We have high level discussions about what we need, and I work alongside colleagues to introduce or refine the tools for the specific goal we are working to."

"It's an exciting space to be working in, with constant development in bioinformatics and genomic medicine. I'm working in projects that will make a difference, and I have a part to play in that - both in the research, and in supporting skills development so we can deliver."

Ben has always been interested in genetics, thanks to his mother who is a tree geneticist "I like that genomics has the potential to really make a difference in someone's life. Being able to pin down a key point in the genome to offer a diagnosis – improving patient care, allowing for prenatal testing, and helping patients and families understand the risk of recurrence within their whanau."

He has observed the surge in interest and respect for genomics since COVID-19 lockdowns, where people were introduced to genomic sequencing. "I want to see genetics and genomics even more widely understood and accepted, to help us deliver the benefits of genomic medicine."

Ben's story is a classic example of what Genomics Aotearoa wants to see from capability-building – the transition from a student to a skilled researcher passing their knowledge on to the next generation of researchers.

PROJECT LEADS:

Prof Stephen Robertson and Assoc Prof Phil Wilcox (University of Otago)

Aotearoa New Zealand Variome

Capturing Genomic Diversity for Disease Diagnosis

Variomes describe the unique genetic signatures of populations. Without a comprehensive variome for Aotearoa, shaped as it is by our unique human history, genomic tools based on overseas non-Māori populations have limited use in genomic medicine for the increasing number of people with Māori ancestry.

PROFESSOR STEPHEN ROBERTSON

Image Credit: **Royal Society Te Apārangi**

The *Variome* project catalogues genomic variation embedded in the genomes of New Zealanders with Māori ancestry, to improve disease diagnosis and to support healthcare research driven by Māori, for Māori. Genomic data sourced from elsewhere are not always informative for Māori. Sometimes variants associated with disease susceptibility in non-Māori populations have no such relationship in Māori, and sometimes there are variants associated with health conditions in Māori that are absent in non-Māori. The *Variome* project seeks to address this knowledge gap.

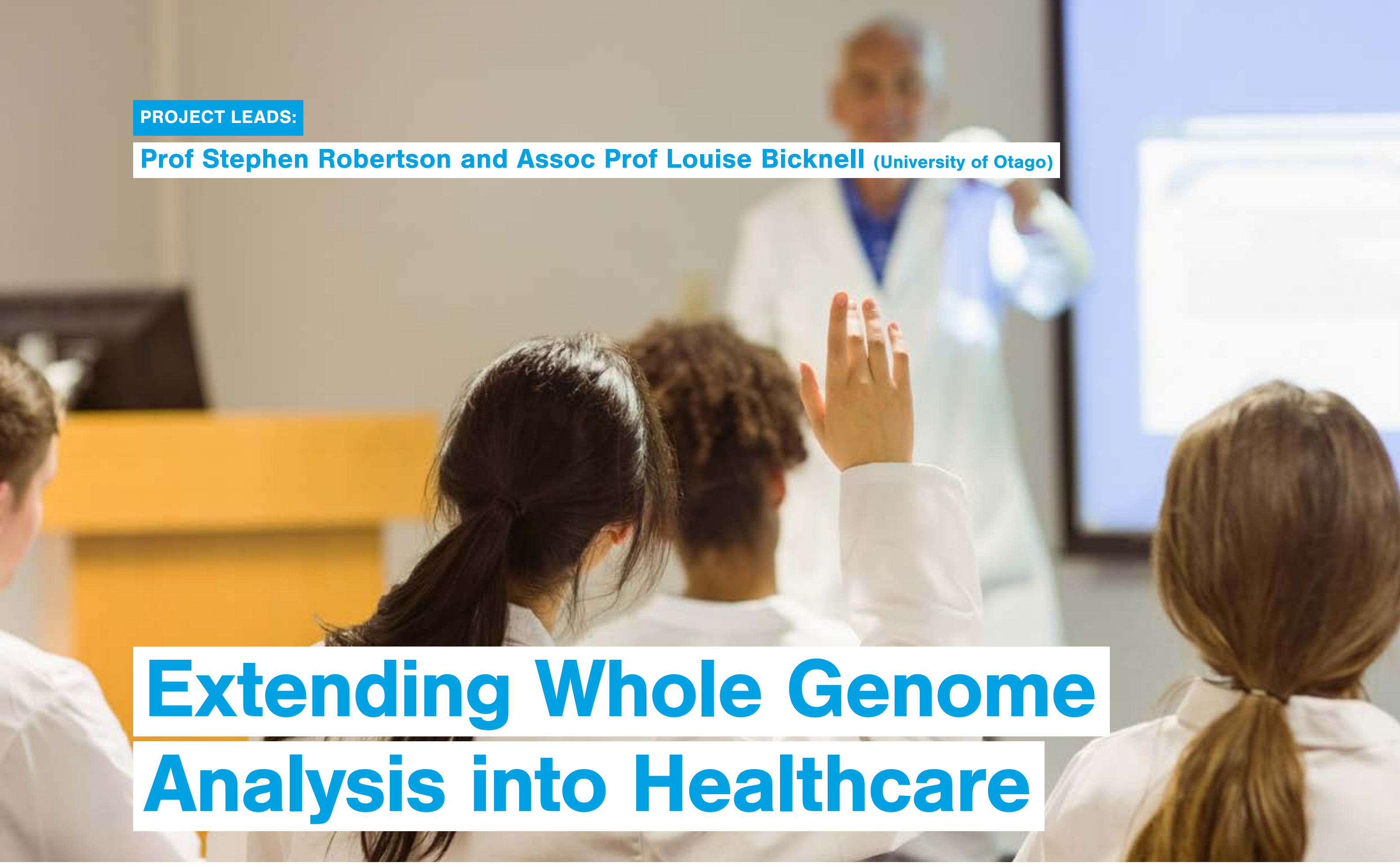
Several communities have engaged with the research and recruitment has continued, with 790 participants recruited to date and plans to recruit a further 200 before the end of August 2023. At the time of writing the project has sequenced 510 genomes, and 280 have been aligned and variant called. The project has developed a pipeline for variant calling in collaboration with Professor Wyeth Wasserman (University of British Columbia and Silent Genomes Canada) who is helping lead a similar Indigenous genomics project for his nation. This collaboration has also been harmonising analysis pipelines and visualisation tools via a secure portal. These collaborations have led to mutually beneficial tool sharing and exchanges.

In addition to the main project which is using short read sequencing as its primary tool, long read sequences are being produced for six individuals, and these have been combined with re-sequenced data to assemble telomere-to-telomere quality genomes in collaboration with the University of California, Santa Cruz (UCSC) which is also co-leading the Human Pangenome Reference Consortium (HPRC).

Establishment of close collaboration with UCSC has enhanced the *Variome* project's substantial international profile. An important aspect of this collaboration is that Māori DNA information is kept in Aotearoa consistent with participant consents. Information exchange includes genomic data generation by UCSD and training of NZ researchers in pangenome construction, for knowledge of inclusion of indigenous ethics and knowledge in STEM education.

The *Variome* project team's international reputation for best practice Indigenous co-led research continues to grow. This year the *Variome* project team contributed to the founding of the Federation of Indigenous Genomics at a meeting in Hawai'i in April with Indigenous scientists from the USA, Canada, Australia, and Aotearoa, as well as an invited presentation by Phil Wilcox at the recent International Congress on Genetics. Specific mention of the project was made in other presentations, including talks by Assistant Professor Karen Miga and former Genomics Aotearoa postdoctoral fellow Ann McCartney.

Genomics Aotearoa wishes to warmly congratulate project co-lead Stephen Robertson, who was awarded the prestigious Royal Society's Hercus Medal for research on genetic conditions impacting children and seeking to establish equitable delivery of genomic medicine for Māori.



PROJECT LEADS:

Prof Stephen Robertson and Assoc Prof Louise Bicknell (University of Otago)

Extending Whole Genome Analysis into Healthcare

Sequencing and analysing a person's entire genome – whole genome analysis (WGA) – is the most comprehensive method of arriving at a genetic diagnosis. In Aotearoa the capability to implement WGA in healthcare has yet to be fully realised, but this Genomics Aotearoa supported project is making significant progress.

The delivery of genome level medical diagnostics in Aotearoa is almost entirely dependent on overseas provisioned services. This project trials the generation, analysis, interpretation, and clinical delivery of genomic diagnostics by teams based in hospitals across Aotearoa.

In the last year a clinical team at Canterbury Health Labs in Christchurch has developed an exome sequencing clinical service in Aotearoa, (the exome is the protein-coding region of the genome which encodes most known disease-related variants), work that was directly leveraged from this Genomics Aotearoa funded project. Clinically accredited programmes are not simple to implement and are governed by strict criteria. This exome sequencing service is a first for the country, and a major milestone, as exome sequencing is considered a fundamental offering for precision medicine.

Under-resourced and understaffed former DHB labs have meant very little time or resource could be dedicated nationally to this project. However, considerable commitment from the team at Canterbury Health Laboratories led by Richard King and Cheng Yee Chan has meant that nearly 100 full genome datasets were analysed by a hospital-based diagnostic laboratory. Developing familiarity with these technologies has led to progress in building new capability in the diagnostic arena in Aotearoa.

The team has also been working on harmonisation of genomic analysis nationwide with the adoption of common tools, a shared analytical platform and analytical approaches and standards. A lack of platform cohesion has been cited by clinical diagnostics teams as a challenge to their work.

Recruitment of participants to this project by clinicians has been purposefully directed towards encouraging Māori to participate. As a result, 44% of recruited individuals self-identified as Māori. To date, 44 whānau have been enrolled into the project (41% Māori and Pasifika), 100 genomes have been sequenced, and 35 whānau datasets (77 genome datasets) have been analysed.

Of the 35 whānau whose genomes have been analysed to completion, a certain diagnosis was made in 12 (34%) and a likely diagnosis in a further three cases. This equates to a current diagnostic rate of 43% which compares favourably with international rates of diagnosis.

All whānau and individuals with either a confirmed or unconfirmed diagnosis were referred to Rare Disorders New Zealand (RDNZ) by their physician and one family had contacted RDNZ at the time of writing.

The project also supported two genomics Wananga to provide training in analysis of whole genome data for identifying causative variants. These were well attended by students (one at *SING Aotearoa*, another at Otago Medical School for Māori medical students).

PROJECT LEADS:

Assoc Prof Paul Gardner (University of Otago)

Dr Joep de Ligt (ESR)

Identifying the Drivers of *Streptococcus pyogenes* to Better Understand Disease Causation

Globally, there are more than 600 million Group A *Streptococcus* (GAS; *Streptococcus pyogenes*) infections occur each year. Māori and Pasifika are disproportionately affected by GAS, including many children. Although GAS infection can result in a range of clinical outcomes, from minor sore throats and skin infections to rheumatic fever and rheumatic heart disease, we still do not fully understand the genetic drivers associated with different outcomes.

TE REO

STOP SORE THROATS HURTING HEARTS

Preventing Rheumatic Fever

Ngā āhuatanga o te kirikā rūmātiki

Tamariki mā, kōrero ki ō mātua, ki tētahi pakeke rānei mehemea kei te mamae tō korokoro

Mātua mā, kia mataara ki ngā tohu o te
- uaua ki te horomi
- uaua ki te kai me te inu

Korokoro mamae

Kua hihiratia ngā korokoro mamae

Haere tōtika ki te rata, ki te nēhi rānei kia hihiratia ngā korokoro mamae katoa.

Ko te korokoro mamae he 'strep korokoro' (ka take mai i te mate huakita ko Group A Streptococcus te ingoa).

Kua whāngaia te 'strep korokoro' ki te rongoā paturopi – kainga te rongoā mō ngā rā 10 **He mea nui kia kainga ngā rongoā kia pau rawa.**

Ka aukatitia te 'strep korokoro' me te rūmātiki kirikā.

Manawa ora, manawa kaha – Hauora, kakama me te manawa ora

Ngā korokoro mamae kāore e hihiratia

Kāore i hihiratia tō korokoro mamae e te rata e te nēhi rānei, ā, ka noho māuiui.

Tērā pea ko te korokoro mamae he 'strep korokoro'.

'Strep korokoro' kāore e whāngaia ki te rongoā (rongoā paturopi).

Tērā pea ka tipu ake te rūmātiki kirikā.

Ka tino māuiui i te rūmātiki kirikā – Ka ngenge, kua mamae, kua pupuhi hoki ngā whatīanga (ngā pona, ngā tuke, ngā whatīanga raparapa me ngā kawititanga o te ringaringa).

Ka māuiui, ka ngenge hoki, ā, kua raru te manawa – mate manawa rūmātiki

ENGLISH

STOP SORE THROATS HURTING HEARTS

Preventing Rheumatic Fever

Steps of rheumatic fever

Kids tell your parents or an adult if you have a sore throat

Adults look for the signs of a sore throat
- hard to swallow
- trouble eating or drinking

Sore throat

Sore throat checked

Go to doctor or nurse straight away to get every sore throat checked.

Sore throat is 'strep throat' (caused by bacterial infection or bug called Group A Streptococcus).

'Strep throat' is treated - take antibiotics for 10 days. It's important to finish all the antibiotics.

Stops 'strep throat' and rheumatic fever.

Healthy and active with strong heart

Sore throats not checked

Don't go to the doctor or nurse to get sore throat checked, stay unwell.

Sore throat could be 'strep throat'.

'Strep throat' not treated with medicine (antibiotics).

Rheumatic fever could develop.

Get very sick from rheumatic fever - tiredness, sore and swollen joints (knees, elbows, ankles and wrists).

Unwell and tired with serious heart problems – rheumatic heart disease

Image Credit: **Health Promotion Agency - Rheumatic Fever Awareness Campaign**

In bacterial infections, the relationships between genotype (genetic causes) and phenotype (invasiveness or resistance to antimicrobial agents) are difficult to predict from genome sequences.

To better understand the mechanisms underlying the range of GAS outcomes, the project aims to:

1. Gain insights into the genetic diversity and population structure of invasive and non-invasive GAS circulating amongst children in Aotearoa and
2. Identify genetic drivers responsible for invasive phenotypes (genotype-phenotype prediction). Community engagement is a major component of the project and critical to its success.

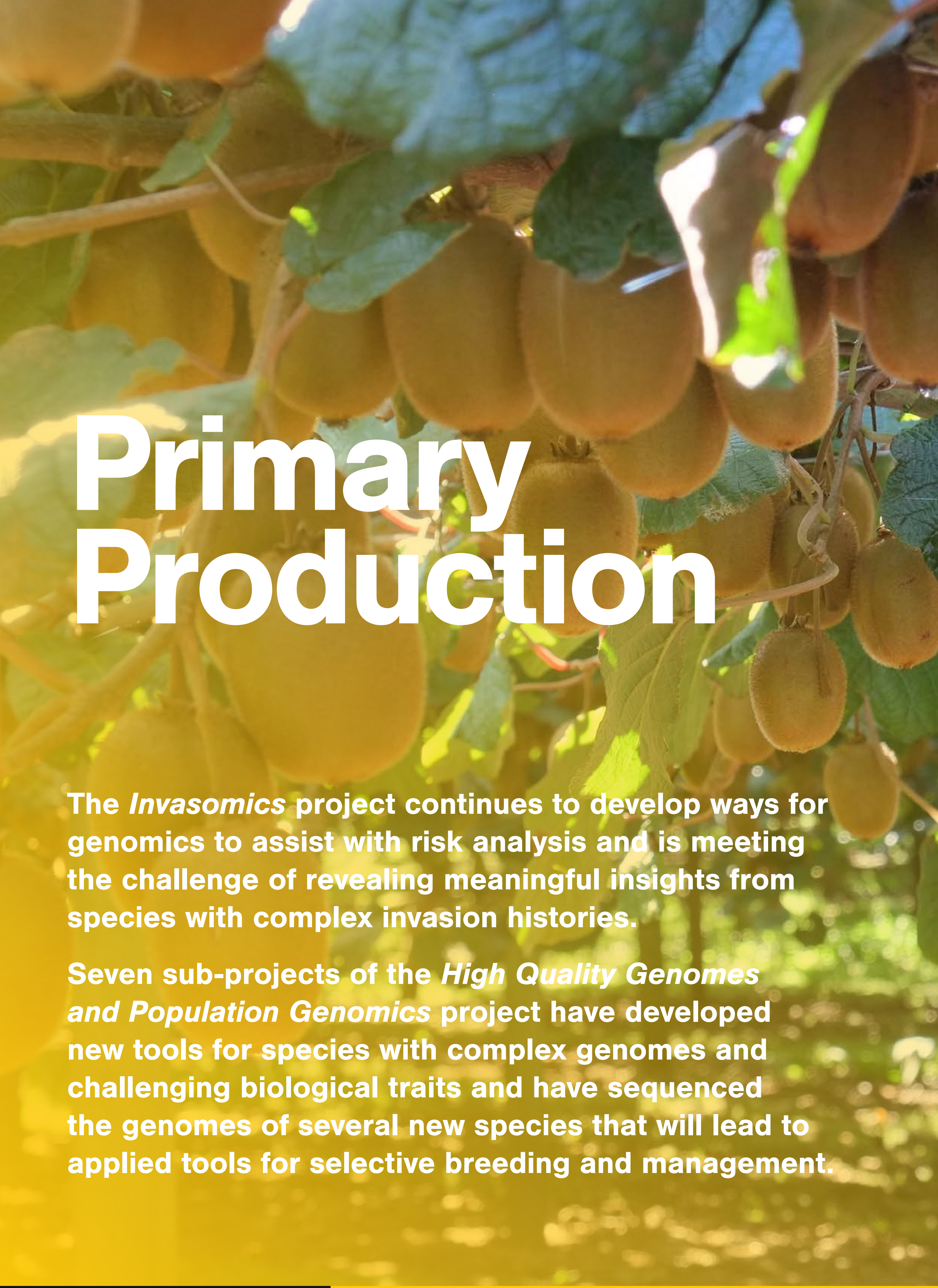
Community engagement led by the National Hauora Coalition (NHC) has raised awareness about GAS infections in our communities and initiated discussions on what researchers can do to adapt their research strategies. Insights from Māori, Tongan, Samoan and Cook Island Māori communities, including youth, have identified three key recommendations covering informed consent, culturally informed guidelines and ensuring Indigenous sovereignty over isolates.

This year, the project has resulted in a deeper understanding of the genetics of *Streptococcus pyogenes*, with the sequencing of 1,968 isolates (500 invasive strains from the ESR collection) and 1,468 non-invasive community isolates (as controls). The resulting population genomics study involved screening genetic markers of antimicrobial resistance, identifying potential virulence genes, and screening variation of target genes for vaccine research. The data generated are being used in the national Manatū Hauora funded vaccination research led by the University of Auckland, which is conducting clinical trials.

In addition, the research identified isolates belonging to the different *emm* types (a gene encoding a cell-surface protein that is commonly used to classify *S. pyogenes* isolates) and used these for grouping of genes/variants that are indicative of invasiveness.

