The Patricia Kailis Fellowship in Rare Genetic Disease

Your Perkins Supporter Newsletter | June 2025

HARRY PERKINS INSTITUTE
OF MEDICAL RESEARCH

IN THIS EDITION

Your support means so much to our family

Our family continues to be so very proud of its association with Professor Gina Ravenscroft at the Perkins. There has been so much to celebrate this year, thanks to your generous and ongoing support along with a transformational gift from the Children's Health & Disability Foundation WA. Your kindness has ensured that the Dr Patricia Kailis Fellowship in Rare Genetic Disease provides financial stability and back-up for Gina and her team as they continue to progress this vital research into rare genetic diseases.

Thank you for partnering with us and for helping to make our mother's legacy possible.

Maria Kailis **Amanda Kailis**
on behalf of the Kailis Family

Thank you so very much for your amazing, and continued, support of the Dr Patricia Kailis Fellowship in Rare Genetic Disease

Your generosity in recent years has ensured that Professor Gina Ravenscroft and her team can continue their ground-breaking research without the risk, faced by most researchers in Australia, of very limited access to funding from government grants. Your support provides reassurance that this important work can continue, no matter how long it might take.

We remain especially grateful to Patricia's family, Maria, Amanda, Alex and George, for their continued support of the Dr Patricia Kailis Fellowship, for believing in Gina and Nigel and their teams' vision for this research–something for which Patricia so passionately advocated.

Our heartfelt thanks and appreciation to the Kailis family–and you–for the loyalty and friendship afforded to us at the Perkins.

We present this update to you with our thanks and our very best wishes and we look forward to hosting you at the Harry Perkins Institute of Medical Research again soon.

Happy reading!

Warm regards,

Shelley Mason
Senior Manager, Key Relationships

An update from

Professor Gina Ravenscroft,
Dr Patricia Kailis Fellow in Rare Genetic Disease

Thank you so much!

As this newsletter goes to print, I am sending you my very best wishes from the European Human Genetics Conference in Milan. Joined by four of our team members–Jevin, Lein, Carolin and Mridul–I feel so lucky that we could attend and benefit from this Congress together.

What a year 2025 has been. The past 12 months has been another incredibly successful year for us in terms of research outputs and publications, winning grants and translation of our research findings. Thank you for partnering with us to ensure that our vital research can continue and go from strength-to-strength.

In 2024, we published six novel human disease genes across a range of different rare diseases including, muscle diseases present at birth or in adulthood, sensory neuropathy, and a neurodevelopmental disorder with global motor and language delay. Each of these discoveries means that several families now have a genetic diagnosis. **They know what the disease in their family–in their child–is**, and what the risk is for the rest of the family and their future children. It also opens new avenues for research for us and for many around the world to better understand the biology underlying these rare conditions.

Your support of me and my team–your kindness to our lab and the research projects that we have underway–means the world to us. Thank you so much.

2024 was such a good year for us, with so many novel diseases described, that I find it hard to believe we will have another year like it but I am constantly surprised–every year–by the generosity bestowed upon our team and our research, that I believe it is possible, thanks to your faith in us.

Thank you for making Patricia's vision a reality–I really do believe that we have established the West Australian "hub" for rare genetic diseases that she always dreamed we would.

Professor Gina Ravenscroft
Dr Patricia Kailis Fellow in Rare Genetic Disease

Introducing Dr Carolin Scriba

"I first joined the lab in December 2018 for a six-week Summer Vacation project, having just earned my undergraduate degree in Molecular Genetics and Biotechnology at Curtin University. In 2019, I continued with the team to complete my Honours, working on the newly discovered repeat expansion in the gene *RFC1*, which causes a progressive neurodegenerative disorder called CANVAS. Following my Honours I worked in the diagnostic laboratory at PathWest as a Medical Scientist, setting up testing for these newly discovered disorders and learning the ins and out of the diagnostic process.

In 2021 I started my PhD, and was fortunate to complete my studies across the Rare Disease Genetics and Functional Genomics group at the Perkins and the neurogenetic diagnostic lab at PathWest. Having a foot in both camps provided greater insight into the diagnostic process and facilitated smoother implementation of the research findings into the diagnostic lab.

With Gina as my incredible supervisor and mentor, the next four years were spent searching for new disease genes and variants, and focusing on improving diagnostic outcomes for patients with ataxias (a type of neurodegenerative condition that can affect the ability to walk, talk and swallow). Receiving a genetic diagnosis is a crucial turning point for many patients and families–being able to contribute to this has been hugely rewarding and a constant driver.

