Perkins

Magazine of the Harry Perkins Institute of Medical Research

"one day...
I want to know the name of my disease."

Stephen

"one day...
We'll find the answer for Stephen"

Professor Nigel Laing AO

Bringing Tomorrow Closer.

Research needed to combat rare diseases

From the Director

People living with a rare disease, regardless of which specific disease they have, often face significant challenges.

Many rare diseases are difficult to identify, many have few treatments and often there is a lack of appropriate health services available to help. As a result families can feel isolated, under-supported and can risk economic hardship.

While researchers are constantly uncovering more information about rare diseases, the exact cause of many is still unknown leaving families without a diagnosis or information about what to expect as the disease progresses. There is also the concern about the likelihood of passing their disease on if they have children.

'Rare disease' is the term given to diseases that affect fewer than 1 in 10,000 people. While each disease might be considered rare, collectively they affect 8-10% of the Australian population. This is similar to the proportion of people living with diabetes or asthma. With more than 8,000 known rare diseases, the community impact is significant.

The majority of rare diseases are thought to have a genetic cause. Our Neurogenetic Diseases Laboratory at the Perkins, led by Professor Nigel Laing AO, hunt for the disease-causing genes lurking in our DNA to help provide answers for families. Identification of a gene gives families choices before passing the gene on to the next generation. Unfortunately, some can occur randomly without a family history of the disease, which makes diagnosing and treating the disease particularly difficult.

Thanks to advancements in technology and understanding, researchers have made significant progress in learning how to diagnose, treat, and prevent a variety of rare diseases. However, there is still much to be uncovered in this area and more research is needed. The reality is that most rare diseases still have no treatment and many families still don't have answers. This is something our researchers are committed to helping overcome.

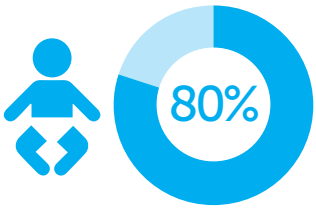
Professor Peter Leedman
Director, Harry Perkins Institute of Medical Research

Source: rarediseasedayaustralia.com.au

ABOUT RARE DISEASES

ONLY **5%** of **RARE DISEASES** HAVE TREATMENTS

RARE DISEASES are responsible for **35% of deaths** in the first year of life.



80% OF RARE DISEASES AFFECT CHILDREN

THERE ARE MORE THAN **8,000** KNOWN RARE DISEASES

Rare diseases affect up to 10% of the Australian population

source: globalgenes.org

one day... we'll find the answer for Stephen

When Stephen Jando was eight-years old, his life was disrupted by a mystery disease. Stephen was a healthy, active kid before he broke his ankle playing basketball at school.

When he was taken to hospital, doctors discovered there were problems with the ligaments and tendons in Stephen's ankles and soon they realised there was a more serious problem with his muscles.

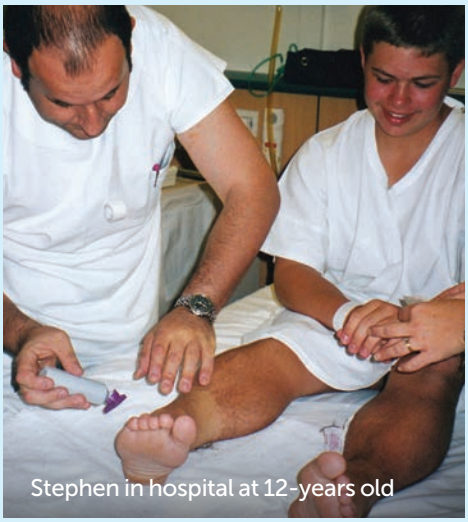
Stephen's muscles were growing out of control, causing debilitating pain and eventually leaving him with difficulties walking, unable to work, or play the sports he once loved.

"The muscles seem to be just growing out of control, they just constantly grow. Since that first hospital visit, I've had fifteen operations on my legs to reduce the size of the muscles," Stephen said.