Postdoctoral fellow Christina Straub was awarded a prestigious Marie Curie Fellowship and will sadly be moving offshore to start a new project in September 2023. Christina's capability-building activities during the project have included supervision of a summer student and a Master's student, and delivering bioinformatics training to other team members at ESR. Meanwhile assistant research fellow Te Whetu Kerekere accepted a further research assistant opportunity with Ngāti Porou Hauora in Tairāwhiti involving metabolic variants that builds on her bioinformatic and genomic skills.

Completion of this highly successful project is scheduled for December 2023, but several new initiatives will build on this foundational work.



Primary Production

The *Invasomics* project continues to develop ways for genomics to assist with risk analysis and is meeting the challenge of revealing meaningful insights from species with complex invasion histories.

Seven sub-projects of the *High Quality Genomes and Population Genomics* project have developed new tools for species with complex genomes and challenging biological traits and have sequenced the genomes of several new species that will lead to applied tools for selective breeding and management.

PROJECT LEADS:

Dr Manpreet Dhami (Manaaki Whenua – Landcare Research)

Dr Ang McGaughran (University of Waikato)

Invasomics

for Better Biosecurity and Invasive Species Management

Brown Marmorated Stink Bug (*Halyomorpha halys*)

Invasive species threaten native biodiversity, primary industries, and health. Ever-increasing international trade, mobility, and the impacts of climate change make Aotearoa highly vulnerable to newly arriving pests and diseases. Our unique ecosystems add a layer of complexity to our responses to these threats. Innovative and customised solutions that minimise the risk of new pests arriving are therefore urgently needed by policymakers and practitioners.

The *Invasomics* project focuses on identifying and validating measurable genetic characteristics that indicate inherent invasiveness. Using biological models, the team aims to identify the genomic features that underlie successful biological invasions and use these features to predict the invasive potential of future invaders posing a high risk to Aotearoa New Zealand.

The project is continuing to develop a comprehensive database for invasive and non-invasive populations and species that represents a resource for the *invasomics* (invasion genomics) community. In addition, the team is producing an effective bioinformatics pipeline to generate high-quality data for key priority species of strategic importance to Aotearoa.

This year the team published findings on the brown marmorated stink bug (BMSB), showing how its complex invasion history can challenge accurate assignment of new incursions to their source populations. A further publication on the shortcomings of data stewardship in invasion biology, and how these limit the value and secondary use of genomic data has just been accepted by *Molecular Ecology Resources.*

The team's extensive assessment of globally available datasets has revealed key barriers that prevent final development of a generalisable model framework (which the team aimed to develop as the first step towards a tool to assist prioritisation of new/other invasive species by end-users, such as MPI). However, identifying these barriers is an important advance that will underpin future grant applications and associated research.

The project has also contributed to the career development of two postdoctoral fellows, Ellie Parvizi and Amy Vaughn, both of whom are moving to other roles within the Aotearoa genomics research community.

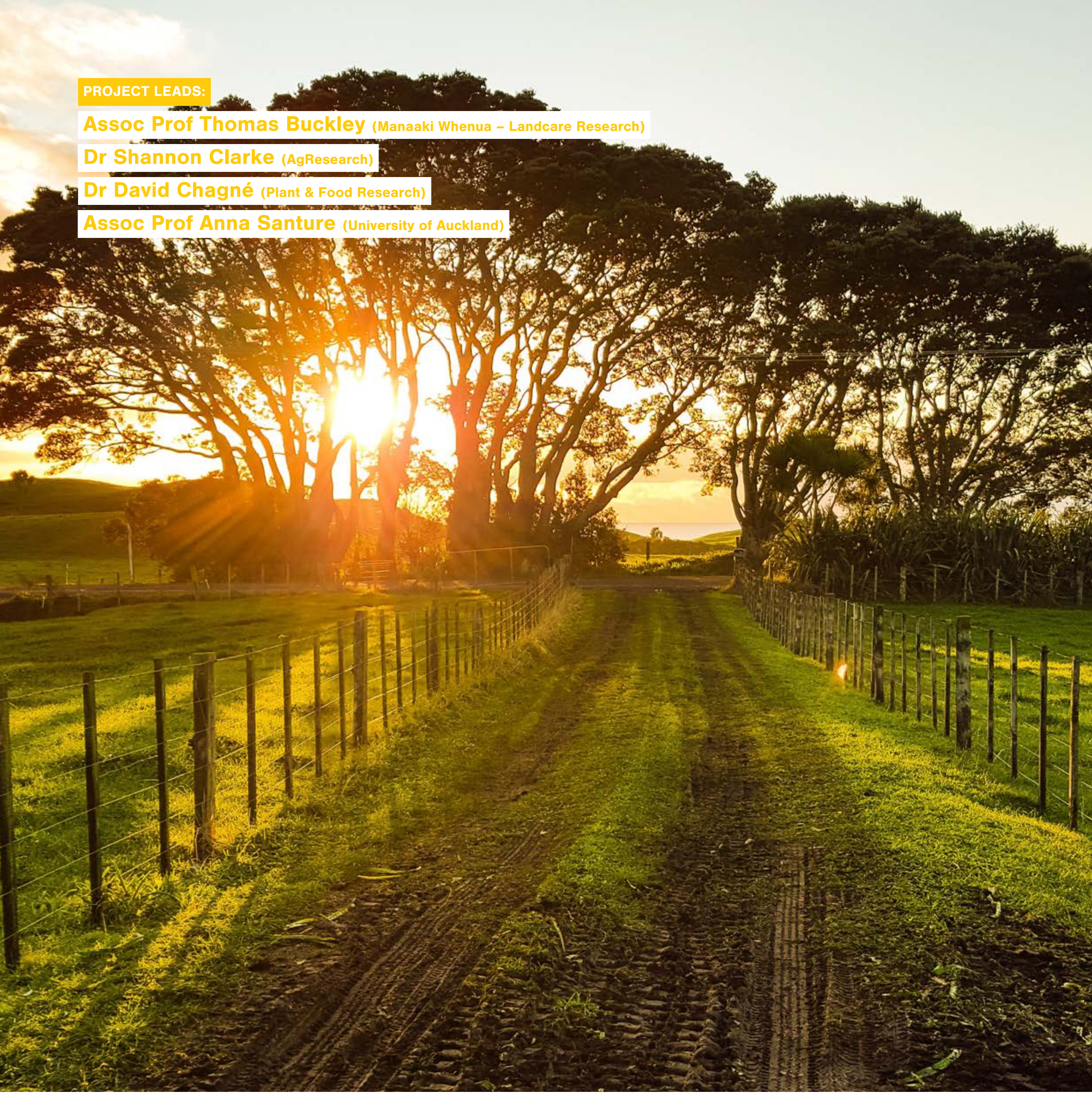
Brown Marmorated Stink Bug eggs (*Halyomorpha halys*)

The project co-leads organised the inaugural Society for Molecular Biology and Evolution (SMBE) Invasomics meeting in November 2022 and devoted a workshop to addressing our current and potential future use of genomic tools in the study of invasive species (from biosecurity surveillance to understanding the evolutionary underpinnings of invasion success). Based on this workshop, the participants are collaborating on a review to be submitted to *Genome Biology and Evolution* later in the year.

Knowledge-sharing across the team has also included presentations and a symposium by several team members at four conferences, and three new bioinformatic pipelines made publicly available on GitHub.

LAIJIAO LAN (INRAE FRANCE), AMY VAUGHAN, ANG MCGAUGHRAN, MANPREET DHAMI, ELLIE PARVIZI, AND MEERAN HUSSAIN (WAIKATO)

Invasomics team and collaborators at the International Congress of Biological Invasions, Christchurch Town Hall, May 2023



PROJECT LEADS:

Assoc Prof Thomas Buckley (Manaaki Whenua – Landcare Research)
Dr Shannon Clarke (AgResearch)
Dr David Chagné (Plant & Food Research)
Assoc Prof Anna Santure (University of Auckland)

High Quality Genomes and Population Genomics

Primary production end-users can use *High Quality Genomes and Population Genomics* tools to better predict biological traits such as the reproduction, appearance, behaviour, or disease susceptibility of a variant, and how these traits differ among populations. Understanding this variation is used to improve economic yields and increase resilience to environmental change.

SUB-PROJECT LEADS:

Dr David Chagné and Dr Chen Wu (Plant & Food Research)

Blueberry (*Vaccinium sect. Cyanococcus*)

Blueberry Polyploid Genome Assembly

Plant & Food Research work with Turners & Growers, who represent blueberry growers to make sure the research is fit for real-world impact on their business, and blueberry breeders who need genomics solutions to develop new cultivars more efficiently.

The new genome assembly of tetraploid blueberry used a combination of high accuracy long read DNA sequencing technology combined with new bioinformatics methods, leveraging a parents-offspring relationship (the trio-binning method). The high quality genome assembly of tetraploid blueberry built by Chen Wu and Ignacio Carvajal (Plant & Food Research) resolved the four sets of chromosomes for this autotetraploid species and enabled Sara Montanari (Plant & Food Research) to identify a candidate gene likely causing the high antioxidant potential of blueberry.

This new method for polyploid assembly is also used for kiwifruit genome analysis at Plant & Food Research.

SUB-PROJECT LEAD:

Assoc Prof Maren Wellenreuther **(Plant & Food Research)**

Finfish Genomics

Improved hoki and Chinook salmon genomes

Chinook Salmon (*Oncorhynchus tshawytscha*)

The genomics field is constantly moving, and resources that were state-of-the-art only a few years ago are already outdated. This sub-project developed improved genomes for hoki and Chinook salmon for the fisheries and aquaculture industries, respectively. High-quality genomes for both species will enable us to study the genetic control of adaptation to warming temperatures, and to elucidate the genes underlying important production traits.

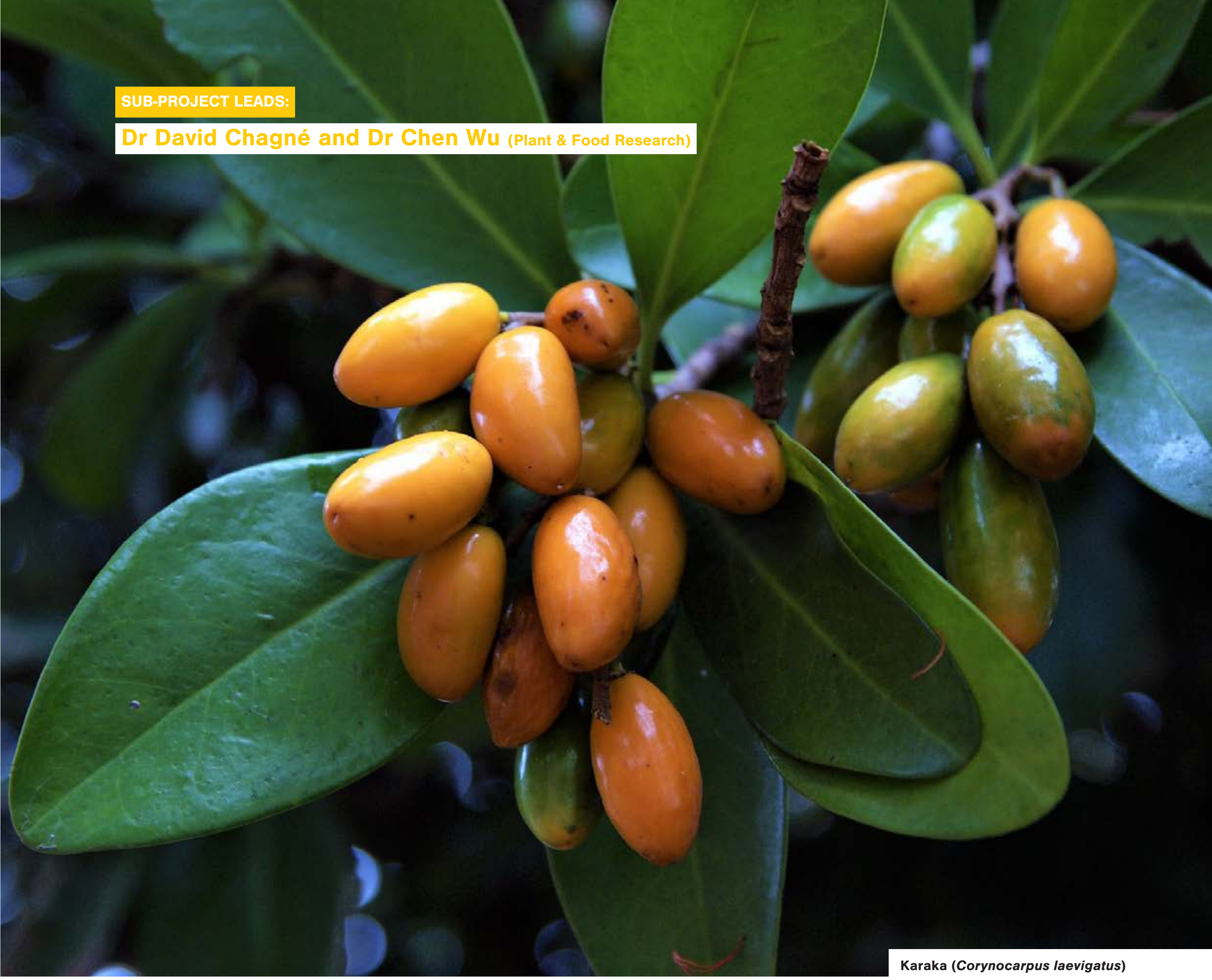
The fisheries industry (DeepWater Group and Te Ohu Kaimoana) and the aquaculture industry (NZ King Salmon, Sanford, and Akaroa Salmon - Mount Cook) are the main end-users of this research.

Te Ohu Kaimoana is involved in consultations about how to manage genomic fisheries data, using the hoki genome as a model. A new data life cycle concept has been developed with Te Ohu Kaimoana to inform decisions along the entire life cycle of data, from project inception, through to sample collection, data analysis, publication, and re-use.

This year, long read sequencing data has been produced using the PacBio hi-fidelity technology for hoki and Chinook salmon. Draft assemblies were produced, and regions linked to the sex determining locus of Chinook salmon were resolved.

SUB-PROJECT LEADS:

Dr David Chagné and Dr Chen Wu (Plant & Food Research)



Karaka (*Corynocarpus laevigatus*)

Developing a High-Quality Genome for Karaka

Karaka is an iconic taonga tree that was an important food source for early Māori settlers, planted at traditional fishing sites as a seasonal resource. But the plant could have commercial potential.

Building a genome for karaka evolved from discussions between Plant & Food Research and Rangitāne o Manawatu around their aspirations to realise the commercial potential of one of their taonga.

A high-quality reference genome of karaka will enable Rangitāne o Manawatu to understand the genetic diversity of karaka trees growing in their rohe, as well as understanding the genetic control of key traits such as toxin production in the fruit and nut, and sex determinism.

To date, a highly complete genome and transcriptome assembly of karaka has been produced. The transcriptome of karaka was produced from RNA extracted from 19 tissue types and enabled researchers to predict approximately 31,000 gene-coding sequences in the karaka genome.

This sub-project has contributed to obtaining a MBIE Vision Mātauranga Capability Fund project led by Rangitāne o Manawatu, with Plant & Food Research and Massey University involved, to understand the phenotypic composition of the toxin in the karaka fruit, which needs to be specially treated before consumption.

SUB-PROJECT LEADS:

Assoc Prof Maren Wellenreuther and Dr David Chagné (Plant & Food Research)

Mānuka flowers (*Leptospermum scoparium*)

The Mānuka Microbiome

Mānuka is a taonga species and a high value production crop. However, nothing is known of the role of the plant's microbiome.

Using metagenomics, this project carried out by Amali Thrimawithana (Plant & Food Research and University of Auckland PhD student) and involving Kim Handley (University of Auckland) investigates the mānuka microbiome across the motu. New this year, the research will contribute to understanding which types of microbes live in association with mānuka and the role these organisms play, especially around adaptation to environmental conditions.

The end-users for this research are Māori landowners who have mānuka growing on their land. The project team has engaged with iwi groups from rohe in Northland, East Cape, Manawatu, and the South Island about accessing samples of mānuka. Māori landowners include representatives of from Ngāti Porou, Te Arawa, Te Rarawa, Ngāi Tahu and Rangitāne o Manawatu. Informed consent has been given for sampling and DNA and RNA sequencing.

SUB-PROJECT LEAD:

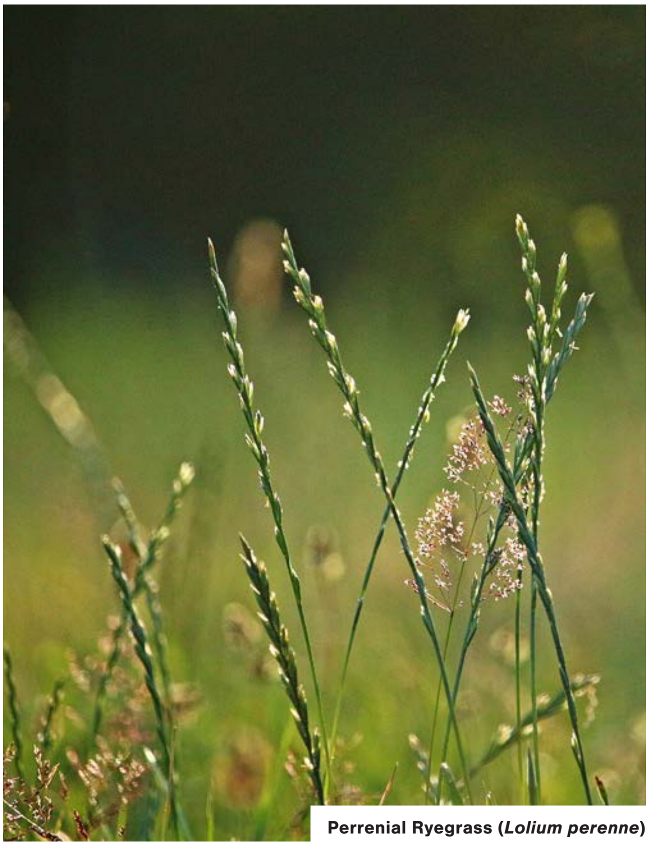
Dr Jeanne Jacobs (AgResearch)

Developing Methods and Tools
for improving reference genomes for ryegrass

Ryegrass is an important animal fodder in Aotearoa, and high-quality ryegrass meets the nutritional requirements for dairy cows, as well as grazing cattle. Ensuring quality fodder is important to farmers, and to the organisations that breed fodder grasses.

This year, in collaboration with researchers at University of Otago, a draft genome assembly was used for a detailed analysis of the self-incompatibility loci in ryegrass. RNAseq analysis on three tissue-types (leaf, root, flowerheads) and eight lines of ryegrass (two parents and six F1 progeny) improved the annotation of the draft genome.

This sub-project lead received a *PromethION Development Grant* from the Genomics Aotearoa associate Bragato Research Institute to develop direct RNA sequencing of ryegrass using nanopore sequencing. The total RNA used is anticipated to give a more complete picture of the RNA landscape, including mRNA isoforms as well as other RNA molecules. An initial trial run on a MinION sequencer showed promising results.