On 14 February, I submitted my thesis entitled, "*Disease gene discovery, and development and implementation of novel tools, to increase diagnostic yield in neurogenetic disease*". With many of the people who have supported me throughout the experience, in the room, it was truly a special moment–see photo above –and definitely a Valentine's Day I will never forget! (and here I am pictured with friend and colleague, Dr Jess Baker).

It has been a privilege to work in this area of research and collaborate with so many generous and enthusiastic scientists. This is a very exciting time to be working in the field of genomics and I am looking forward to what the future will bring."

Commissioned artwork sets new lab tradition at the Perkins

We were thrilled to receive this beautiful artwork from the very talented India Parker, who lives with Limb Girdle Muscular Dystrophy. Starting in 2025, every PhD student who graduates from the Ravenscroft and Laing labs will receive a print of the Geraldton Wax (a nod to Western Australia) and the DNA helix (a nod to our research). Don't you just love it?

Raine Priming Grant aims to improve lives for patients and families

Dr Mridul Johari's project, *From Coding to Cryptic–Exploring the RNA World of Inclusion Body Myositis (IBM)*, secured $247,815.63 in funding from the Raine Medical Research Foundation in their 2024 grant round.

Inclusion Body Myositis (IBM) is the most common late-onset muscle disease, characterised by progressive muscle weakness and difficulty swallowing, significantly impacting the quality of life of those affected. With no effective treatments available, Mridul's research aims to fill critical knowledge gaps by uncovering the molecular mechanisms driving this debilitating disease.

"It is an absolute honour to receive this recognition and support from the Raine Medical Research Foundation. This funding will allow us to explore the untapped potential of regulatory RNAs in IBM and could pave the way for transformative treatments. By focusing on the molecular triggers of disease progression, we hope to provide new insights that will ultimately improve the lives of patients and their families."

Mridul's innovative project delves into the roles of cryptic RNAs and peptides in IBM, which are often overlooked in standard studies but could hold the key to understanding the disease's complex interplay between immune system abnormalities and muscle degeneration.

Using cutting-edge technologies such as total RNA sequencing, spatial transcriptomics and proteomics, the research will map interactions between RNAs and proteins within affected muscle tissues.

Mridul's research is further strengthened through collaborations with leading experts, including Prof Merrilee Needham (from Fiona Stanley Hospital), A/Prof Andreas Roos (Proteomics, University of Duisburg-Essen) and Prof Alistair Forrest (Systems Biology & Genomics, Harry Perkins Institute of Medical Research), Neuropathology WA and the Myositis Association of Australia.

Together, they aim to unlock new diagnostic and therapeutic pathways for IBM, bringing hope to those living with this challenging condition.

Stay tuned! We look forward to sharing the outcomes of Mridul's project in the months to come.

Ravenscroft and Laing Lab Retreat – a day out in Mandurah

Gina's and Nigel's teams took time out of their busy research schedules to enjoy a day of goal setting and professional development sessions that focused on preparing papers, giving conference presentations and planning lab work. They even squeezed in a game of Frisbee and visited one of the Mandurah Giants by Thomas Dambo! This was the fourth annual Ravenscroft and Laing Lab Retreat which aims to introduce new members to the team and share the group's guiding principles.

Our heartfelt thanks to the Children's Health & Disability Foundation WA

The Perkins remains so truly grateful to the Children's Health and Disability Foundation WA for its support. Thanks to a transformative gift of $1 million–received in early 2025–the Fellowship's campaign target has been met. The generous donation has also enabled the Ravenscroft and Laing labs to purchase a piece of equipment that, until now, could only be accessed interstate or overseas.

So many of you joined us on Tuesday, 8 April to mark this momentous occasion–thank you very much. It is thanks to your care, kindness and loyalty that The Patricia Kailis Fellowship in Rare Genetic Disease has been made possible. Your support of Gina, Nigel and their teams is so very appreciated!

Gina named Premier's Mid-Career Scientist of the Year

In August, Professor Gina Ravenscroft was announced as the Premier's Mid-Career Scientist of the Year. Awarded for her research into "the genetic causes of rare but serious neuromuscular diseases. She has made significant discoveries, including identifying previously unknown disease genes and her research helps provide a diagnosis and paves the way for new treatments."

The Goggle Squad go for Gold!