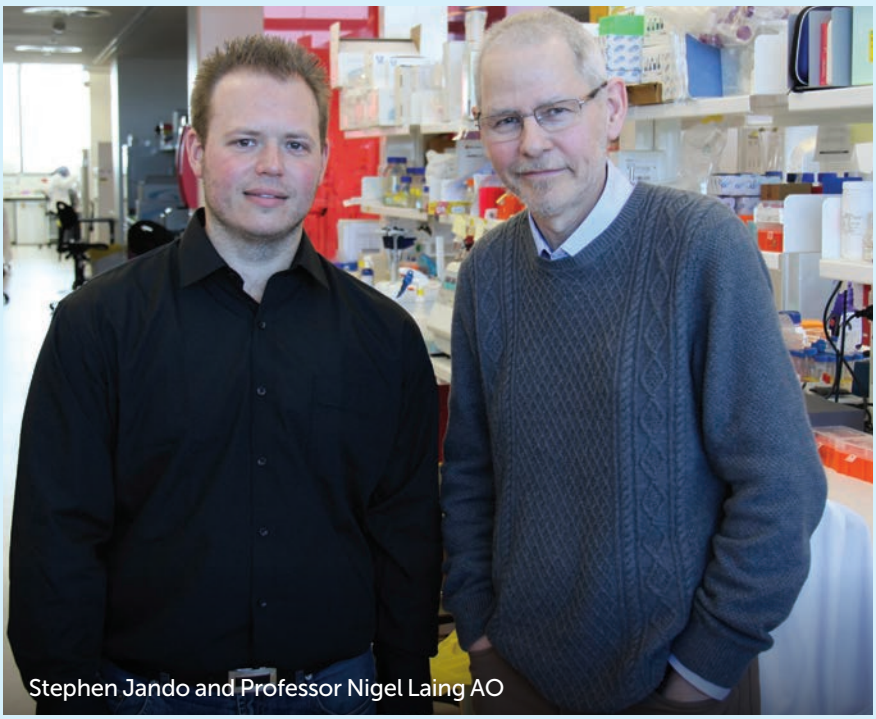
Stephen and his family are desperate for a diagnosis, to give him the ability to plan for the future.

"They've found some sort of muscular disease that they're not certain of what it is or what's caused it," Stephen said.

Stephen hopes that a diagnosis might lead researchers to find a way to treat his disease, which could prevent further surgeries and reduce the constant pain he experiences.



Stephen in hospital at 12-years old



Stephen Jando and Professor Nigel Laing AO

"I can't remember a day without pain anymore, and I probably don't want to remember a day without pain because then I'll know what I'm missing."

Professor Nigel Laing AO, who heads the Perkins Neurogenetic Diseases Laboratory, believes that it's only a matter of time before the cause of Stephen's disease is discovered.

"One day we'll know the answer, we'll know what's causing Stephen's problem. I'm absolutely convinced about that," Professor Laing said. "What I don't know is how long it's going to take."

"Everyone in my group is aimed at trying to find as many disease genes as fast as we can, to help as many people, patients and families as we can."

Professor Laing described the method of hunting for the genes that cause rare diseases, as adding detail to a map.

"We're gradually filling in the whole map of every disease gene that we humans suffer from," Professor Laing said.

"When we look at a patient like Stephen who hasn't got an answer, that's what drives me. If we don't succeed in finding all the genes, then there will still be patients who don't get an answer, ever. So there's a lot of work to do, but one day we'll know the answer."

Watch Stephen tell his moving story online at perkins.org.au

WA's biggest fundraising ride raises $4.1 million

In the six-year history of the MACA Ride to Conquer Cancer, generous West Australians have raised a total $27.5 million for cancer research at the Perkins.

This year 855 riders raised more than $4.1 million by riding 200km in the two-day event held over the weekend of 21-22 October.

"With one in two Australian men and one in three Australian women diagnosed with cancer before the age of 85, the Ride is a remarkable tribute to people who suffer from this terrible disease", Professor Peter Leedman said.