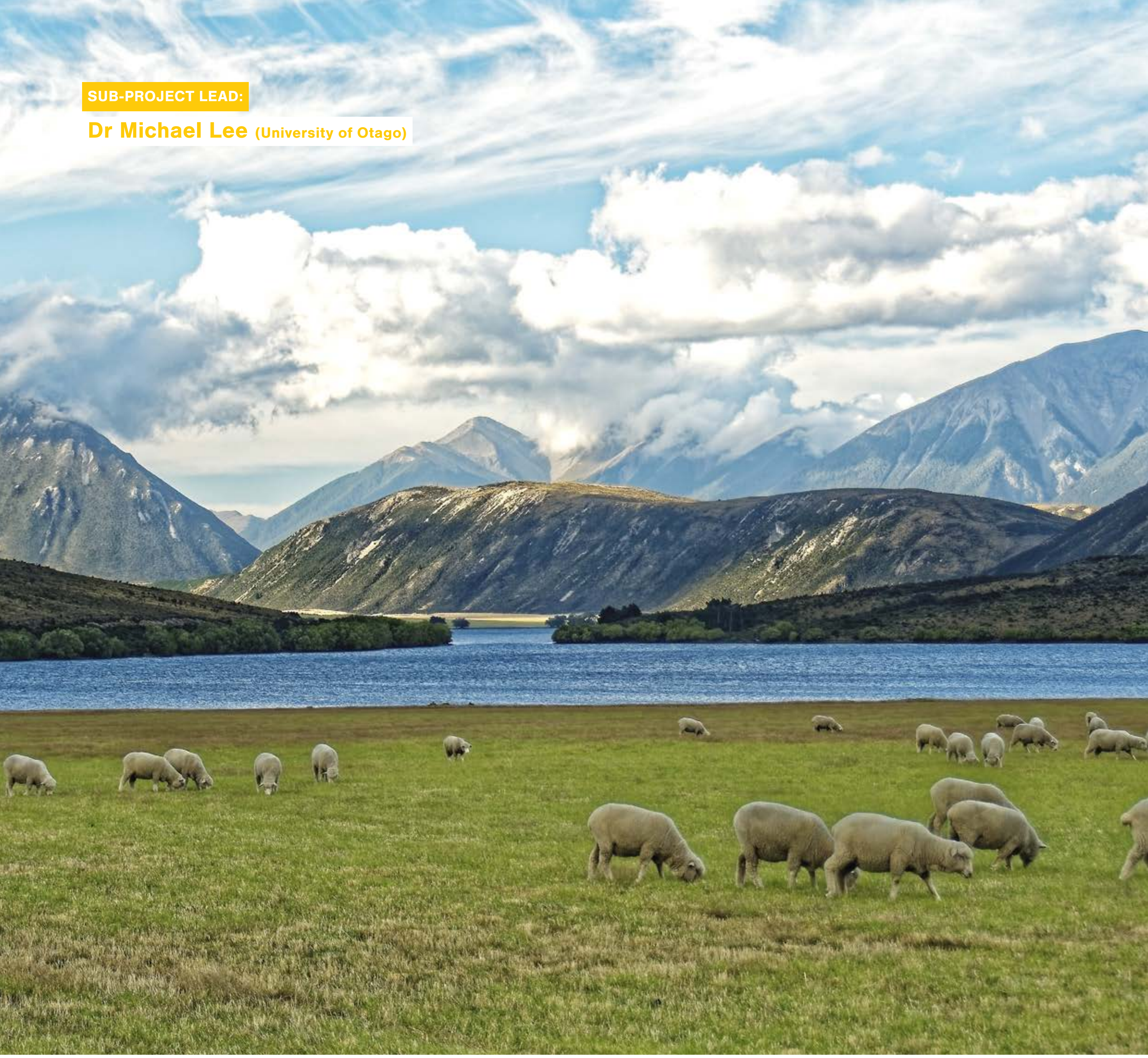


Perrenial Ryegrass (*Lolium perenne*)

Sheep Image Credit: **Bernard Spragg**

SUB-PROJECT LEAD:

Dr Michael Lee (University of Otago)

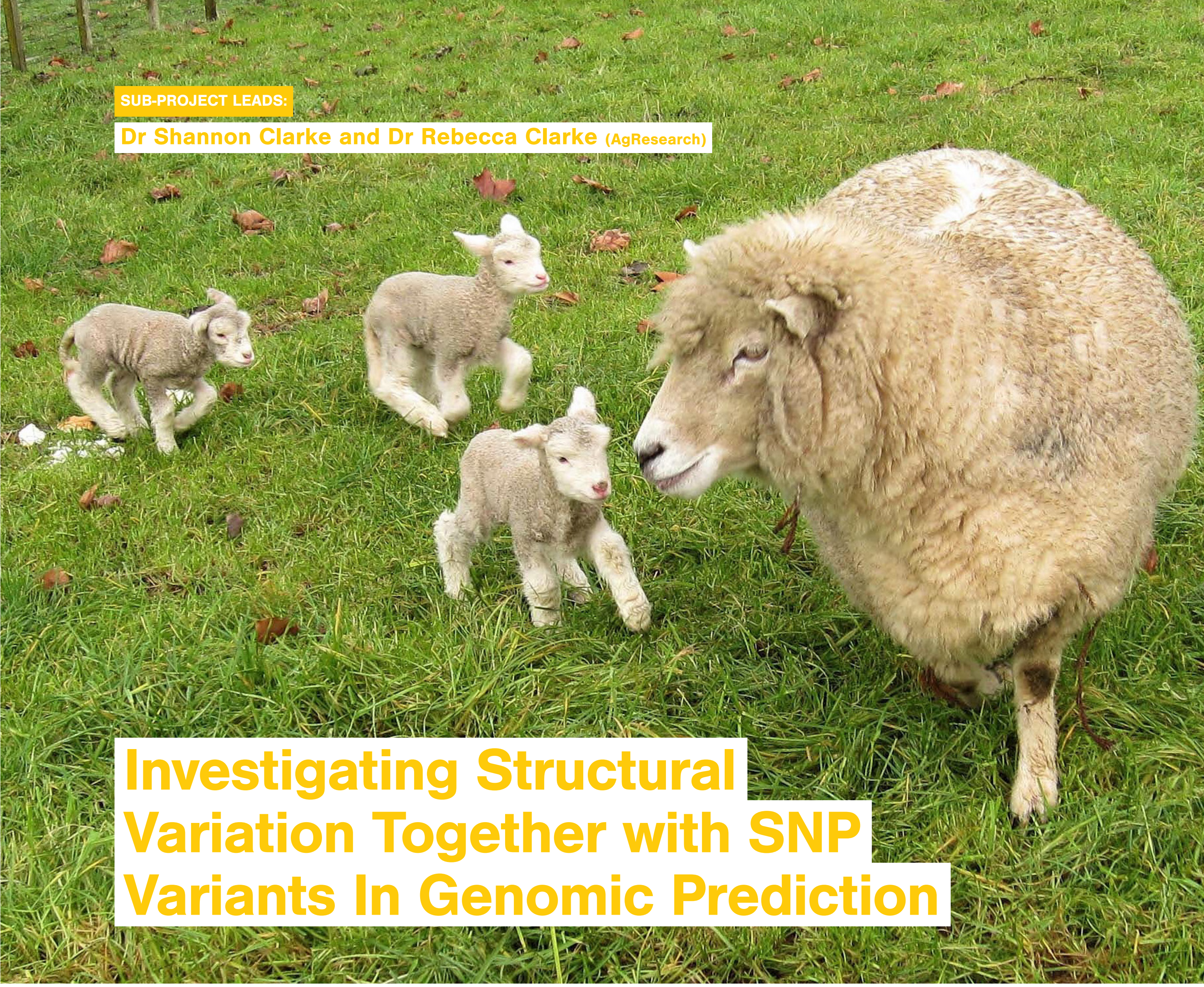


Characterising Inter-Chromosomal Linkage Disequilibria

with high density SNP chip data

Primary producers require a better understanding of genomic data to ultimately use these data to better predict genetic merit and increase the rate of genetic improvement. Beef + Lamb NZ Genetics is a key collaborator on this sub-project, using sheep as an exemplar.

To date this sub-project has processed high density SNP chip data (600K chip) from >23,000 animals and developed the analysis methods to allow linkage disequilibrium (epistasis) to be calculated from each pair of SNPs (that is, after quality checking about 250 billion pairwise comparisons). The next step is to optimise a panel of single nucleotide polymorphisms for genomic prediction in Aotearoa New Zealand sheep.



SUB-PROJECT LEADS:
Dr Shannon Clarke and Dr Rebecca Clarke (AgResearch)

Investigating Structural Variation Together with SNP Variants In Genomic Prediction

The national and international sheep research communities and sheep breeding entities need new genotyping tools for their industry genomic pipelines to improve genomic prediction.

This sub-project uses existing sheep genomic resources to investigate methods for structural variation detection and incorporation into genomic prediction. Genomic resources from pangenome assembly, copy number variation inclusion on SNP array platforms and long read adaptive sequencing have been integrated. Furthermore, SNP variation from thousands of sheep resequencing data sets has generated millions of variants for annotation on the sheep pangenome.

Long read sequencing and subsequent assembly of six Aotearoa New Zealand sheep breeds is underway (The Ovine Pangenome Project). These assemblies will contribute to the *Genome Graphs* project, which will capture structural variation between breeds with diverse phenotypes. Furthermore, variant calling from > 3500 whole genome short read sequencing sheep data sets has been incorporated on the functionally annotated sheep reference genome. Refinement of adaptive sequencing with Oxford Nanopore Technology (ONT) for validation of 59 regions of interest across the genome simultaneously has seen an increase in multiplexing level with the GridION and PromethION.

Rachael Ashby (the sub-project's postdoctoral fellow) has helped with Genomics Aotearoa training on basic R skills, and a workshop on GitHub best practise, among other efforts toward capability building.

Thanks in part to her role as a science lead for the *HQG+PG* project as well as the Science leadership team, Shannon was promoted to a Principal Scientist at AgResearch. Genomics Aotearoa is delighted to have been able to contribute to Shannon's career progression.

Members of the team presented at the Advances in Genome Biology and Technology conference in Texas in March 2023, both on travel scholarships. Postdoctoral researcher Rebecca Clarke was invited to present her work on adaptive sampling, and Shannon and other team members presented at four other events. One publication has resulted from the work.

Image Credit: **Pam Kiwinz** (CC BY 2.0)

Te Ao Māori

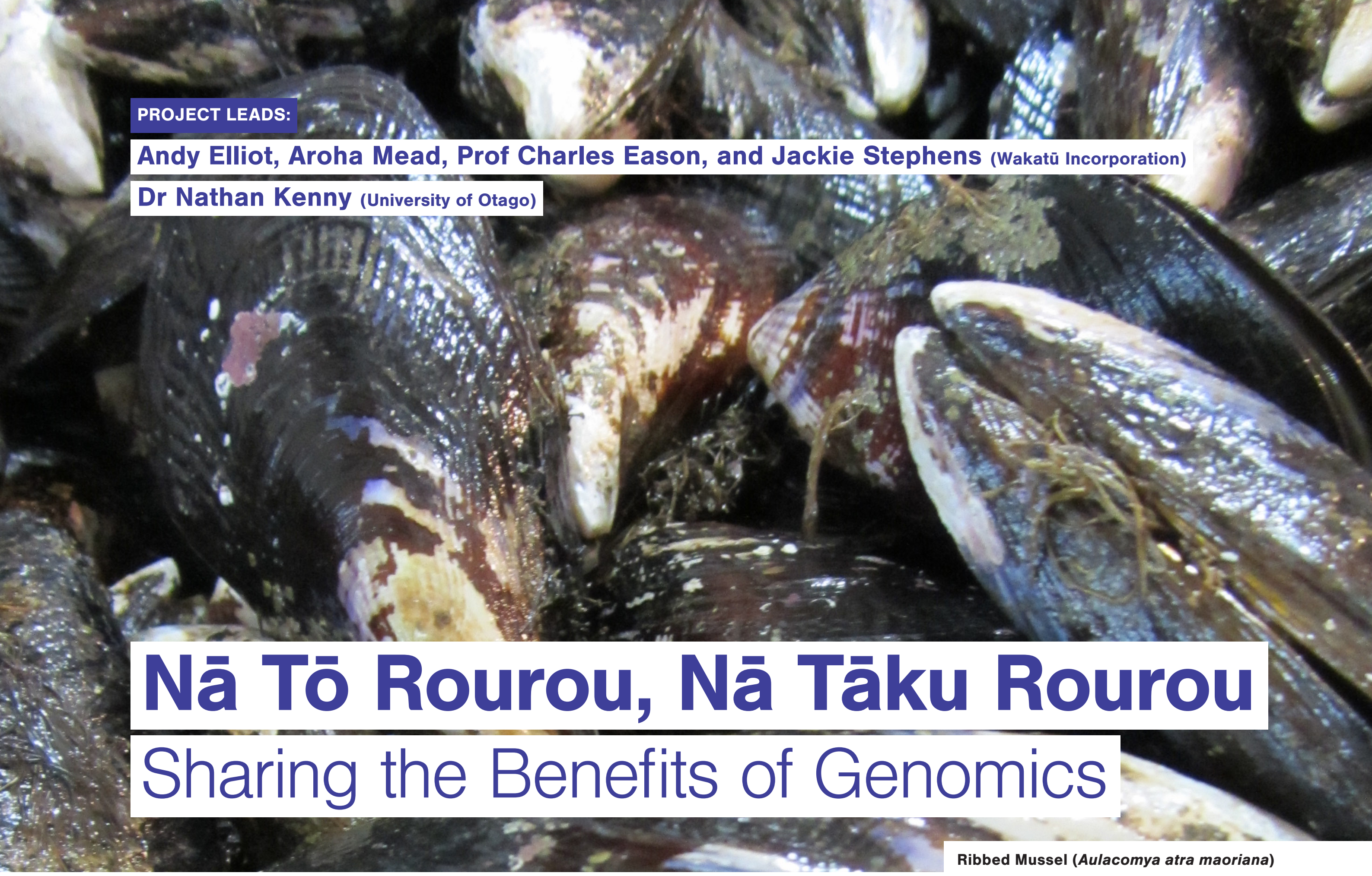
Progress and achievements acknowledged this year include six projects supported by the Indigenous Genomics Platform, together with the ongoing *Aotearoa Genomic Data Repository*, *Ruatau: Connecting Māori genomic scientists and communities*, and support for *SING Aotearoa: Mentoring emerging Māori genomics researchers*.

Indigenous Genomics Platform

Bringing Co-Leadership of Genomics Research to Life

The IGP increases opportunities for Māori groups, including hapori Māori, to lead projects and engage directly with genomics researchers, and for our research to better reflect Māori end-user needs.

Now in its second year, the IGP platform continues to grow. Three new IGP projects were awarded in this past year and four new projects have been funded for the coming year (2023-2024). The growing interest in the IGP, despite it being a relatively recent establishment, is testament to the need for, and value of, an approach that places those who benefit from the research in leading roles.



PROJECT LEADS:

Andy Elliot, Aroha Mead, Prof Charles Eason, and Jackie Stephens **(Wakatū Incorporation)**

Dr Nathan Kenny **(University of Otago)**

Nā Tō Rourou, Nā Tāku Rourou

Sharing the Benefits of Genomics

Ribbed Mussel (*Aulacomya atra maoriana*)

Nā Tō Rourou, Nā Tāku Rourou aims to better understand the genomic basis of productivity traits of high-value marine species, and ensure the ownership of this research is assigned to the right individuals, communities, or institutions according to established access, utilisation, and benefit-sharing (ABS) principles.

Many initiatives propose to sequence the genomes of species. In some cases, these initiatives are not sensitive to the Wai 262 Te Tiriti o Waitangi claim or United Nations Convention on Biological Diversity and Nagoya Protocol implications. Nā tō rourou, nā tāku rourou establishes a model for collaboration among Māori organisations and research institutions, with ownership and governance of genomic data by Māori, using exemplar datasets to test the model.

Wakatū Incorporation's strategy of cataloguing, characterising, and developing a deep understanding of the endemic species of Te Tauihu is the perfect vehicle for testing this Māori-centred model.

The project focuses on how the commercial benefits of culturally responsible genomic work can be vested long-term in the kaitiaki of the original sample. In an increasingly globalised world, where genomic sequencing is routine, kaitiaki run the risk of losing control of the ability to protect the genetic resources, data, and traditional knowledge associated with their taonga.

The project is sequencing the genomes of kopakopa (the ribbed mussel *Aulacomya atra maoriana*) and karengo (a seaweed within *Porphyra sp.*), marine species of significant interest to Wakatū and the iwi of Te Tauihu. Genomes of previously identified individuals with traits related to growth rate, adult condition and divergent phenotype will then be re-sequenced. The project will generate high density linkage maps, relationship matrices, gene predictions, and other useful resources for breeding and production pipelines.

The project will prove how genomic information can be gathered, used, and safeguarded by Indigenous organisations to drive forward their aspirations. The outputs will extend on Wakatū's existing benefit sharing programmes.

The project is still in the initiation stages, including recruitment, training, and initial experimentation. Team members have been involved in several communications events, and assistant research fellow Marc Bailie is a keen contributor to the Genomics Aotearoa training programme.

The first genomic sequences are being assembled and those assemblies optimised. Other work continues as scheduled, for delivery in the second year of the project.

Despite still being in the initial stages, the project has stimulated an additional partnership between Otago and Wakatū, focused on kawakawa diversity. That in turn has led to opportunities to apply for other external funding, with the aim of growing collaborative research in this field.

Image Credit: **Rodrigo Fernández** (CC by-SA 4.0)

PROJECT LEADS:

Chris Insley (Te Arawa Fisheries, Smart Māori Aquaculture)
Assoc Prof Peter Ritchie (Te Herenga Waka – Victoria University of Wellington)
Assoc Prof Maren Wellenreuther and Dr David Chagné (Plant & Food Research)

Development of Genomic Resources for the Selective Breeding of Haku

Kingfish (*Seriola lalandi*)

Globally and nationally, the demand for seafood is growing. Aquaculture programmes in Aotearoa rely almost exclusively on the farming of three species: kuku or kūtai (greenshell mussels), tio (Pacific oysters), and Chinook salmon. This highlights a need to increase resilience of the sector through diversification.

Haku (kingfish) are an important traditional food for Māori, but no quantitative data on Māori customary non-commercial catch are available. Because of the coastal distribution of the species and its inclination to strike lures, it is likely that historically Māori caught considerable numbers of haku. The project is designed to lay the groundwork for the aquaculture of haku (specifically warehenga or Yellowtail kingfish).

This year has involved further developing the relationships underpinning the project. The co-leads have met to discuss an intellectual property plan and a data management plan, and have developed a sampling protocol using pre-prepared sampling kits that have been distributed to local fishers. So far around 50 haku genomic samples have been taken from several locations around the rohe including Pudney Rock, Ohiwa, Plate Island, Astrolabe Reef, and Whakaari (White Island).

Carla Finn is the PhD student working on the project supervised by Peter Ritchie, supported by Maren Wellenreuther and David Chagné. Among other work, Carla will generate a high-quality genome assembly for haku.