In late 2024 the Perkins launched a new peer-to-peer fundraising event to raise funds for medical research–the Perkins Plunge–a 12-hour overnight relay swim that took place from 6pm to 6am on 9-10 November.

The Ravenscroft and Laing labs were keen to get behind the initiative and signed up their team of 11 students, researchers and colleagues from the neuromuscular diagnostics team at PathWest, to form *The Goggle Squad*.

With the cumulative long distance swimming experience of approximately zero years, they jumped in the pool in June and started their training in preparation for the event!

Come November, with six months of swimming under their belts, confidence had grown, and they were ready to take on the Plunge. "The event was incredible," recalls Dr Carolin Scriba. "Spirits were high and sleep forgone–we all had a wonderful time, with lots of friends, family and members of the lab dropping in throughout the night to support us."

The team also raised over $30,000 for medical research at the Perkins and were named the top fundraising team overall.

The team reports that since the event most of them have kept swimming and are looking forward to taking part again in November this year. Go Goggle Squad!

It is our pleasure to share with you an excerpt from Professor Gina Ravenscroft's speech on Tuesday, 8 April 2025. A celebration of all that the Patricia Kailis Fellowship in Rare Genetic Disease has achieved to date–thanks to you.

"I am so incredibly grateful to Patricia, the Kailis family and all of you for your support of this Fellowship and **I am honoured to be the recipient of this Fellowship and the custodian of Patricia's and Nigel's rich rare disease legacy here in WA. I never imagined in my wildest dreams that I'd find myself in this position.**

I have spoken before about the perilous journey that is a career in medical research, the short-term contracts, the highly competitive grant cycles where only 1 in 10 applications is funded and the impact that this environment has in terms of morale and productivity–sometimes it feels like we spend more time and energy applying for grants than actually doing the research. I want to share a little example. I have been a member of the World Muscle Society, the major international society in our field since 2007 and I have been an active member since then. Every year, we must renew our membership and every year there is the option to renew for one year or for three years…every year I renewed annually. Having the security of the Patricia Kailis Fellowship has meant that I have stopped worrying so much about whether I would still have a job in a few years' time. Three years ago, I renewed my WMS membership for three years, and just last week I renewed again for another three years. **It's a small thing, but it made me reflect and I think it illustrates what a difference the Kailis Fellowship has made to me personally.**

It also means that I can just get on with the research, with leading the wonderful team that I get to work with each day. I calculated overnight that my group has more than 100 years of collective experience in neuromuscular and genetics research.

All four of our post-doctoral researchers, including me, are supported by philanthropy–myself with the Kailis Fellowship, Dr Rhonda Taylor and Dr Josh Clayton with Safe Harbour Giving Circle Fellowships and Dr Mridul Johari with a grant from the Raine Foundation–two of us supported by pioneering West Australian women.

Being a large research team and being successful with grant funding and having philanthropic support, not just through this initiative, but via other families who also generously support our research, means that we have the capacity to take on projects that provide answers to families even when it's not some shiny new discovery. In some ways each family enrolled in our research is its own project and we have over 300 families from whom we have whole genome sequencing data. Our cohort makes up more than one tenth of all rare disease families with whole genome sequencing in the country.

Last year we identified two variants, two changes in the DNA, in a Western Australian man with an unusual muscle disease. He first sought medical attention when he was in his late 30s and his muscle weakness progressed rapidly with him becoming wheelchair dependent within a few years. **He was obviously very concerned.**

We did whole genome sequencing on him and his parents and found two variants, one inherited from each of his parents, in an important muscle gene–cofilin 2. There had only been 11 variants described in this gene ever before and these ones were both new–we didn't know enough about them to be sure that they were the cause of his condition.

Lein Dofash, a talented PhD student in the team, worked really hard to develop a way to tell if these variants in cofilin impacted the protein function. She was able to show that both variants rendered the protein non-functional. I recently saw this patient at a community day, and he knew we were trying to find the cause of his condition. I gave him an update and I saw the wave of relief wash over his face. He had two young boys, and he had been very worried that they might develop the same condition he had. Because we now know the likely cause of his disease are these two variants in cofilin 2, and because this is a recessive disease, he can't pass this condition on to his boys. **For this father and his family it was the most important study we could do.**

Similarly, just last week I had an email from a paediatric neurologist in Brisbane. We had been working on a family of hers with childhood onset sensory neuropathy, a nasty condition. In this family the disease is dominant, the grandmother was affected, she'd passed the condition on to her son and he in turn had passed it to his two children. This was the email from Anita:

> "***Thank you so much for getting a diagnosis–after 16 years of trying for this family!!*** *I thought we would never get there! The good news is that one of the affected children is planning a family, so this will provide information to allow genetic counselling.*"

Looking forward, I'm really excited about two projects within the team. The first is aiming to address a huge unmet need to better understand the genetics underlying recurrent pregnancy loss and providing answers for these couples. Around 300 miscarriages occur every day in Australia and for 20 per cent of these women, miscarriages will be recurrent. We are using a new genomic technology that relies on creating optical maps of the DNA to try to find answers. Recruitment so far is going quite well, via word of mouth, General Practitioners, fertility clinics and obstetricians, and some of the stories from the couples are heartbreaking–five, eight, ten miscarriages with no answers. If we can provide a genetic diagnosis to just a small portion of these couples, that will be huge progress.

The other project is also enabled by very generous philanthropic support from The Children's Health & Disability Foundation WA as well as Hearts and Minds Investments Limited and with thanks to Magellan Group. This has enabled us to purchase a bespoke piece of equipment that allows Dr Rhonda Taylor's team to generate 3D engineered muscle tissues from patient cells. We started out with patient blood cells from France, and Dr Josh Clayton converted them to stem cells. We could then change these stem cells in muscle and grow muscle cells here in Perth from these patients. Many of the blood cells were stored decades ago. It's remarkable what can be done with these precious banked samples. We can grow these muscle cells in culture, on a flat dish and these are useful for some aspects of our work, however the major consequence of many diseases we work on is muscle weakness. We need a way to model this weakness so that we can then test therapies. Using this Mantarray equipment Rhonda and her team can make 3D engineered muscles from these patient cells and measure the force they produce, we can measure how weak they are compared to healthy control muscle, and we can also test therapies in these engineered tissues to see if they improve muscle force. **Can we rescue the muscle weakness?**

I hope this gives you some insights into the ongoing projects within the team and into how your support of medical research is moving the dial for patients and families affected by genetic diseases.

I also want to make sure I pay this incredible gift forward–to those within my team and those to come. Compared to many research groups we are fortunate to have a little stability, and I'll continue to work hard to make sure our research excellence speaks for itself. May we continue to secure grants and philanthropic support so that we can keep the team together and improve the lives of patients and families.

Thank you again."

This (very large) photo was taken at the Australasian Neuromuscular Network Conference on the Sunshine Coast in early 2025. Dr Rhonda Taylor, Dr Josh Clayton, Emeritus Professor Nigel Laing AO and Professor Gina Ravenscroft all spoke at the Conference, giving five presentations in total. In June 2026 the group looks forward to hosting a joint meeting in Perth.

Mantarray Equipment a game-changer for our research–thank you!

"In June 2024 I was given the incredible opportunity to travel to Seattle to learn how to make 3D skeletal muscle tissues using patient cells. I spent most of my time in the laboratory of one of our collaborators, Professor David Mack. We first started working with this team in 2023 when they submitted an application to a charitable foundation in the US that funds research into Nemaline Myopathy. Our team had been studying Nemaline Myopathy since 1999 when Nigel and the team discovered that genetic changes in the skeletal muscle actin gene are a primary cause of this disease.

Since joining the team in 2018 I had been creating stem cell models of Nemaline Myopathy using patient cells. We could show that these stem cells could be instructed to turn into muscle cells, but we were limited in what we could do with these when grown on a traditional flat surface in the lab. We needed a more "functional" system–something that we could use to test whether our patient cells made muscles that modelled Nemaline Myopathy, like we see in patients.

Here is where our path fortuitously crossed with David–his original application for funding proposed to use an innovative piece of equipment he had co-developed and commercialised, the Mantarray! This neat piece of equipment allows you to make up to 24 engineered muscle tissues in a single 'plate' and measure how strong they are and how well they respond to signals that tell muscles to contract. We started collaborating and soon their team was able to show that our cells sent from Perth could make good 3D muscle tissues and that a novel treatment we had been exploring improved the function of patient tissues–great!

But...we considered–"this work is being funded for now, but the funding means we can only study our cells from Nemaline Myopathy patients, and when the funding runs out what then?" We decided it would be worthwhile for someone in our team to get hands-on experience with the Mantarray to learn the skills and assess whether it could be a worthwhile investment.