"We are incredibly grateful for the continued support of everyone involved in the Ride. The community of riders, donors, volunteers, and sponsors make a significant difference to our research."

Mandurah rider, Darren Hospital, who rides with team Woodside, the second largest team in the event said his family and friends had inspired him to take part.

"My motivation for doing the Ride is the memory of my sister Lisa who recently passed away from ovarian cancer at the age of 47, my father John who passed away from prostate cancer at the age of 49 and my best mate Adam Dean who recently passed away from bowel cancer at the age of 49," Darren said.

Title sponsor MACA, and their 200-strong team rode with 'Frosty' armbands in honour of team member Daniel Frost, who at the age of 33 died of bowel cancer in March.

"Our employees, our community of friends and family members, have all been personally impacted by cancer and it is a continued honour to support the Perkins advancements in medical research in order to better the lives of cancer patients and support their mission to conquer cancer," said MACA Director Geoff Baker.

New drug to supercharge immune cells in the fight against cancer

Perkins researchers have developed a new cancer treatment with the dual ability to normalise out-of-control tumour blood vessels and boost the body's immune system.

Tumours can become resistant to the body's immune system by creating a barrier of tangled blood vessels that feed the cancer while locking out immune cells.

Professor Ruth Ganss, head of the Perkins Cancer and Cell Biology Division, said the new treatment worked by generating more "normal" blood vessels and lymph-node-like structures within the cancer, which together enabled immune cells to better reach the cancer core.

"Our drug strengthens the immune response against tumours by inducing these lymph-node-structures together with normalised blood vessels, producing immune cells that infiltrate deep into the cancer. There are currently no single treatments available which can produce these two features in cancers."

"Our research shows that once our drug has triggered the lymph-node-structures within the cancer, current immunotherapies that have been approved for clinical use, can work more effectively," Professor Ganss said.

"We envision that a combination of our drug and existing immunotherapies, will greatly enhance the outcomes for patients in the future."

Funds from the MACA Ride to Conquer Cancer support the vital work of Professor Ruth Ganss.

Kathy's legacy to create tsunami of hope

Kathy Bunce had been busy training for her third MACA Ride to Conquer Cancer when she began feeling unwell in July this year.

Eight weeks later, on Sepember 8th, a very rare and very lethal cancer took the life of the beloved 48-year-old mother, wife, sister and friend.

Before being diagnosed, Kathy had hoped to personally raise $50,000 for cancer research at the Perkins and rally together 50 riders to join her team FiTi50 and take on the 200km challenge.

FiTi50 went on to become the top community team, with 32 riders raising more than $135,000 for the Perkins and Kathy herself raising more than $30,000 for cancer research.

Fellow riders, friends and colleagues of Kathy's held posters in her honour at the event, and Kathy was recognised by Perkins Director, Professor Peter Leedman, as a driving force for the event.

Kathy's younger sister Suzie said that Kathy had been determined during her illness, telling family that although she might not win her fight against cancer, by supporting new research, others would.

"Even as she got sicker and lost her hair and her body was subjected to indescribable pain, Kathy had a steadfast belief that research would help find answers, just not in time to help her. But she was confident that, in the future, others might survive when she did not," Suzie said.

Despite being in hospital, Kathy's dedication to the Ride and the cause never wavered.

"She'd be outside Cancer Ward G73, on a stationary exercise bike between chemotherapy treatments, clocking up the kilometres," Suzie said.

Kathy underwent two rounds of intense chemotherapy and radiotherapy, but neither were successful in stopping the cancer. Tragically, the cancer that ultimately took her life was so rare that there were no targeted treatments to help.

Kathy (third from left) and her three sisters

> "Kathy was a natural leader and one of those people who rolled up her sleeves and got things done and inspired others to do the same. Kath didn't believe in making ripples, she believed in starting a tsunami."

"Kathy had been inspired to join her first ride in 2015 in honour of our mother who passed away in June 2013 from breast cancer. Research had improved treatment options for breast cancer and mum had been able to live cancer free for 10 years before it came back. Participating in the Ride strengthened Kathy's passion to advocate for cancer research," Suzie said.