Image Credit: **Naomi Springett** (CC BY NC ND)

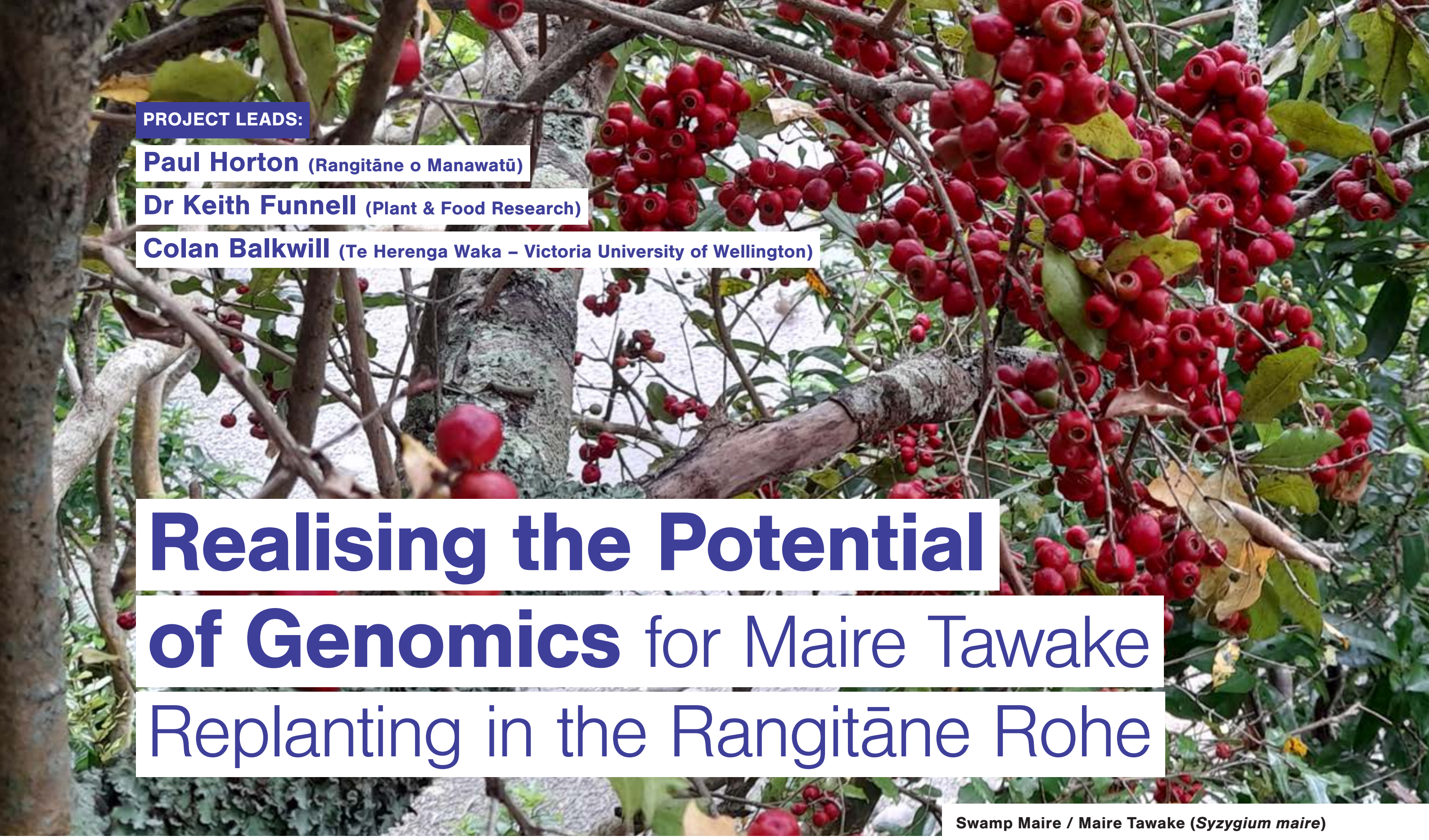
PROJECT LEADS:

Paul Horton (Rangitāne o Manawatū)

Dr Keith Funnell (Plant & Food Research)

Colan Balkwill (Te Herenga Waka – Victoria University of Wellington)

Realising the Potential of Genomics for Maire Tawake Replanting in the Rangitāne Rohe



Swamp Maire / Maire Tawake (*Syzygium maire*)

Maire tawake (swamp maire; *Syzygium maire*), a nationally critically threatened tree, is the subject of active restoration in the Manawatū region, within the rohe of Rangitāne o Manawatū.

Populations of maire tawake are small and fragmented, typically surrounded by farmland. With fewer than 12 mature trees within the rohe, the lack of genetic diversity may hinder the establishment of a naturally sustainable forest population through the replanting programme alone. By integrating mātauranga Māori with genomics-based pūtaiao, Rangitāne o Manawatū hopes to inform plans for forest restoration, including this species.

The demand for skills and data that support Māori aspirations is growing. This partnership enables Rangitāne o Manawatū to better participate in the Aotearoa science system, facilitated by researchers. Rangitāne o Manawatū will gain access to a broad range of scientific expertise. In return, Rangitāne o Manawatū will support scientists authentic understanding of mātauranga and kaitiakitanga.

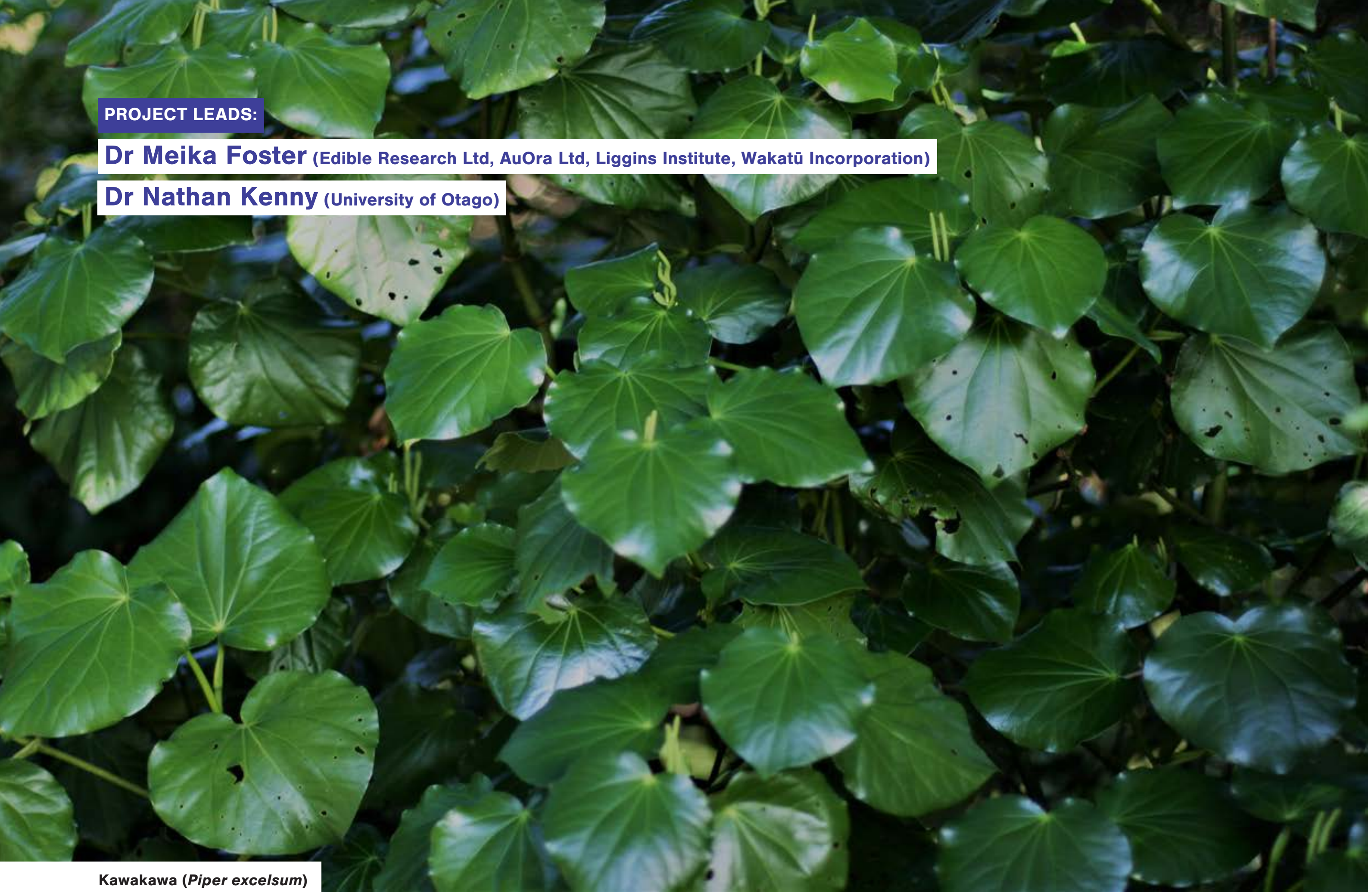
Together with habitat suitability mapping, genomic information will be built into an integrated restoration model for maire tawake in the Manawatū, allowing finer scale identification of suitable planting locations and genetic material within the rohe.

This year, hui and frequent informal exchanges are allowing the partners to plan, implement, and interpret the results of the project.

Genomic data from 135 maire tawake seedlings were aligned to the high-quality maire tawake genome to produce a large dataset of 720,000 DNA variants. The size of this is unprecedented for a threatened taonga tree. Kinship analysis provided in-depth information about the genetic relationships among the seedlings and adult trees from the rohe, which is being used to select which seedlings to prioritise for replanting and restoration, and to prioritise collection of germplasm. Meanwhile, ecological analysis in the Rangitāne o Manawatū rohe has helped identify suitable sites for replanting.

Short videos have been taken by Rangitāne o Manawatū Kia Ora FM to document the journey of Rangitāne o Manawatū engaging with genomics-based pūtaiao, including tikanga associated with sampling of foliage that was used for DNA extraction. This digital information is currently in post-production.

Image Credits: **Karin Van Der Walt - Otari Native Botanic Garden**



PROJECT LEADS:

Dr Meika Foster (Edible Research Ltd, AuOra Ltd, Liggins Institute, Wakatū Incorporation)

Dr Nathan Kenny (University of Otago)

Kawakawa (*Piper excelsum*)

Phylogenetic and Transcriptomic Analysis of Kawakawa for Rongoā and Kaitiakitanga

New this year, this project seeks to add genomics information to the rich existing mātauranga on kawakawa to deepen kaitiaki understanding of this taonga.

Kawakawa (*Piper excelsum*), a taonga in every sense of the term, is of great ecological and cultural importance. While multiple varieties of kawakawa are known from across the motu, to date there have been no investigations of the diversity, population structure and inter-relatedness of sub-populations of this species using a range of technical tools. Using a tikanga-informed pathway for sample acquisition, processing, data sovereignty and benefit-sharing, the team is using a transcriptomic approach to understand kawakawa diversity and bridge this knowledge gap.

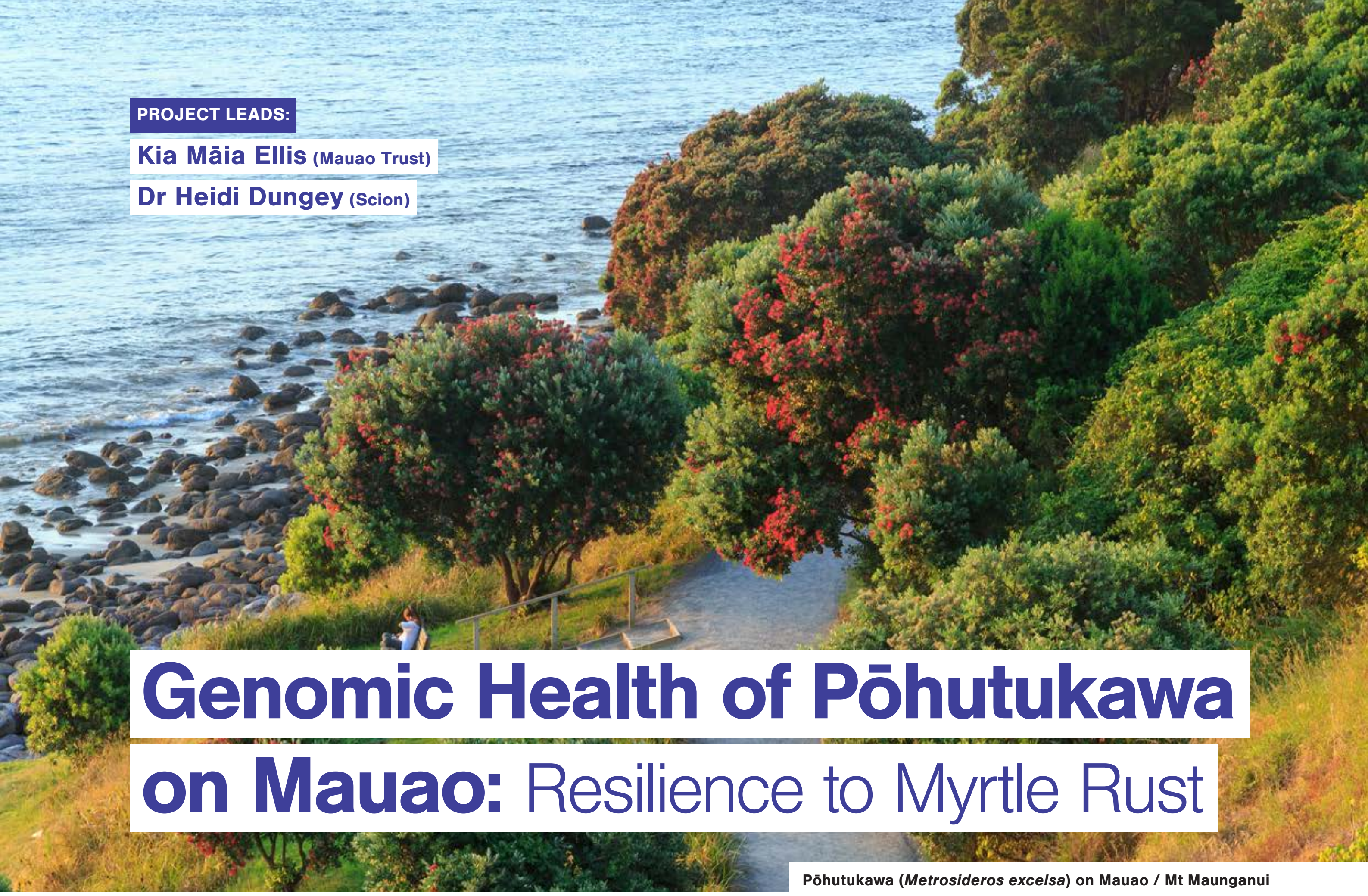
Robust data showing the whakapapa relationships among kawakawa could inform decisions around eco-sourcing to protect biodiversity in revitalisation or horticulture initiatives. In turn, genetic information aligned with mātauranga may provide additional insights regarding the efficacy of local kawakawa varieties for health and wellness applications.

A deeper understanding of the traits of regionally specific kawakawa varieties could lead to new high-value products with distinct provenance and narratives that support tikanga-led economic development.

Molecular work commenced in March 2023. Samples of kawakawa have been gathered, mainly from Te Tauihu, with some from further afield. These samples will be supplemented with those from other rohe as this work progresses.

A robust protocol for extracting high quality RNA has been tested and verified, which required optimisation due to the high phenolic content of kawakawa leaves. Library preparation is near complete, and sequencing is pending.

The postdoctoral fellow on this project, Roseanna Gamlen-Greene (Waikato-Tainui, Pākehā) started working on the project in late February 2023.



PROJECT LEADS:

Kia Māia Ellis (Mauao Trust)
Dr Heidi Dungey (Scion)

Genomic Health of Pōhutukawa on Mauao: Resilience to Myrtle Rust

Pōhutukawa (*Metrosideros excelsa*) on Mauao / Mt Maunganui

Another new project this year, Tauranga Moana hapū and the Mauao Trust are working together with Scion scientists to determine whether the pōhutukawa trees on Mauao are likely to be native or introduced.

The project will use genetic sequencing and leaf shape to help determine whether pōhutukawa on Mauao are native, introduced or hybrids. Developing a genetic sequencing strategy for pōhutukawa will inform pōhutukawa management decisions for iwi under attack from myrtle rust and provide capacity development in genomics for mana whenua.

While the project is in its very early stages, hapū are involved in the development and participation at every level of the project to ensure that the research outcomes provide capacity development of local kaitiaki and ongoing protection and management of Mauao.

The most exciting outcome to date has been securing a Māori candidate at Toi Ohomai Institute of Technology to commit their passion, time, and energy to enrol in master's level research in genetic sequencing to answer genomic questions.

Māori principles and values are woven throughout the research, with close alignment with the Mauao Trust strategy for the health and well-being of Mauao. This is the first research project on Mauao to be led by a Māori organisation. It provides a unique opportunity for Māori to achieve specific skills in genomics and build capacity of local kaitiaki to be directly involved in biosecurity management on Mauao.

Pōhutukawa (*Metrosideros excelsa*) on Mauao / Mt Maunganui

PROJECT LEADS:

Prof Nick Rahiri Roskruge **(Tāhuri Whenua)**
Prof Peter Dearden **(University of Otago)**
Assoc Prof Jen Tate **(Massey University)**

He Wae Kai Pakiaka:

Genomics of Māori Traditional Root Crops as Insurance for Food Security

The Māori perspective of horticulture recognises a growing and urgent need for food security alongside food sovereignty of what can be termed traditional Māori foods. Recent examples of this need are the effects of recent cyclones, where some iwi and hapū interests have lost entire crops and seed material for the next season.

New this year, *He Wae Kai Pakiaka* seeks to provide a strategic and secure approach to the gathering of both mātauranga and plant materials, supported by science through characterisation of landraces, which are then retained under international germplasm protocols to ensure accessibility for future generations.

The National Māori horticulture collective, Tāhuri Whenua, leads the project and draws from their kaumātua rōpū and broad membership to gather and manage plant materials and mātauranga into a collection with appropriate protocols.

Genomic approaches will be used to characterize genetic traits of kūmara and taewa to support and accompany accessions to the Centro Internationale de la Papa (International Potato Centre/CIP) in Peru, which hosts data and a germplasm collection for a range of traditional crops, as a food security insurance policy.

An additional outcome of He wae kai pakiaka is transferability to future applications across other Indigenous communities of Te Moana-nui-a-Kiwa. The traditional knowledge component of the project will add a layer to support Indigenous oversight of the resource alongside the science activity.

In the words of project leader Nick Roskruge, “The whakatauakī that underpins our project - He wae kai pakiaka – a literal reference to being grounded in our approach – applies to us as people, and all the natural resources we work with – we are all whānau of Papatūānuku”.