Fortunately, David and his team were happy to have me visit, so I spent a busy few weeks preparing cells to ship to his lab in Seattle. I went to Seattle and spent seven weeks learning how to use the Mantarray and analyse the data it produces. I also got some hands-on training at Curi Bio, the Seattle-based company that manufactures the Mantarray. **During my time in Seattle, I was able to make 168 muscle tissues from several of our patient and healthy control cells from Perth.** Although this was a very busy time, I was able to showcase what we

could do with this technology. I was incredibly grateful for this opportunity–it was only possible due to the generous support our team has received via philanthropic kindness.

The story doesn't end there! When I returned from Seattle I started investigating the possibility of us getting a Mantarray of our own. Given the cost of the equipment, we would have had to apply for funding from at least two funding bodies and wouldn't have received a result until the end of 2025. Even then, success was not guaranteed!

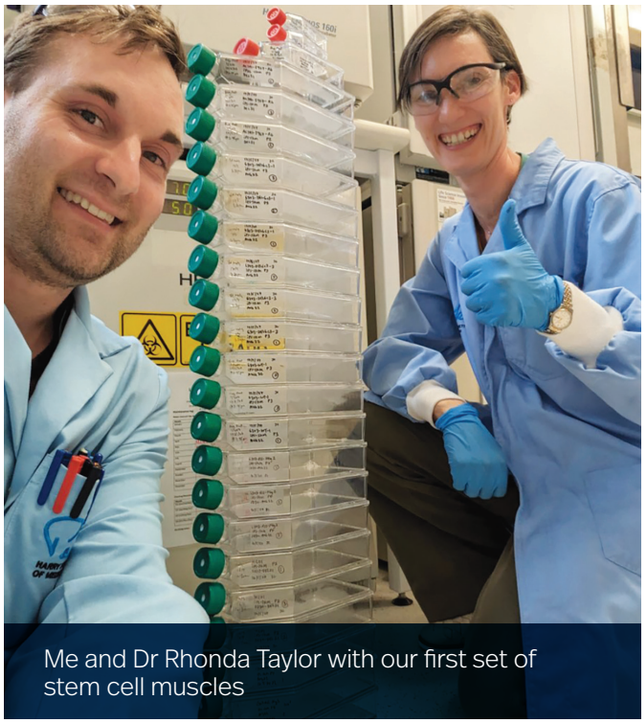
On my first day back at work in 2025 Gina called me into her office to chat with her, Nigel and Rhonda. I was nervous as the tone of the summons sounded ominous, but that changed instantly–Gina simply said, "we've just received the most amazing gift [at the time I didn't know it was from the Children's Health & Disability Foundation WA]... how soon could you sort out a Mantarray?"

Through my connection with David Mack and Curi Bio, I was able to arrange a generous discount, and we had a Mantarray in Perth by the start of February! Since then, I have conducted the install and got the machine up and running. I will be commencing work on the first batch of Perth tissue in June (a year after my trip to Seattle) and am very excited to see where this work goes!

I am incredibly grateful to everyone that has made this acquisition possible–a funding application would have taken weeks or months away from the lab, and may not have succeeded. Timing meant we were also able to beat the tariffs which may have put the Mantarray of reach. Having access to the Mantarray technology in our lab dramatically accelerates and improves the work that we can do now and puts us one big step closer to developing systems and treatments for neuromuscular diseases.

Again, thank you to the Children's Health & Disability Foundation WA, the Kailis Family and everyone associated with the Patricia Kailis Fellowship–your support is amazing."

- Dr Josh Clayton



Me and Dr Rhonda Taylor with our first set of stem cell muscles

Developing treatments for childhood muscle diseases using patient-derived cells

In 2024 and again in 2025, Dr Rhonda Taylor was awarded funding from the Channel 7 Telethon Trust to support research that aims to develop new treatments for muscle diseases affecting children.