PROFESSOR PETER DEARDEN, PROFESSOR NICK RAHIRI ROSKRUGE, AND SAII SEMESE

PROJECT LEADS:

Prof Mik Black (University of Otago)
Dr Rudiger Brauning (AgResearch)
Assoc Prof Libby Liggins (Massey University)

Mahuika, one of the supercomputers which make up the High Performance Computing Facility at NIWA's Greta Point Campus

The Aotearoa Genomic Data Repository: A Nationally Significant Database for Taonga Species

The *Aotearoa Genomic Data Repository* has been implemented in alignment with the *Te Mata Ira* and *Te Nohonga Kaitiaki* guidelines and follows the principles of Māori Data Sovereignty. This repository helps organisations or individuals exercise their kaitiakitanga over taonga, while facilitating controlled data management and sharing.

The AGDR now hosts genomic data from 25 species (13 new datasets over the past year) across 30 projects and has received 12 applications for access this year. It is now possible to incorporate Biocultural Labels for data sets hosted in the AGDR.

The applications for access included one each for kanakana (lamprey) and hoki, two for snapper, and eight for mānuka. Most of the applications, including four applications for mānuka, were approved. Although no applications were declined, four are awaiting additional information from the researchers.

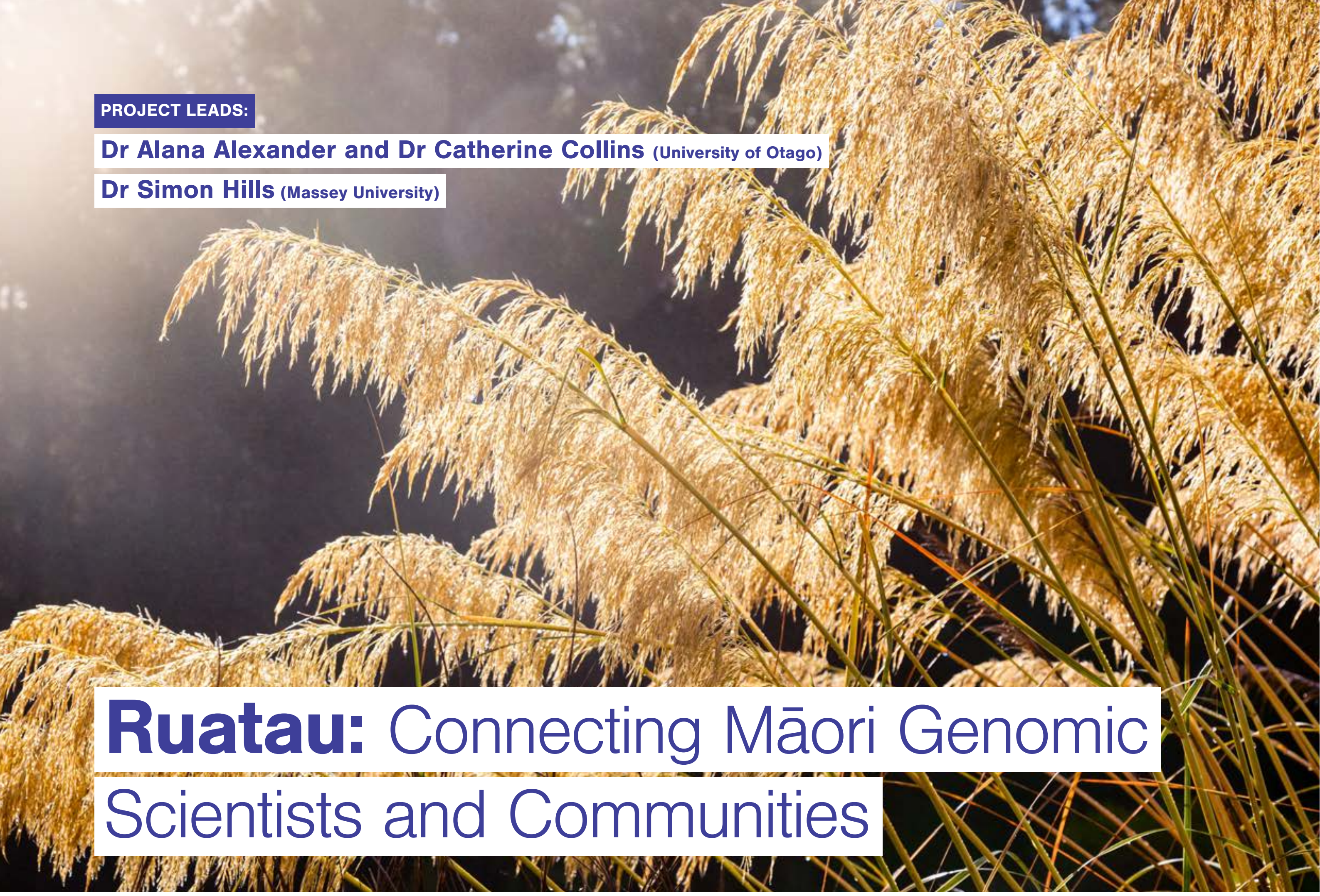
The approved projects relating to mānuka included:

- Evolutionary study of the *CER1* gene family in Myrtaceae.
- A phased genome of *Melaleuca quinquenervia* to shed light on allelic variation of NLR-type resistance genes.
- Transcriptome analysis of myrtle rust in mānuka.
- C-C coupling in the biosynthesis of natural compounds.

The project team is currently working with the recently established Māori advisory group to provide ongoing guidance on the operation of the data repository.

A manuscript describing the AGDR has been accepted, pending minor revisions for a *Molecular Ecology Resources* special issue *Indigenous Contributions to Molecular Ecology*. The manuscript describes the mātauranga Māori and Māori data sovereignty principles underlying the repository, and includes contributions from both Ben Te Aika (first author) and Tracey Godfery.

The AGDR was promoted at the eResearch NZ Conference in Hamilton in February 2023, with a presentation on *Building a taonga species data repository for Aotearoa New Zealand.* Mik is also participating in the wānanga being run within the MWLR led project *Towards a Te Tiriti based DNA Reference Library for Aotearoa's Indigenous Genome*, which included a presentation with Claire Rye (NeSI) on the AGDR – *Wānanga 3: Database architecture, process, and pathways.*



PROJECT LEADS:

Dr Alana Alexander and Dr Catherine Collins **(University of Otago)**

Dr Simon Hills **(Massey University)**

Ruatau: Connecting Māori Genomic Scientists and Communities

Empowering Māori communities to make informed decisions about important genomics issues such as hauora (health) of people, or kaitiakitanga (guardianship) of taonga is vital. Māori scientists (kaipūtaiao Māori) can help through understanding community perspectives, priorities, and concerns. But Māori are under-represented in academia and in genomics research. Furthermore, many Māori genomics researchers grew up Te Ao Pākehā and feel that they need more grounding in Te Ao Māori.

Ruatau is developing processes and resources to increase the capability of both Māori communities and Māori scientists to achieve and contribute to genomic mātauranga aspirations. Specifically, *Ruatau* helps support an increase in unique whānau-led research, culturally informed genomics research, Māori participation in research, as well as the creation of a network of Māori with interests in genomics.

The project explicitly focuses on Māori (researchers and communities) and is being implemented following kaupapa Māori best practices for engagement, including co-design. This builds capability and capacity in these researchers, as well as supporting the development of leaders in genomics within local communities.

The project is currently in a resource-building phase. Some of these resource materials are now available for end-users to access and have been provided via prototype genomics wānanga to representatives of Kāti Huirapa Rūnaka ki Puketeraki (Puketeraki).

The team has communicated and met with representatives of Ōtākou marae to discuss a potential genomics wānanga with the Ōtākou community. Co-lead Alana Alexander has also met with representatives of Komiti Kaupapa Taiao (Kāti Huirapa Rūnaka ki Puketeraki) and the East Otago Taiāpure committee to run a genomics mini-wānanga at their request, with materials also circulated to the wider community via zoom videos, to help with their capacity for screening research requests, and to provide feedback on the materials. This was a useful opportunity to trial the genomics wānanga materials being assembled.

As well as engagement with community groups, Alana was a keynote and invited speaker to conferences, the co-leads contribute to the *GENE411* and *ZOOL424* courses at Otago, and the companion resources being developed for kaipūtaiao upskilling are publicly available. This year Alana won a Genetics Otago Student Supervisor Award 2022.

PROJECT LEAD:

Assoc Prof Maui Hudson (University of Waikato)

SING Aotearoa: Mentoring Emerging Māori Genomics Researchers

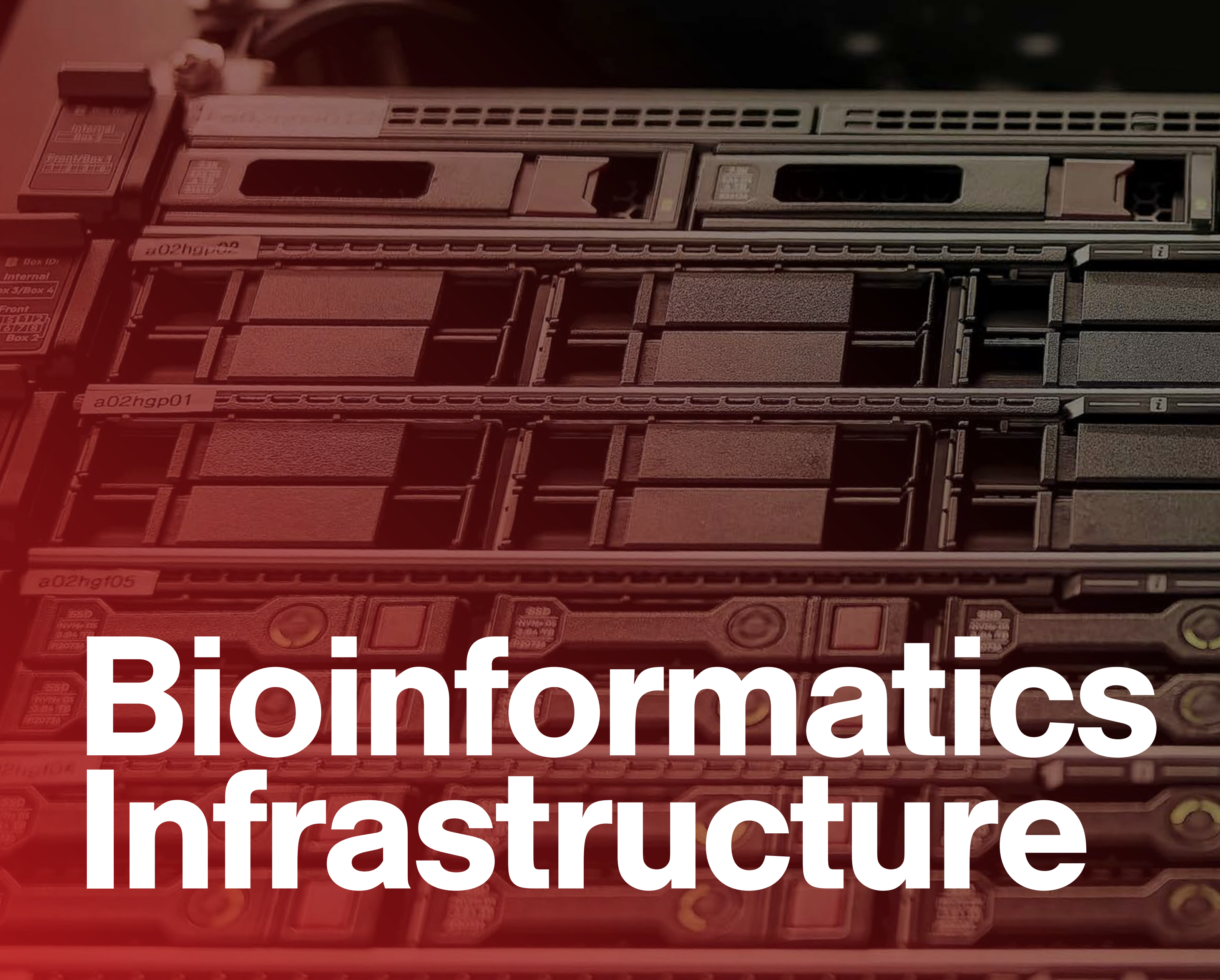
Genomics research is still an emerging discipline that has significant potential benefits for Māori, particularly in the environment, health, and socio-economic sectors. The Summer Internship for Indigenous Genomics (SING Aotearoa) initiative delivers a training programme for pakeke and tauira Māori interested in learning about genomics and bioinformatics, and the ethical and cultural issues around genomics research.

A kaumātua role for *SING Aotearoa* has been established, which was a key action from the last wānanga.

This year 25 participants attended the four-day SING2023 residential wānanga. Local hapū/iwi were key contributors to the event with key kaumātua present and opening SING2023, establishing the integral connection among hapū, iwi, and local community with participants, local and international speakers, and facilitators. *SING Aotearoa 2023* was a highly effective and engaging wānanga program, with a unique and enriching learning experience for interns, and a platform for Māori to connect and engage with the genetics and genomics in a meaningful and culturally appropriate way.

A part of the event, the group had a hīkoi (walk) on Maungatautari (Sanctuary Mountain), where mātauranga Māori from ahi kaa (local residential experts) provided insights and understanding about the whenua, ngahere (forest), plants and animals.

Four *SING* interns also had the opportunity to attend Queenstown Research Week in 2022.



Bioinformatics Infrastructure

This year Genomics Aotearoa’s bioinformatics programme has supported four ongoing projects with a primary focus on building bioinformatics infrastructure. These are *Bioinformatics Capability, Genome Graphs, Distributed Sequencing,* and *High Quality Genomes and Population Genomics.*

The work would not be possible without the Genomics Aotearoa/NeSI partnership. New Zealand eScience Infrastructure (NeSI) provides critical support for bioinformatics research in Aotearoa. The NeSI strategy to support research aligns with Genomics Aotearoa’s commitment to increase the capability of our genomics researchers. This strategic alignment makes us ideal partners.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PROJECT LEADS:

Prof Mik Black (University of Otago)
Dr Rudiger Brauning (AgResearch)
Dr Kim Handley (University of Auckland)
Dr Joep de Ligt (ESR)

Bioinformatics Capability

Supporting Aotearoa’s Genomics Researchers’ Technical Needs

Bioinformatics combines biology, computer science, mathematics, and statistics to enable the tools and strategies needed to analyse genomic data. This is the key discipline that underpins Genomics Aotearoa’s work. Although researchers arrive at bioinformatics from a variety of fields, they all share the same problem - how to analyse large, complex sets of data to extract biological meaning.

The greater the capability in bioinformatics, the more we can do with the information we gather, and the bioinformatics requirements of the research community drive our other infrastructure needs: computing resources, data management and training.

The *Bioinformatics* project enables national leadership to support and develop on-going capacity and capability in Aotearoa. The project also fulfils the technical requirements to support the cultural infrastructure of Genomics Aotearoa, such as the AGDR.

The team is strongly engaged with the eResearch community. In February 2023 Genomics Aotearoa was the lead host (along with NeSI and REANNZ as co-hosts) of the annual eResearch NZ Conference. This was a hybrid online and in-person event in Hamilton with 143 registrations. Session topics included *Equity Challenges in eResearch* and *Indigenous Data Sovereignty*. Several Genomics Aotearoa-affiliated researchers presented at the meeting (Annie West, Joseph Guhlin, Natalie Forsdick, Libby Liggins, Maui Hudson, Zoe Yang, Ben Halliday, Mik Black).

The partnership with NeSI continues to be critical to bioinformatics capability, with Genomics Aotearoa researchers using more than 900,000 hours of CPU time over the past year, across 17 different projects.

The training partnership between Genomics Aotearoa and NeSI is also a major strength of this project, involving 73 workshops run over the past four years, with 1,521 participants. Tyler McInnes and Jian Sheng Boey have managed this training programme over the past year.

Participants in this year's workshops included:

- 248 attendees at 14 bioinformatics workshops. A substantial proportion of the 248 workshop attendees were from outside Genomics Aotearoa affiliated organisations.
- 30 early career researchers attended a two-day training and networking event that included sessions on the interface of science and Te Ao Māori, and media training at the Auckland University Shortland Street studio. Guest speakers included Mike Bunce, Julie Everett-Hincks, Anna Santure, and Libby Liggins.
- More than 25 Aotearoa based researchers from universities, CRIs, government and industry representatives) have attended the Metagenomics Summer School at the University of Auckland.

Workshop material is available via GitHub.

CASE STUDY:

Early career researcher support is a key focus for building capability

Genomics Aotearoa is a substantial contributor to the development of postdoctoral fellows and early career researchers in Aotearoa.

Since 2017, 56 early career researchers have contributed their expertise to Genomics Aotearoa projects, making it one of the most significant funders of early career researchers across research organisations in Aotearoa. Many of those have worked on key primary production and conservation initiatives within Genomics Aotearoa's *High Quality Genomes and Population Genomics* projects.

The organisation also supports PhD students across its partner organisations.

Bioinformatics lead Mik Black said the beauty of Genomics Aotearoa is its collaborative environment.

“We are a partnership of 10 universities and Crown Research Institutes, which gives us a natural collaborative advantage. And while institutional connectivity is an issue for early career development in Aotearoa, the cross disciplinary nature of our projects means that our researchers are working with different research institutes, iwi, government organisations and private sector groups.”

Genomics Aotearoa makes a point of bringing all the postdoctoral fellows from their different research institutions together at least once a year to upskill, share and collaborate.

Mik said Genomics Aotearoa recognises the importance of early career researchers to the future of science in Aotearoa and relies on the postdoctoral fellows to innovate and to pass on their expertise.

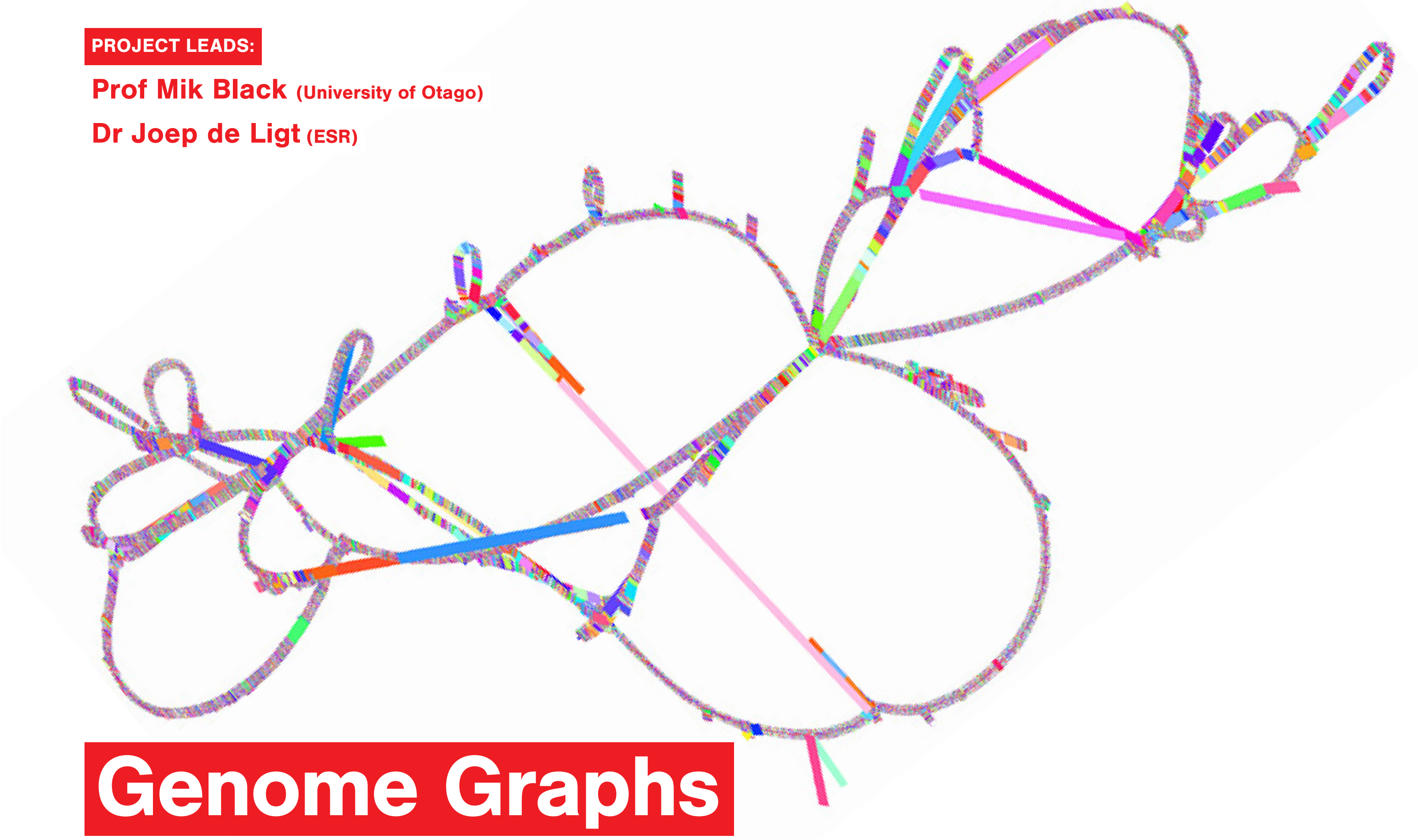
“Genomics Aotearoa postdoctoral fellows and other early career researchers have been leading the development of bioinformatic pipelines – processes to gather and analyse genomic information – resulting in outcomes that are of international standard.”

“The ECRs also play a key role in training programmes – something Genomics Aotearoa has invested heavily in – to the point that some of the programme attendees have become trainers themselves. Our training participants appreciate taking their skills back into their workplace, both to use on their research and to pass on to their teams.”

PROJECT LEADS:

Prof Mik Black (University of Otago)

Dr Joep de Ligt (ESR)



Genome Graphs
to Unravel Pangenomes

Pangenome analysis is increasingly being used to assess the entire genomic variation in a population, or even a species. But pangenome analysis is a challenge for bioinformatics. Graph-based analyses offer an improved approach to detect genomic variation. Although graph-based bioinformatic methods are relatively new, they are experiencing strong uptake internationally, and have been used in several new Genomics Aotearoa projects.

This project explores the use of genome graphs for pangenome construction, and for performing genome-wide variant analysis using graph-based methodology.

As well as their trailblazing work for infectious disease analysis, the team has been diligently working towards planned Genomics Aotearoa training sessions, ensuring that the benefits of these cutting-edge technologies reach the wider genomics community in Aotearoa. Next year promises to be an exciting one as the team continues to push the boundaries of graph-genome methodologies, foster collaborations, and make significant contributions to the field of genomics.

The *Genome Graphs* project has linkages with several other Genomics Aotearoa-funded projects.

The team has engaged with the *Rakeiora* and *Variome* project teams and will provide a report on the effectiveness of Genome Graph methods for the analysis of genomic data from admixed populations, which is of clear relevance to the genomic data for Māori collected the projects.

Several other successful collaborations include construction of a sheep pangenome using the PGGB software, with Rudiger Brauning (AgResearch), and work with Tammy Steeves and Jana Wold from the University of Canterbury on a Kākāpō pangenome (one of Aotearoa's unique taonga) for comprehensive genetic variation analysis. Patrick Biggs (Massey University) is a new collaborator on the project and is working on the application of graph-based methods for the analysis of bacterial genomes and ensuring that pipelines run on smaller/local infrastructure.

As well as three publications in review from the team, postdoctoral researcher Zoe Yang presented at two significant conferences, three new pipelines have been developed and made available on GitHub, and Mik Black has incorporated genome graph content into the *GENE315 – Genomes* course in third year Genetics at the University of Otago.

CASE STUDY:

Genome graphs lead to unique insights into infectious diseases

A pangenome is an integrated collection of the genomes of multiple individuals from a specific species.

Until recently, genomics research used a single ‘reference’ genome to provide a baseline, rather than a collection of genomes. This means analysis and findings are always influenced by the reference genome. No single genome can represent the genetic diversity of an entire species, and rarer, but important diversity is easily missed.

Now that sequencing genomes is much less expensive, it is possible to sequence many genomes for a species. Sometimes an entire species can be sequenced, as is the case for kākāpō.

However, combining genomes is not straightforward. Genomics Aotearoa’s *Genome Graphs* project team is testing new techniques to better understand infectious diseases, as were used in the recent seminal paper on the first human pangenome.

Pangenome graphs are data structures used to represent and compare the genetic variation within a species or a population, providing a comprehensive view of their genetic diversity.

The aim of the Genomics Aotearoa *Genome Graphs* project is to make this new analysis process accessible to people across Aotearoa in many different sectors. The initial focus is on research in human health and primary industries, but it has broad applicability.

The project team works with the international group who created the Pangenome Graph Builder (PGGB), which is led by Erik Garrison at the University of Tennessee Health Science Center. The PGGB toolset is highly scalable, applicable to all species, and is highly regarded amongst current techniques for constructing pangenomes.

Project researchers Zoe Yang and Joep de Ligt tested PGGB in the analysis of pathogen genomes, specifically to analyse *Neisseria meningitidis* disease in Aotearoa. A resulting manuscript is recently published with Frontiers in Genetics. One of the reviewers commented that the paper addresses both an urgent and an intriguing issue, “Obviously, pangenome graphs have great advantages over linear reference genomes when working with diverse pathogenic species. However, this seems to be the first study of that kind.”

As Joep says, “Ultimately, our innovative approach paves the way for enhanced comparative genomics and a deeper comprehension of genetic variations in (infectious) diseases, thereby enhancing our understanding pathogen evolution and spread.”

The next year promises to be an exciting one as the team continues to push the boundaries of graph-genome methodologies, fostering collaborations, and hosting training workshops to contribute to capability in research institutions across the motu.

PROJECT LEADS:

Dr Joep de Ligt and Dr Donia Macartney-Coxson (ESR)

Distributed Sequencing

Towards Real-Time Outcomes in Animal and Human Health

Distributed sequencing in the field has promising benefits for a range of applications. Small, portable sequencing devices are now able to capture genomic data in real time without the need for specialised facilities.

This project tests the application of distributed sequencing to sites of need, developing and implementing pipelines to enable real-time analysis and reporting for decision making at these sites.

As a result, a more diverse group of people will be empowered to use these techniques and timely information will inform treatment decisions and improve outcomes. While Oxford Nanopore Technology (ONT) is quickly being adopted within the Aotearoa research community, this project aims to place the technology and the associated bioinformatic and reporting tools directly at the point of need: in the hands of human and animal healthcare professionals.

This project has two trial applications: human health and animal health.

The first year of the project has focused on recruitment, which is now in the final stages of completion. Due to unexpected major changes in currency exchange, the project has decided to move from using the Oxford Nanopore Technology (ONT) GridION sequencer, to the smaller MinION sequencer. This provides an excellent opportunity to validate the protocols on the very equipment that veterinary clinics will be using in the future.

Initial discussions have been held with a Ngā Wairiki Ngāti Apa representative regarding how the research could be aligned with a mātauranga approach, especially the potential of this project to protect and enhance the whenua. The team has links with a Ministry for Primary Industries *Sustainable Food and Fibre Futures* project on regenerative agriculture – Whenua Haumanu – and through this have established contacts with Māori regenerative agriculture practitioners in Taranaki, with hopes to test the potential of this research on their sheep farms.

PROJECT LEADS:

Assoc Prof Thomas Buckley (Manaaki Whenua – Landcare Research)
Dr Shannon Clarke (AgResearch)
Dr David Chagné (Plant & Food Research)
Assoc Prof Anna Santure (University of Auckland)

Pōhutukawa (*Metrosideros excelsa*)

High Quality Genomes and Population Genomics

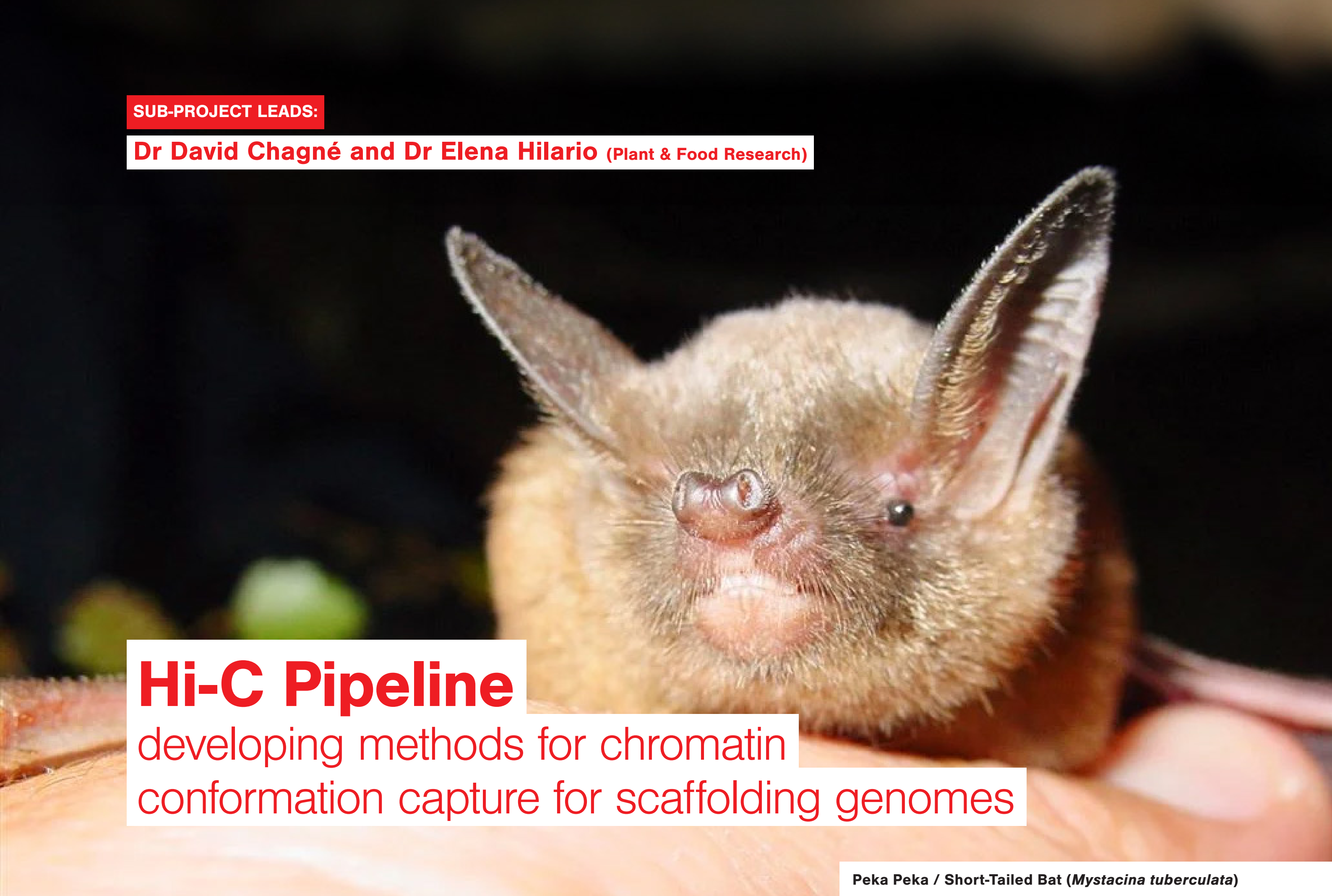
High Quality Genomes and Population Genomics sub-projects have produced multiple bioinformatic tools which are available on our on GitHub. Not only does this work empower our current researchers but it provides a framework for future research infrastructure.

SUB-PROJECT LEADS:

Dr David Chagné and Dr Elena Hilario (Plant & Food Research)

Hi-C Pipeline

developing methods for chromatin conformation capture for scaffolding genomes



Peka Peka / Short-Tailed Bat (*Mystacina tuberculata*)

Hi-C sequencing is a high throughput chromosome conformation capture technique to analyse spatial genome organization and map higher order chromosome folding and topological associated domains.

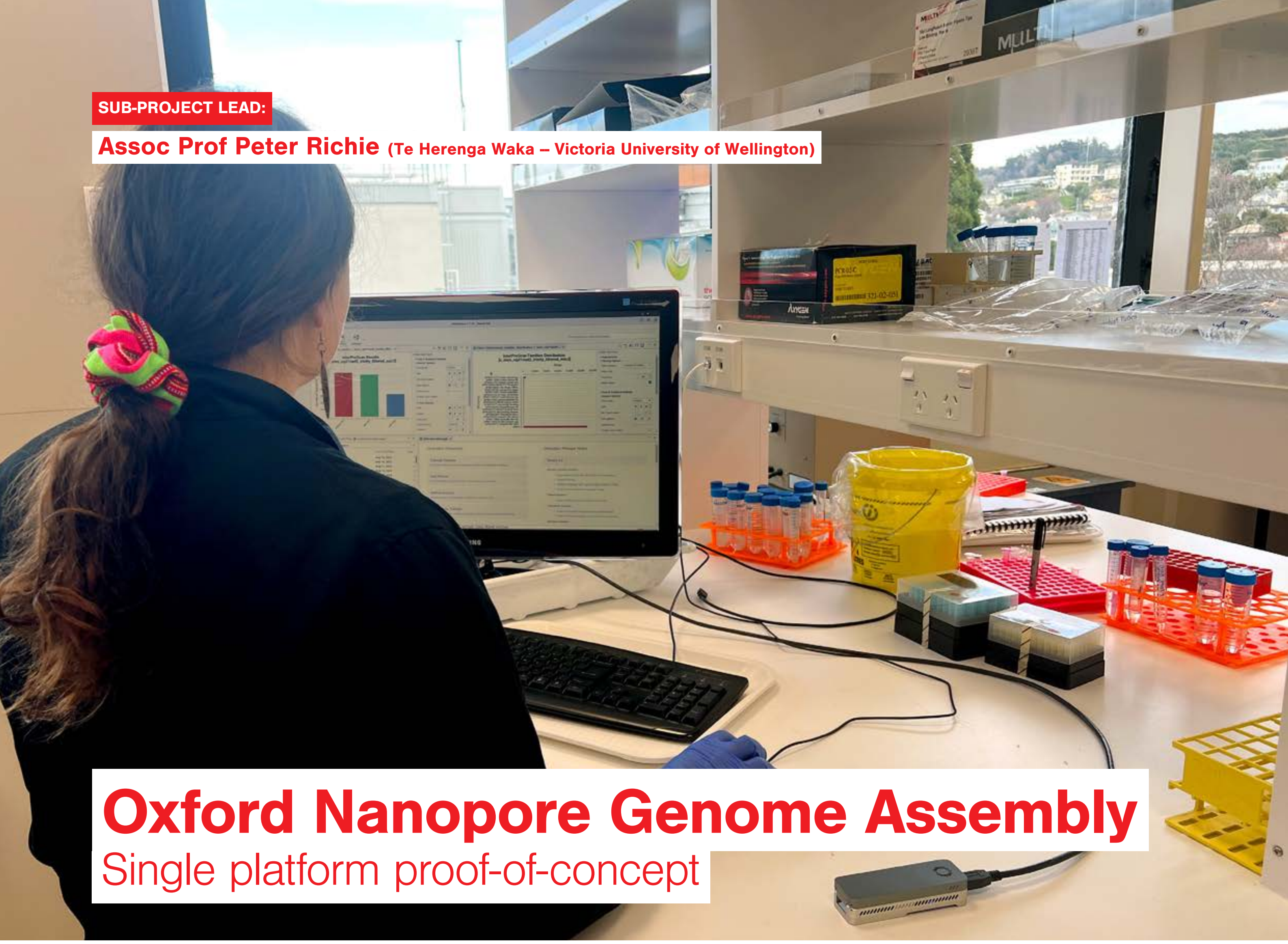
This sub-project will help researchers constructing genome assemblies using the Hi-C technique. Commercial solutions exist for Hi-C techniques, however, they often do not work well, or need extensive optimisation to be implemented for some recalcitrant species.

This year the sub-project team optimised protocols for Hi-C and tested these across a wide range of species: myna bird, peka peka, kiwifruit (multiple ploidies), karaka, *Eucalyptus*, grape, *Wahlenbergia* (plant), sea urchin and finfish.

Sharing knowledge has been a major focus. Many Genomics Aotearoa and other collaborators were trained by Elena Hilario and Ignacio Carvajal (Plant & Food Research) on methods for Hi-C. Co-leads David and Elena presented at several conferences, and the team has developed a number of pipelines.

The team is grateful for the collaboration of Ashley Jones from the Australian National University, Canberra. Ashley has been a collaborator since 2019 and is a talented molecular biologist and an experienced bioinformatician with sound knowledge of long read sequencing.

Karaka (*Corynocarpus laevigatus*)



SUB-PROJECT LEAD:

Assoc Prof Peter Richie (Te Herenga Waka – Victoria University of Wellington)

Oxford Nanopore Genome Assembly

Single platform proof-of-concept

The dual objective of this sub-project is to generate reference genomes for key species in the marine environment that will provide the resources to promote the application of genomics in fisheries and climate change science, and to be a proof of concept that a single sequencing platform can be used to generate genome assemblies.

The outcomes of the sub-project are directly relevant to academic and government institutions that want to use reference genomes for the benefit of Aotearoa's fisheries. Anyone who wants to use Oxford Nanopore for genome assembly can use the sub-project's bioinformatics pipeline.

Nine species will be sequenced as part of this research: hapuka, haku (kingfish), blue moki, kahawai, kuparu (John Dory), two toroa (albatross) sub-species, marine sponge, and black coral. Initial datasets have been sequenced for six out of nine species and genomes produced that could be used in population genomic analyses. The genome data produced will be accessible via the AGDR, and methods will be publicly available.

Regardless of the encouraging preliminary findings, the amount of data generated was insufficient to critically filter or optimise the genomes. More data are being sequenced and first insights from the expanded dataset for one of the species (hapuka) suggests significant improvement is possible.

Genomics Aotearoa Associate, the National Institute of Water and Atmospheric Research (NIWA), has provided tissue for kingfish and will benefit from the output for their aquaculture breeding program. Preliminary results have been shared with NIWA.

Members of the team have presented at several domestic and international events, and one publication has been produced. As well as postdoctoral fellow Tom Oosting, five PhD and postgraduate students are contributing to the project and sharing the learning journey.