Rhonda is the team leader of the Disease Models and Therapies team. This team has two aims:

1) developing new patient-centric cell models to better understand muscle diseases, and

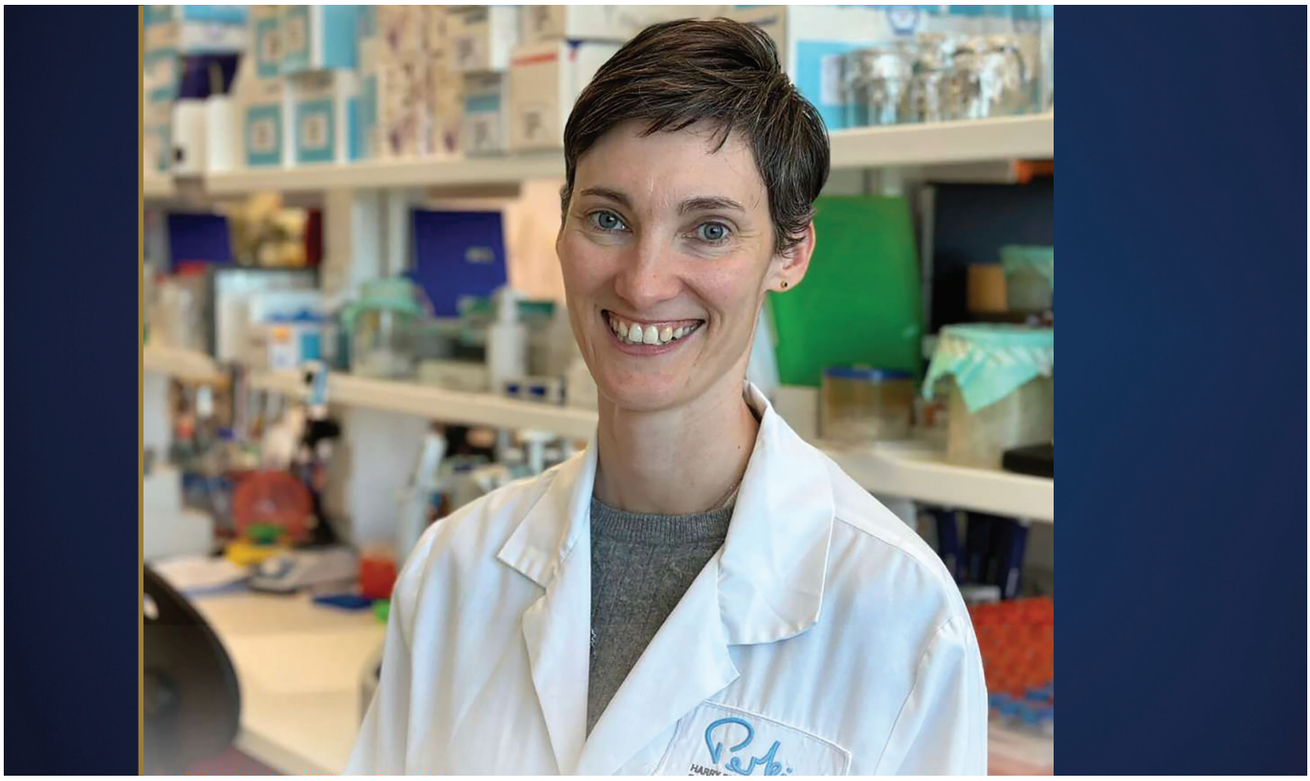
2) designing and testing new treatments for muscle disease using these models.

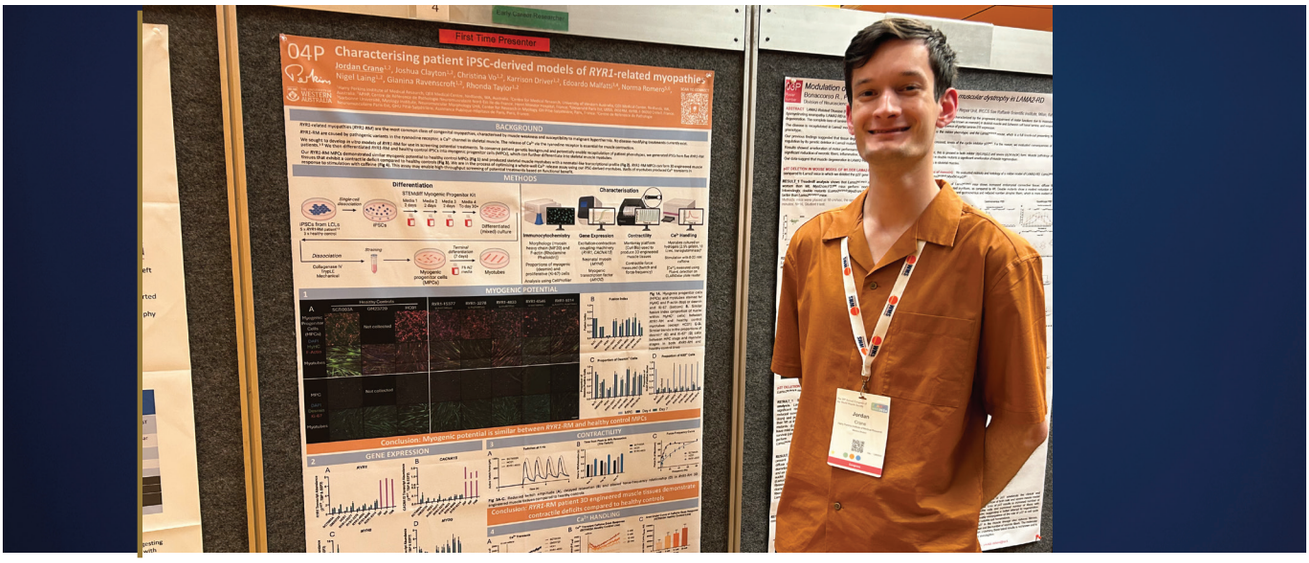
The current funding from the Channel 7 Telethon Trust specifically supports the team to design and test a range of treatments called antisense oligonucleotides (ASOs). ASOs are a class of drug that can target messenger RNA (mRNA). mRNA is a molecular intermediate that enables the DNA code to be translated into a functional protein. ASOs can be designed to target specific mRNAs for removal from the cell, reducing or eliminating the protein that would normally be produced from that mRNA.