Engagement and partnership with Te Ātiawa ki te Upoko o te Ika a Māui Pōtiki (Fisheries) Trust includes regular meetings with Mark Fenwick and trustees, co-led design of a proposal *Are New Zealand's fisheries species responding to climate change by evolutionary adaptation?* and developing a written deed/agreement/MoU with the Trust for the collection, potential export of samples, and subsequent sample and data analyses and management, related to fisheries genetics research.

Image of Roseanna Gamlen-Greene Credit: **Josh Byford-Pothan**

Perrenial Ryegrass (*Lolium perenne*)

BEAKERtaxa

A novel model for species identification

The BEAKERtaxa sub-project is investigating the application of natural language processing methods to identify species from DNA kmers (short DNA sequences that are used for genome analysis), which will be of value to bioinformaticians and researchers locally and internationally. Researchers will be able to access BEAKER through a public GitHub repository where issues and suggestions can be raised. The focal species is ryegrass, and its symbiotic endophytes.

BEAKERtaxa is a collaboration with pasture growers from Minderoo in Perth, Australia. The collaboration involves the exchange of ideas and knowledge as well as testing once the model is developed further. Ryegrass test datasets have been generated in collaboration with Marty Faville (AgResearch, Grasslands) to aid development and testing of both BEAKER and benchmarking current bioinformatic methods for taxonomic assignment.

Work completed so far includes development of the BEAKER model (developed by Joseph Guhlin from the University of Otago), review of current taxonomic binning methods, with a plan for benchmarking these against BEAKERtaxa, and setting up datasets for testing and benchmarking. These test datasets include publicly available mock bacterial communities as well as selecting, growing, and sequencing of five perennial ryegrass cultivars with three *Epichloë festucae* symbiotic endophytes.

Rachael has presented at several events and won an award at the Advances in Genome Biology and Technology 2023 (AGBT) conference.

Public Engagement

Our genomics exhibition at Tūhura Otago Museum was revamped this year and became our major outreach initiative.

We also continued our fortnightly Friday seminar series, interactions with the media, monthly newsletters, and public engagement through social media.

genome
SCIENCE OF LIFE

The feedback on the exhibition at Tūhura Otago Museum in May last year was so positive that we were invited to display it again over the summer months, from early November 2022 until mid-February 2023.

The exhibition highlighted how the field of genomics is increasingly becoming part of our everyday lives, impacting our health and wellbeing, our primary industries and our environment.

Told in stunning picture panels, the exhibition narrated stories on the foundations of the science of genomics, its history, genomics in Aotearoa, and the role genomics played in the COVID-19 pandemic. During the exhibition 20,841 people visited Tūhura Otago Museum, in addition to 4,212 visitors during the first showing last year.

The exhibition was also supported by four community programmes.

Brave New World: Genomics in Aotearoa Panel Discussion

In association with the exhibition, in December 2022, a panel of some of Aotearoa's leading researchers discussed the power of genomic solutions in health, conservation, primary production, and how this brave new world could impact on our futures in Aotearoa. Sixty-three people attended in person with a further 133 people taking part online.

We thank James Maclaurin (Co-Director of the Centre for AI and Public Policy) for hosting the event, together with:

- Amanda Black (Tūhoe, Whakatōhea, Whānau-ā-Apanui) (Lincoln University, BioProtection Aotearoa)
- Jemma Geoghegan (University of Otago)
- Mike King (University of Otago)
- Justin O'Sullivan (University of Auckland, Liggins Institute)
- Tammy Steeves (University of Canterbury)

Ancient DNA and Dinosaur Day

Visitors met with University of Otago ancient DNA experts Nic Rawlence, Catherine Collins, and Michael Knapp in the *Southern Lands, Southern People* gallery where, surrounded by moa skeletons, they brought the pre-history of Aotearoa to life. On the same day Nathan Kenny and his team demonstrated how to extract DNA from a banana and how real-time sequencing works, and there was also a marathon screening of the first three Jurassic Park films.

All The Small Things Event

The science that shapes our lives was celebrated with an evening event in February 2023. The diverse programme included live music from local band *Bo and the Constrictors*, TED-style talks, open labs that showcased tiny science from a variety of the University of Otago's science departments, give-away sour dough starters and DNA strawberry daiquiris.

We thank the presenters who contributed to this lively event:

- Joep De Ligt (ESR) and Jemma Geoghegan (University of Otago) on the COVID-19 pandemic in 2020.
- Neil Gemmell (University of Otago) on searching for the Loch Ness Monster.
- Charlotte King (University of Otago) on forensic cold cases from the goldrush era.

Secrets of the Past Event

Genomics Aotearoa teamed up with the New Zealand Festival of Nature, Wild Dunedin to present a talk in April 2023 at Toitū. Ancient DNA and forensic experts Michael Knapp, Nic Rawlence, Catherine Collins and Charlotte Paul described Aotearoa at the time of the moa and Haast eagle, when people first arrived on our shores, and in the gold mining era, all through the bones they left behind. The event was attended by 60 people.

Media Engagement


RNZ

NEW ZEALAND / SCIENCE

Research sheds light on Kākāpō pandemic mystery

3:41 pm on 8 November 2022

Peter Dearden talked to media about the crucial role genomics played in tracing the single strain of *Aspergillus* responsible for wiping out four percent of the kākāpō population on Whenua Hou (Codfish Island) in the Foveaux Strait.


nzherald.co.nz

The hihi is one of the 'unluckier species' to live but can be saved from extinction

1 Jun, 2022 09:00 AM

Anna Santure's hihi genomics project received wide media interest, including an NZ Herald article, and interviews on Te Hiku, bFM, and Radio New Zealand.


Stuff opinion

How we can realise the huge potential of genetic modification technology

Peter K Dearden · 09:07, Jun 16 2023

Over the year Peter has championed genomics in the media broadly, including an interview with Radio New Zealand's Kim Hill on genomics, conservation, and the velvet worm, and the huge potential of genetic modification technology.


RNZ

SCIENCE / COVID-19

Genome sequencing and the pandemic

From Our Changing World, 5:00 am on 24 November 2022

Also on Radio New Zealand, Jemma Geoghegan and Joep de Ligt talked about how genomics was used to help decision-making in the COVID-19 pandemic.


RNZ

SECURITY

Does New Zealand need to up its biosecurity game?

From Nights, 9:08 pm on 14 June 2023

Ang McGaughran promoted *Invasomics* in The Conversation; and on Radio New Zealand Nights.


RNZ

SCIENCE / HEALTH

Planning for Aotearoa's genomic medicine future

From Our Changing World, 5:00 am on 1 December 2022

Cris Print and Phil Wilcox discussed the future of precision medicine in Aotearoa on RNZ

SING Aotearoa was featured in a TVNZ Te Karere item, which included interviews with Phil Wilcox and keynote speaker *SING* USA, Krystal Tsosie.

eResearch Conference 2022

Genomics Aotearoa again co-hosted this conference alongside NeSI and REANNZ, at the University of Waikato in February 2023. The theme was *Capability, Connectivity and Equity*, with a strong focus on research and capability-building initiatives that involved partnerships with Māori.

Although attendance was disrupted by Cyclone Gabrielle meaning some people were only able to attend online, 143 people registered to hear about work from across the spectrum of eResearch.

Education Outreach Programme

This year Genomics Aotearoa initiated an education outreach pilot to promote genomics in education.

Efforts to build a home-grown workforce of genomic biologists and bioinformaticians depend on a pipeline of students having an interest in such careers when they embark on tertiary study. Currently, genomics is discussed only briefly in the secondary school classroom. As a long-term workforce strategy, Genomics Aotearoa is looking to provide professional development opportunities to secondary school teachers, highlighting how genomic data impacts biological knowledge, and ways this can be incorporated into current NCEA assessments and student experimental work.

Co-ordinated by Dr Thierry Lints, Genomics Aotearoa held its first workshop for teachers at the Fale Pasifika at the University of Auckland, in collaboration with Pūtahi Manawa (Healthy Hearts for Aotearoa New Zealand). Key speakers included Mike Bunce (Senior Science Advisor of the Department of Conservation), educators who shared their expertise in conducting environmental DNA sampling studies with primary and secondary school students, and leaders of organisations dedicated to increasing Māori representation in STEM careers, including genomics. A significant positive outcome was that post-workshop, a majority of teachers reported a significantly increased interest in pursuing environmental DNA sampling activities with their students, and most teachers had an increased, or significantly increased, desire to run DNA-based practical lessons in the classroom (such as PCR and gel electrophoresis).

Ongoing work includes the development of carefully crafted and tightly integrated genomics teaching resources that are culturally responsive and appropriate for the learning progression of students across curriculum levels. Further workshops are planned for the coming year.

DR THIERRY LINTS (EDUCATION OUTREACH CONSULTANT)

Seminar Series and YouTube

This year Genomics Aotearoa hosted 20 fortnightly seminars via Zoom.

Nine of these were international speakers, including two presentations from the Human Pangenome Reference Consortium which introduced the project, and described some of the technical and ethical, legal, and social issues of human pangenome construction. The first was given by Nanibaa' Garrison and Ann McCartney, and the second by Karen Miga. On more than 50 people attended each of the 20 seminars with a total audience of 1016 attendees over the year. A list of presenters from the past 12 months is below, and information about previous seminars can be found on our website.

Our YouTube channel continues to grow.

Most of the content is Genomics Aotearoa and Asia-Pacific Genetic seminars, along with videos of Genomics Aotearoa activities. In just over a year, the channel has received 41,095 views and gained 512 subscribers.

Asia-Pacific Genetics
Seminar Series

Genomics Aotearoa also hosts the Asia-Pacific Genetics Seminar Series.

A collaboration between the Genetics Society of Japan (GSJ) the Genetics Society of Australasia (GSA), the Genetics Society of China, and the International Genetics Federation. This quarterly seminar series aims to further develop the field of genetics and promote international exchange among researchers in Asia and Oceania and attracts a global audience.

The four seminars so far were presented by: Prof. Emeritus Tomoko Ohta (Japan), Prof. Lisa Matisoo-Smith (Aotearoa New Zealand), Prof. John Bowman (Australia), and Prof. Guojie Zhang (China).

Shannon Clarke	AgResearch	Genomic Solutions: underpinning, implementation, and impact
Santiago C. González-Martínez	INRAE and University of Bordeaux (France)	Ecological genomics of forest tree adaptation in the face of climate change
Thomas Buckley	Manaaki Whenua Landcare	Using genomics to understand the evolution of NZ terrestrial invertebrates
Greg Jones	University of Otago	From epigenome-wide discovery to clinical utility, delivering on the promise of 'precision' cardiovascular disease risk prediction
Michael Hoggard	University of Auckland	Environmental viruses and their influence on the microbial dynamics of aquatic ecosystems
Patrick Sullivan	University of North Carolina and Karolina Institute	Lessons for conservation biology and human genetics from Zoonomia (alignment of 241 Eutherian mammals)
Zhongchi Liu	Maryland University	Wild strawberries as a model to investigate regulatory mechanisms of strawberry reproduction
Imogen Foote	Victoria University of Wellington	A whole-genome approach to resolve the genetic structure of Antipodean and Gibson's albatross populations
Peter Gogarten	University of Connecticut	Inteins as a tool to trace the transfer of genetic information
Roger Hellens	Kiwifruit Breeding Centre	The Kiwifruit Breeding Centre and critical issues on breeding faster
Elizabeth Ross	University of Queensland	Nanopore sequencing for cattle breeding
Andrew Allan	University of Auckland	Gene editing in perennial tree crops; why, how, what?
Alice Popejoy and Nanibaa' Garrison	UC Davis University of California	The Human Pangenome Reference Consortium (HPRC): Facing the ethical, legal and social issues (ELSI) of human pangenome construction
Leah Kemp	Garvan Institute of Medical Research	Why is it so hard to actually do reproducible research in computational biology? Lessons learnt from a bioinformatician's past self
Kirsten Boggs and Zornitza Stark	Sydney Children's Hospitals Network Victorian Clinical Genetics Services	Acute Care Genomics: rapid rare disease diagnosis on a national scale
Karen Miga and Ann McCartney	UC Santa Cruz, NIH and NHGRI	The goals of the HPRC, and broader application of the methods to other species
April Jauhal	University of Auckland	Baleen whale olfaction: Do whales smell?
Cecilia Deng	Plant and Food Research	Long Read Sequencing: Options, Applications and Bioinformatics
Richard Espley Chen Wu	Plant and Food Research	Bilberry genome assembly – how identifying a complex loci is a step closer to breeding a novel anthocyanin-enriched red-fleshed blueberry
Lisa Matisoo-Smith	University of Otago	Ka mua ka muri: Why understanding the Pacific past is important for Pacific genomic research today

CASE STUDY:

Seminars share knowledge with the global genomics research community

Genomics Aotearoa’s Friday seminar series has blossomed, from a good idea that linked our researchers during the 2020 COVID-19 lockdown, to growing a Aotearoa New Zealand genomics community.

“The series has become a firm fixture in the genomics events calendar, where researchers want to invite overseas colleagues to contribute and participate, are volunteering to speak or organise a seminar. We enjoy a steady stream of enquiries”, says Claire Grant, Communications Manager.

The audience comes from more than 50 organisations, including universities and research institutes both here and overseas, service providers and schools. They include researchers, technicians, managers, policy makers, students, lecturers, health care providers and business development representatives.

Most seminars are recorded and uploaded to our Genomics Aotearoa YouTube channel; our international presenters are a major drawcard.

Genomics Aotearoa Co-Director Peter Dearden said there is wide-ranging value gained from hosting the hour-long seminars online on a Friday afternoon. “We have created a genuine sharing community that is supporting our work in growing genomics capacity and capability.”

Peter has managed some great discussions and questions over the last three years. “It’s been encouraging to see so many people quiz the speaker about their work and use the opportunity to discuss their own approaches with peers.”

“It’s gone from being a welcome contact between researchers from different institutions during the COVID-19 lockdown, to having an enduring forum open to anyone involved in genomics and bioinformatics; it’s been heartening to see cross disciplinary networks forming.”

“The international component is invaluable for us and creates opportunities to connect with overseas researchers.”

Former Genomics Aotearoa postdoctoral fellow Ann McCartney is working on the Human Pangenome Reference Consortium in the United States and has organised several international seminars.

“Working with Genomics Aotearoa, I had world class training in the production of high-quality reference genomes in a way that recognises, respects, and includes the rights of Māori. The seminar series provides a unique opportunity for international research collaborators to share and receive feedback on their research,” Ann said.

Early career researchers also appreciate they have a comfortable and supportive environment to present in, as Michael Hoggard from the *Environmental Microbiomes* project found.

“The seminar series is a great opportunity to present work to an audience of leaders and peers in the genomics community. It’s also an important way to break down research silos and facilitate interaction, collaboration, and knowledge sharing among groups with similar research interests,” he said.

Contributing Researchers

GA1802

TE NOHONGA KAITIAKI

Maui Hudson	University of Waikato
Chris Battershill	University of Waikato
Jason Mika	Massey University
Phil Wilcox	University of Otago
Jacinta Ruru	University of Otago
Matt Stott	University of Canterbury
Robert Brookes	University of Waikato
Tuti Nikora	University of Waikato
Ariane Thompson	University of Waikato
Lisa Warbrick	Massey University
Amanda Clarke	University of Waikato
Daniel Patrick	University of Auckland

GA1805

BETTER BREEDING VALUES

Dorian Garrick	Massey University
Michael Lee	University of Otago
Phil Wilcox	University of Otago
Shannon Clarke	AgResearch
Rudiger Brauning	AgResearch
David Chagné	Plant & Food Research
Christine Couldrey	LIC
Neville Jopson	AbacusBio
Andrew Hess	AgResearch
Yu Wang	Massey University LIC
Brittany Jones	University of Otago
Joshua Rich	University of Otago AgResearch

GA1807

CELL FREE GENOMICS

Parry Guilford	University of Otago
Mik Black	University of Otago
Fiona Hely	AbacusBio
Jonah Duckles	AbacusBio
Luna Zhang	AbacusBio
Miles Benton	ESR

GA1808

GENOMIC TRANSLATIONAL ONCOLOGY

Cris Print	University of Auckland
Peter Tsai	University of Auckland
Richard King	CDHB
Chang Ho	WDHB
Lindsey Harbour	ADHB
Henry Chan	WDHB
Kim Gamet	ADHB
Jaclyn Ting Fowler	ADHB

GA2202

SELECTIVE BREEDING OF KINGFISH

Maren Wellenreuther	Plant & Food Research University of Auckland
Chris Insley	Te Arawa Fisheries
Peter Ritchie	Victoria University of Wellington
David Chagné	Plant & Food Research
Carla Finn	Victoria University of Wellington

GA2206

IDENTIFYING PŌHUTUKAWA ON MAUAO

Heidi Dungey	Scion
Kia Maia Ellis	Mauao Trust
Jaroslav Klápště	Scion

GA1813

SING AOTEAROA

Maui Hudson	University of Waikato
Phil Wilcox	University of Otago
Katharina Ruckstuhl	University of Otago
Tuti Nikora	University of Waikato
Kimiora Hēnare	University of Auckland

GA1809

GENOMIC TYPING PATHOGENS

Jenny Draper	ESR
Xochitl Morgan	University of Otago
Paul Gardner	University of Otago
Patrick Biggs	Massey University
Christina Straub	ESR
Xiaoyun Ren	ESR
Joep de Ligt	ESR

GA1811 / GA2104

BIOINFORMATICS CAPABILITY

Mik Black	University of Otago
Peter Dearden	University of Otago
Helge Dzierzon	Plant & Food Research
Rudiger Brauning	AgResearch
Kim Handley	University of Auckland
Carmen Astudillo-Garcia	University of Auckland
Roy Storey	Plant & Food Research
Dan Jones	Plant & Food Research
Ngoni Faya	University of Otago
Miles Benton	ESR
Rachael Ashby	AgResearch
Leah Kemp	ESR
Libby Liggins	Massey University
Michael Hoggard	University of Auckland
Jian Shen Boey	University of Auckland
Joep de Ligt	ESR
Tyler McInnes	University of Otago

GA1812

CLINICAL GENOMICS

Stephen Robertson	University of Otago
Cheng-Yee Chan	CDHB
Natasha Henden	ADHB
Jo Martindale	CCDHB
Padmini Parthasarathy	University of Otago
Peter Tsai	University of Auckland
Kate Gibson	Genetic Health Service NZ
Rachel Stapleton	Genetic Health Service NZ
Ian Hayes	Genetic Health Service NZ
Patrick Yap	Genetic Health Service NZ
Kate Neas	Genetic Health Service NZ
Colina McKeown	Genetic Health Service NZ
Nerine Gregersen	Genetic Health Service NZ

GA2103

RUATAU

Alana Alexander	University of Otago
Catherine Collins	University of Otago
Simon Hills	Massey University
Kimiora Hēnare	University of Auckland
Karaitiana Taiuru	Independent contractor
Lisa Warbrick	Independent contractor
Phil Wilcox	University of Otago

GA2201

SHARING THE BENEFITS OF GENOMICS

Nathan Kenny	University of Otago
Andy Elliot	Wakatū Incorporation
Aroha Mead	Wakatū Incorporation (Subcontractor)
Charles Eason	Wakatū Incorporation
Jackie Stephens	Wakatū Incorporation
Katerina Achilleos	University of Otago
Marc Bailie	University of Otago
Aleisha Chalmers	University of Otago

GA1901

RAKEIORA

Cris Print	University of Auckland
Stephen Robertson	University of Otago
Phil Wilcox	University of Otago
Helen Wihongi	University of Auckland
Peter Tsai	University of Auckland
Jennie Harre Hindmarsh	Ngāti Porou Hauora
Donia Macartney-Coxson	ESR
Joep de Ligt	ESR
Miles Benton	ESR
Kimiora Hēnare	University of Auckland
Liz Goodin	University of Otago
Polona Le Quesne Stabej	University of Auckland
Ben Curran	University of Auckland
Claire Rye	NeSI
Matt Pestle	NeSI
Eiran Perkins	NeSI
Kenny Zhao	NeSI
Martin Kennedy	University of Otago
Hugh Stuart	Ngāti Porou Hauora
Frances King	Ngāti Porou Hauora
Ben Te Aika	University of Otago
Huti Watson	Ngāti Porou Hauora
Caroline Koia	Ngāti Porou Hauora
Jun Huh	NeSI
Jade Scott	University of Auckland CTNZ
Ben Halliday	University of Otago
Rochelle Style	University of Auckland
Ben Rangihuna	University of Otago
Alastair Lamont	University of Otago

GA2105

EXTENDING WHOLE GENOME ANALYSIS

Stephen Robertson	University of Otago
Louise Bicknell	University of Otago
Miles Benton	ESR
Leah Kemp	ESR
Natasha Henden	ADHB
Jo Martindale	CCDHB
Cheng-Yee Chan	CDHB
Kim McGuiness	Rare Diseases NZ
Padmini Parthasarathy	University of Otago
Sam Hawarden	University of Otago
Emma Wade	University of Otago

GA2204

ANALYSIS OF KAWAKAWA

Nathan Kenny	University of Otago
Jackie Stephens	Wakatū Incorporation
Meika Foster	Wakatū Incorporation
Roseanna Gamlen-Greene	University of Otago

GA2004

IDENTIFYING DRIVERS OF *STREP...*

Paul Gardner	University of Otago
Joep de Ligt	ESR
Julie Bennett	University of Otago
Nigel French	Massey University
Christina Straub	ESR
Te Whetu Kerekere	University of Otago
Toby Dawn-Sugrue	University of Otago

GA2205

DISTRIBUTED SEQUENCING

Joep de Ligt	ESR
Kristene Geydes	Massey University
Donia Macartney-Coxson	ESR
Patrick Biggs	Massey University
Rhys White	ESR
Georgina Dawson	ESR
Max Berry	Wellington Hospital
Jane Clapham	ESR
Andrea Grana	Massey University
Anjan Purkayastha	ESR

GA1806 / GA2101 / GA2303

ENVIRONMENTAL METAGENOMICS

Kim Handley	University of Auckland
Gavin Lear	University of Auckland
Bevan Weir	Manaaki Whenua Landcare Research
Louise Weaver	ESR
Matt Stott	University of Canterbury
Charles Lee	University of Waikato
Susie Wood	Cawthron
Hwee Sze Tee	University of Auckland
David Waite	University of Auckland
Carmen Astudillo-Garcia	University of Auckland
Chanenath Sriaporn	University of Auckland
Jian Shen Boey	University of Auckland
Michael Hoggard	University of Auckland
Jemma Geoghegan	University of Otago
Murray Cox	Massey University
Maui Hudson	University of Waikato
Annie West	University of Auckland
Millie Ram	University of Auckland
Esme Spoelstra	University of Auckland
Sophie Palmer Dale	University of Auckland
Craig Cary	University of Waikato

GA2003

GENOME GRAPHS

Mik Black	University of Otago
Joep de Ligt	ESR
Zoe Yang	ESR
Nuzla Ismail	University of Otago

GA2203

SWAMP MAIRE

Paul Horton	Rangitāne o Manawatū
Keith Funnell	Plant & Food Research
Colan Balkwill	Victoria University of Wellington
Debbie Te Puni	Rangitāne o Manawatū
Alana Nuku	Rangitāne o Manawatū
Jon Procter	Rangitāne o Manawatū
Julie Deslippe	Victoria University of Wellington
David Chagné	Plant & Food Research
Wayne Blissett	Rangitāne o Manawatū

GA1810

EPIGENOME WIDE ASSOCIATION STUDY

Greg Jones	University of Otago
John Pearson	University of Otago
Aniruddha Chatterjee	University of Otago
Basharat Bhat	University of Otago

GA2102 I

INVASOMICS

Angela McGaughran	University of Waikato
Manpreet Dhami	Manaaki Whenua Landcare Research
Ellie Parvizi	University of Waikato
Amy Vaughan	Manaaki Whenua Landcare Research

GA1803 and GA2301

AOTEAROA NZ VARIOME

Stephen Robertson	University of Otago
Phil Wilcox	University of Otago
Lisa Matisoo-Smith	University of Otago
Murray Cox	Massey University
Tony Merriman	University of Otago
Andrew Sporle	University of Auckland
Allamanda Faatoese	University of Otago
Huti Watson	Ngāti Porou Hauora
Liz Goodin	University of Otago
David Markie	University of Otago
Wyeth Wasserman	University of British Columbia
Ben Rangihuna	University of Otago
Brittany Hewitson	University of British Columbia
Sam Hawarden	University of Otago
Ben Halliday	University of Otago

GA2302

MĀORI TRADITIONAL ROOT CROPS

Nick Roskruge	Tahuri Whenua
Peter Dearden	University of Otago
Aleise Puketapu	Tahuri Whenua
Simon Apang Semese	Massey University
Hema Wihongi	Tahuri Whenua
Te Mauri Apiata	Tahuri Whenua
Jennifer Tate	Massey University

PF2050

TACTICAL GENETIC CONTROL OF RATS

Bjorn Oback	AgResearch
Goetz Laible	AgResearch
Susie Szakats	AgResearch
Donisha Liyanagama	AgResearch
Ming Li	AgResearch
Bobby Smith	AgResearch
Catherine Collins	University of Otago
Maren Wellenreuther	Plant & Food Research University of Auckland
Brooke Whitelaw	University of Otago
Sara Belcher	Victoria University of Wellington
Symon Palmer	Victoria University of Wellington
Ocean Mercier	Victoria University of Wellington
Alan King-Hunt	Victoria University of Wellington
Inaya Clark	Victoria University of Wellington

GA1801 / GA2002

HIGH QUALITY GENOMES

Thomas Buckley	Manaaki Whenua Landcare Research
David Chagné	Plant & Food Research
Jeanne Jacobs	AgResearch
Murray Cox	Massey University
Neil Gemmell	University of Otago
Peter Dearden	University of Otago
Anna Santure	University of Auckland
Emily Telfer	Scion
Maren Wellenreuther	Plant & Food Research University of Auckland
Chen Wu	Plant & Food Research
Ross Crowhurst	Plant & Food Research
Cecilia Deng	Plant & Food Research
Susan Thomson	Plant & Food Research
Elena Hilario	Plant & Food Research
Richard Newcomb	Plant & Food Research University of Auckland
Tammy Steeves	University of Canterbury
Gary Houliston	Manaaki Whenua Landcare Research
Duckchul Park	Manaaki Whenua Landcare Research
Patrick Biggs	Massey University
Roy Storey	Plant & Food Research
Orin McCormick	RAPiD Genomics
Matthew Tinning	Australian Genome Research Facility
Alana Alexander	University of Otago
Ann McCartney	Manaaki Whenua Landcare Research
Joseph Guhlin	University of Otago
Roger Moraga	TeaBreak Bioinformatics
Jessie Prebble	Manaaki Whenua Landcare Research
Laura Duntsch	University of Auckland
Florian Pichlmueller	University of Otago
Talia Brav-Cubitt	Manaaki Whenua Landcare Research
Manpreet Dhami	Manaaki Whenua Landcare Research
Chris Smith	Manaaki Whenua Landcare Research
Caroline Mitchell	Manaaki Whenua Landcare Research
Emily Koot	Plant & Food Research
Sara Montanari	Plant & Food Research
Seung-Sub (Shane) Choi	University of Auckland
Shannon Clarke	AgResearch
Peter Ritchie	Victoria University of Wellington
Michael Lee	University of Otago
Libby Liggins	Massey University
Rudiger Brauning	AgResearch
Christine Couldrey	LIC
Kim Handley	University of Auckland
Amali Thrimawithana	Plant & Food Research
Andrew Hess	AgResearch
Charles Hefer	AgResearch
Rachael Ashby	AgResearch
Annabel Whibley	University of Auckland
Natalie Forsdick	Manaaki Whenua Landcare Research
Tom Oosting	Victoria University of Wellington
Anna Clark	University of Otago
Monica Vallender	University of Otago AgResearch
Vanessa Arranz	Massey University
Jana Wold	University of Canterbury
Sarah Bailey	University of Auckland
Catherine Collins	University of Otago
Iggy Carvajal	Plant & Food Research
Imogen Foote	Victoria University of Wellington
Rebecca Clarke	AgResearch
Mitchell Weston	AgResearch
Carla Finn	Victoria University of Wellington
Manon Broadribb	Victoria University of Wellington
Miriam Pierotti	Victoria University of Wellington
Ben Roberts	Victoria University of Wellington
Melissa Nehmens	Victoria University of Wellington
Keith Funnell	Plant & Food Research
Chris Kirk	Plant & Food Research
Renata Blissett	Rangitāne o Manawatū
Richard Espley	Plant & Food Research

Glossary of Genomic Terms

TERM	DEFINITION
Allele	Alternative versions of a single gene. Used to characterise variation in the genome and compare among individuals.
Assembly	DNA sequencing technology cannot read whole genomes in one go; instead, it reads short pieces of bases from a genomic sequence. Sequence assembly aligns and merges fragments to form a longer DNA sequence to reconstruct the original genome.
Bioinformatics	The science of analysing genomic data.
Candidate genes	Genes of interest related to phenotypes or disease states.
Clinical geneticist	This is a medical doctor with special training in genetics who meets with patients to evaluate, diagnose, and manage genetic disorders. Clinical geneticists also assist in the management of genetic diseases by identifying preventable complications through early and accurate diagnosis and surveillance.
CRISPR-Cas9	CRISPR-Cas9 (Clustered Regularly Interspaced Short Palindromic Repeats) is a method of genetic manipulation consisting of two key molecules that introduce a change into the DNA. The molecules are: An enzyme called Cas9 which can cut strands of DNA at a specific location in the genome so that short sections of DNA can then be added or removed. A piece of RNA sequence called guide RNA (gRNA), located within a longer RNA which guides Cas9 to the correct part of the genome to cut.
DNA methylation	DNA methylation is one type of epigenetic mechanism that modifies the expression of genes and therefore their effects. DNA bases are modified by addition of a methyl group. Methylation can change the activity of a DNA segment without changing the genome sequence.
eDNA	Environmental DNA, or eDNA refers to many tiny traces of genetic material that remains as living things pass through water or soil. It can be used to survey the environment to detect traces of diseases or invasive species or rare species. Environmental DNA can be assessed using metabarcoding.
Epigenetics	Epigenetic mechanisms are those that modify inherited (genomic) gene expression, that is how, and when, those genes are switched on (or off). These epigenetic switches may be due to environmental exposure or lifestyle factors. Non-genetic factors include the environment like diet, gut microbiota, toxin, and drug exposure, psychological and physical stressors, and levels of activity throughout life. Measuring the epigenetic changes that occur in diseases, including cancer and heart disease, can provide understanding of the underlying mechanisms.
Epigenome	The set of all epigenetic modifications to an individual's genome.

TERM	DEFINITION
Eukaryote	An organism whose cells contain a nucleus surrounded by a membrane and whose DNA is bound together by proteins (histones) into chromosomes. Animals, plants, and fungi are eukaryotes.
Exemplar research	A research model that provides leadership and examples for further research in similar fields.
Finishing the sequence	Finishing an assembly involves refining the genomic sequence to eliminate sequencing errors and to close gaps.
Genetic admixture	The presence of DNA in an individual from a distantly related population or species, because of interbreeding between populations or species who have been reproductively isolated and genetically differentiated. Admixture results in the introduction of new genetic lineages into a population.
Genetic counsellors	These are healthcare professionals with training in human genetics and counselling who guide patients and their whānau with a genetic disorder through the process of understanding and making informed healthcare decisions.
Genetic gain	The rate of genetic improvement within a breeding population over time. An important concept in conventional quantitative genetics and breeding, genetic gain can be defined as the increase in performance achieved annually through artificial selection.
Genetic-linkage mapping	Illustrates the order of genes on a chromosome and the relative distances between those genes.
Genome annotation	The process of attaching biological information to genetic sequences.
Genome duplication	A widespread phenomenon in plant genome evolution, where an organism can sometimes inherit two copies of the genome of its parents, instead of only one copy. The doubling of DNA then persists over generations and the duplicated copies can result in the evolution of new functions.
Genome map	Helps scientists to define which parts of the genome are physically linked to each other. The landmarks on a genome map that aid navigation might include short DNA sequences, regulatory sites that turn genes on and off, and genes themselves.
Genomic signature	Genomic regions of DNA sequences that provide information about the activity of a specific group of genes in a cell or tissue.
Genome variation types	Genome variations include mutations and polymorphisms. Mutation is often used to refer to a variation that is associated with a specific human disease, while the word polymorphism implies a variation that may or may not affect a physical characteristic. Genetic variations also include gene deletions, gene additions and structural variations (see structural variants).
Genome-wide association studies	A search for parts of the genome associated with characteristics of interest, one example being human diseases.
Genotype (noun)	An organism's set of genetic variations.
Genotype (verb)	To determine genetic variation in a genome.
Germline	The cells which develop into eggs and sperm.

TERM	DEFINITION
Haplotype	Combinations of alleles from several locations on the same chromosome that are inherited together. Used to infer the relationships among genes (and species) and the characterize the evolutionary processes that shape their ancestry.
HiC	Hi-C sequencing is a high-throughput chromosome conformation capture technique to analyse spatial genome organization and map higher-order chromosome folding and topological associated domains.
Imputation	The mathematical process of replacing missing data with estimated values.
Introns	Regions of non-coding DNA sequence within genes.
Long read sequencing *Also known as:* **Third generation sequencing**	Involves new forms of sequencers that can read long distances down one strand of DNA. There are currently two effective technologies: Pacific Biotechnology (PacBio) - an imaging approach that allows the detection of the incorporation of single labelled base pairs one after another into a strand of DNA being replicated. Oxford Nanopore (Nanopore) - uses tiny, charged pores that a strand of DNA is drawn into, and as each base passes through the hole it changes the charge in a way that can be measured. These changes in charge are then assigned to each base and the sequence is built up from there.
Linked read technology	Uses a unique barcode system to label short DNA sequences from individual molecules that are close to each other on the genome, so they can be linked to create longer sequence reads.
Metabarcoding	Metabarcoding is a type of sequencing that enables the simultaneous identification of many organisms within one sample. The aim is to assess species composition within a sample.
Metabolomics	This process detects chemicals or metabolites and provides a read-out of what chemicals are in a tissue at any one time.
Microbiome	The community of microorganisms (such as fungi, bacteria, and viruses) that exists in a particular environment. The environment could be anything from the body of an animal or plant or soils and waterways.
Mutations	The changing of the structure or function of a gene, caused by the alteration of single base units in DNA, or the deletion, insertion, or rearrangement of larger sections of genes or chromosomes. The resulting variant form may be transmitted to subsequent generations.
Non-coding DNA	Genes (coding DNA) account for a small percentage of the DNA in the genome - knowing the entire genome sequence will help scientists study the parts of the genome outside the genes. Non-coding DNA includes the regulatory regions that control how genes are turned on an off, as well as long stretches of DNA of unknown function.
Nonsense mutation	A DNA mutation that results in a non-functional protein.
Nucleotides and bases	A single *base* of DNA is made up of Adenine (A), Cytosine (C), Guanine (G) or Thymine(T). Base pairs are the two opposing nucleotides (a base, a sugar molecule, and a phosphate group) on a double-stranded DNA molecule. Adenine pairs with Thymine, and Cytosine pairs with Guanine.
Omics or **'omics**	Omics is the collective name for the disciplines that characterise and quantify pools of biological molecules that translate into the structure, function, and dynamics of organisms. Examples include genomics, proteomics, metabolomics, metagenomics, phenomics and transcriptomics.

TERM	DEFINITION
Pangenome	A pangenome (also pan-genome or supragenome) is a combined collection of genes from all strains or varieties within a group of organisms.
Phenotype	Observable characteristics influenced both by an organism's genotype and by the environment.
Pathway	In genetics, a pathway is a set of genes that work together in a biological process.
Pharmacogenomics	This is the study of how genomic variation within the individual or their disease (including gene expression, epigenetics, germline, and somatic mutations) influences one person's response to drugs. The aim is to optimise drug therapy by maximising therapeutic effect and minimising adverse effects.
Pipeline	A process for the preparation, development, production, and analysis of genomic data.
Population genomics	The large-scale application of genomic information to study populations, including entire genomes of an entire species (e.g., kākāpō).
Prokaryote	Unicellular microbial organism that lacks a nucleus. All bacteria are prokaryotes. A related group are Archaea – these are also unicellular but are different to bacteria.
Proteomics	RNA (made from turned-on genes) is translated into protein. Proteomics is a technique to look at the broad range of proteins in a cell or tissue (using mass spectrometry). We can usually identify which proteins are present in a cell and what they are doing.
RNA	RNA (ribonucleic acid) is the molecule that takes information from DNA to make protein and has many other activities. RNA is only made from genes in the DNA that have been activated.
Short read sequencing *Also known as:* **Next generation** or **second-generation sequencing**	A DNA sequencing method that produces relatively short fragments of DNA sequences, typically ranging from a few dozen to a few hundred base pairs in length. This sequencing approach is characterized by its ability to generate a large volume of sequencing data quickly and cost-effectively.
SNPs	About 90 percent of human genome variation can be accounted for by single nucleotide polymorphisms, or SNPs (pronounced *snips*). These are variations that involve just one nucleotide, or base. These markers are used to genotype individuals to better understand population genetic variation and can be used to detect genes that are under selection.
Somatic	Refers to the cells of the body in contrast to the cells that make sperm or eggs (germline cells).
Structural variation or **Structural Variants (SVs)**	Structural variation describes individual or group differences in genome structure, such as gene deletions, insertions, duplications, inversions, and translocations. These variant regions are scattered throughout genomes and are often associated with gene expression changes and observable differences among individuals (phenotypic differences).
Transcriptomics	Sequencing RNA from a tissue or cell to measure the set of active (expressed) genes.
Variome	The complete set of genetic variations found in populations of species.
Whole genome sequencing (WGS)	The process of determining the complete DNA sequence of an organism's genome.

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