Rhonda and her team have designed a range of ASOs to selectively target disease-causing mRNA, but not healthy mRNA copies. This strategy aims to prevent toxic, disease-causing proteins from being made and therefore restore function to the remaining healthy mRNA copies.

We take this opportunity to acknowledge and thank the Channel 7 Telethon Trust, Hearts & Minds Investments Limited, Magellan Group and the Safe Harbour Giving Circle for the generous philanthropic support that has enabled Rhonda and her team to continue this vital research. Thank you so much!

(l-r) Jordan Crane, Dr Rhonda Taylor and Jeremy Garcia from the Disease Models and Therapies team at Harry Perkins Institute of Medical Research. Right; A patient-derived muscle culture (myotubes) treated with an antisense oligonucleotide, demonstrating no cellular toxicity after treatment. Myotubes are stained for myosin (green), actin (red) and nuclei (blue).





"As part of his PhD project, Jordan Crane has designed a range of ASOs targeting a gene that is a common cause of childhood muscle disease. In December 2024, Jordan was able to show that two of these ASOs successfully reduced the abundance of disease-causing mRNA, without significantly reducing levels of the healthy mRNA in patient-derived muscle cells. This was an exciting moment as it was the first evidence of this strategy working in our team. We still have a lot of additional testing to do to ensure that the ASOs we have developed are safe and do not have additional unintended effects in the cell (called 'off-targets'). However, this preliminary result is a great first step and could potentially lead to a life-saving treatment in the future.

With the support of additional team members Jeremy Garcia and Anna Johnstone, the team are now validating these preliminary results with additional patient cell lines and investigating a broad range of additional ASOs targeting multiple muscle disease genes."

- Dr Rhonda Taylor, Safe Harbour Fellow (2024–2026)

A few words from Emeritus Professor Nigel Laing AO

As all of us involved with the Patricia Kailis Fellowship know, one of Patricia's many pioneering endeavours was to invent, in the 1960s, cascade carrier screening to identify which women in Duchenne muscular dystrophy families were carriers. Women who are carriers of Duchenne have a 50 per cent chance that any son will have Duchenne. The carrier screening Patricia invented along, Byron Kakulas enabled couples to avoid having boys affected with Duchenne, if that was what they wanted to do. The carrier screening reduced the incidence of Duchenne muscular dystrophy in Western Australia–a world first.

We have carried on Patricia's work in carrier screening with Mackenzie's Mission, the Australian Reproductive Carrier Screening Project. The major update about Mackenzie's Mission is that the principal research paper resulting from Mackenzie's Mission was published in the extremely prestigious

(l-r) Professor Martin Delatycki, Prof Edwin Kirk, Rachael and Johnathon Cassella (Mackenzie's parents) and their sons, Emeritus Prof Nigel Laing AO

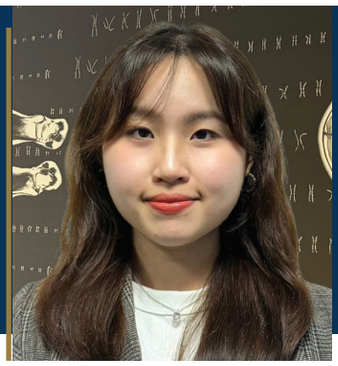
New England Journal of Medicine in November last year. The publication has received wide acclaim around the world. When a scientific journal publishes a paper that it considers more important than others, it sometimes asks another authority in the field of research to write an editorial on the paper. The *New England Journal of Medicine* published an editorial on the Mackenzie's Mission paper by Professor Lidewij Henneman who is based in Amsterdam. Professor Henneman is a world leader in human genetics, including attempts to introduce carrier screening in the Netherlands, and in research into the ethics of human genetics. In her editorial, Professor Henneman wrote, "The overall conclusions can inform a global research agenda for years to come." In other words, Western Australia once again is a pioneer in carrier screening.

Professor Henneman also wrote, "For Australia, the question is whether expanded carrier screening will become accessible at no cost to those who opt to undergo screening". This is true. We have done the research. We showed with Mackenzie's Mission that just under one in 50 of the couples we tested had a 25 per cent chance with every pregnancy of having a child affected with one of the 750 severe recessive diseases we screened for.

What then are the Federal and State Health Departments going to do with this information? What action are they going to take to make government-funded Mackenzie's Mission-level expanded carrier screening available to all Australians? Depressingly, I was reminded earlier this year by a wise person from the Eastern States, that it took 15 years from the pilot study of bowel cancer screening for bowel cancer screening to be implemented as a government funded program and made free to Australians. A similar timeline would mean that it would be 2039 before Mackenzie's Mission carrier screening is government-funded in Australia. This is surely not an acceptable timeline. In those 15 years, thousands of Australian children will be born and suffer from severe recessive diseases.

In a completely different outcome, Medicare rebated carrier screening for the three relatively common recessive genetic diseases cystic fibrosis, fragile X syndrome and spinal muscular atrophy has been free to Australian couples since 1 November 2023. When a Medicare rebate is approved by the Medical Service Advisory Committee (MSAC) and made available, you never know how much the item number rebate is going to be used. For example, Medicare item number 73360, which pays for reanalysis of a patient's genomic data, has been used only 74 times in the whole of Australia since July 2021. On the other hand, usage of Medicare Item Number 73451 for the three-disease carrier screen is consistently running at an annual equivalent of around 120,000 tests a year. Australia has approximately 300,000 births a year. Therefore, usage of the three-disease carrier screen by 120,000 couples a year may be taken to indicate that around 40 per cent of couples having a child in Australia each year are using the free three-disease carrier screening test. This to me indicates a very high appetite for carrier screening amongst Australians who are having children. This makes me confident that in the fullness of time Australian couples will have government-funded Mackenzie's Mission-like carrier screening available to them. I will be doing my best to ensure that it does not take 15 years for it to happen. I hope you will come along for the ride.

My sincere thanks for your support and your kindness and your generosity in helping our teams reach new research heights, year on year."



New PhD student to assist Gina's miscarriage project

"I'm Jasmine Chew, a first-year PhD student in the Rare Disease Genetics and Functional Genomics Group, under the supervision of Professor Gina Ravenscroft. I'm originally from Malaysia and moved to New Zealand for my tertiary education, where I developed a background in medical laboratory science and diagnostic genetics.

Before relocating to Perth to pursue my PhD, I worked as a Medical Laboratory Scientist in Auckland, specializing in chromosomal microarray analysis for constitutional, prenatal and pregnancy loss cases.

Working in this field has ignited my passion for research in perinatal and rare disease genetics, inspiring me to improve genetic diagnoses and make a meaningful impact on families affected by unexplained conditions. My PhD research focuses on utilising advanced genomic technologies, such as optical genome mapping and whole genome sequencing, to better understand the genetic causes behind unexplained recurrent pregnancy loss and rare disorders associated with congenital foetal anomalies. Establishing genetic diagnoses in these patient cohorts has significant clinical implications, including understanding recurrence risk for informed genetic counselling and future reproductive planning.

I'm truly excited and grateful to be part of your community. Being surrounded by researchers, mentors and supporters who share a passion for making a difference is incredibly motivating. I look forward to learning from and contributing to this community that values research excellence and positive outcomes for families."

In January 2025 Gina was interviewed by ABC Perth Mornings Nadia Mitsopoulos. Listen to her interview where she talks about her research goals for recurrent pregnancy loss.

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