"Kathy was a natural leader and one of those people who rolled up her sleeves and got things done and inspired others to do the same. Kath didn't believe in making ripples, she believed in starting a tsunami."

"My sister said at the end of her life that she had no regrets, but that she had more to do and that was to help others in cancer treatment believe that, with research, there's hope."

"Kathy would be so happy to know she has inspired others to act. I'm so proud of her legacy."

You can help Kathy realise her goal to raise $50,000 for cancer research.

Look out for our Christmas appeal featuring Kathy and her brave fight to help the Perkins.

Perkins trials facility leading the way towards better health

Breakthrough treatments and innovative new technologies are helping Linear Clinical Research deliver better health to the community.

Linear CEO, Dr Michael Winlo said the Perkins-owned Phase 1 clinical trials facility was world renowned as a cutting-edge hub for local and international trials of promising new treatments.

"It's a little-known fact that a Perth-based clinical research team is playing a major role on the world stage," Dr Winlo said.

Linear is currently testing a broad range of new medicines to tackle some of the major diseases that affect the community. The trials feature more than 20 new cancer medicines, including immunotherapy drugs, and new therapies for multiple sclerosis – a condition with few effective treatments.

New treatments for joint disease, osteoarthritis, cystic fibrosis, pulmonary fibrosis and even methamphetamine addiction are being trialled at the Nedlands facility.

Dr Winlo said Linear is at the forefront, rolling out new state-of-the-art technology that will enhance data capture and trial efficiency, to improve insights for biotech companies and help bring life-saving drugs to patients sooner.

"Linear is committed to being one of the world's most patient-centric and innovative clinical trial sites".

"By embracing new technology, we not only attract more therapies and trials to Perth but also greatly accelerate the speed at which beneficial therapies get to patients," Dr Winlo said.

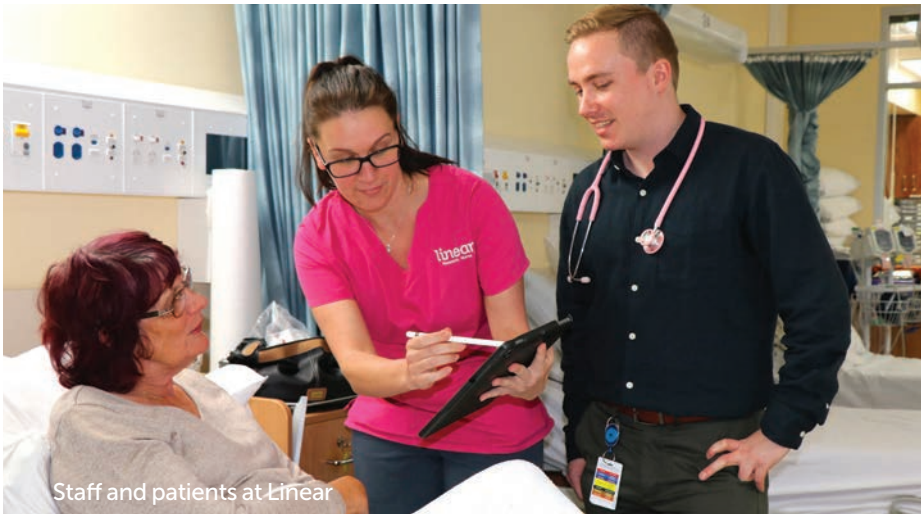
One of the drugs soon to be trialled is a new treatment for chronic kidney disease, developed by Perkins researchers and their collaborators.

"It marks a unique milestone in the WA biotech scene whereby a locally discovered breakthrough, will also commence a crucial trial in Perth's most specialised clinical trials unit," he said.

The drug, called DMX-200, has the potential to revolutionise treatment for chronic kidney disease, a condition which affects as many as one in 10 adult Australians, and could significantly reduce the need for dialysis and kidney transplants.

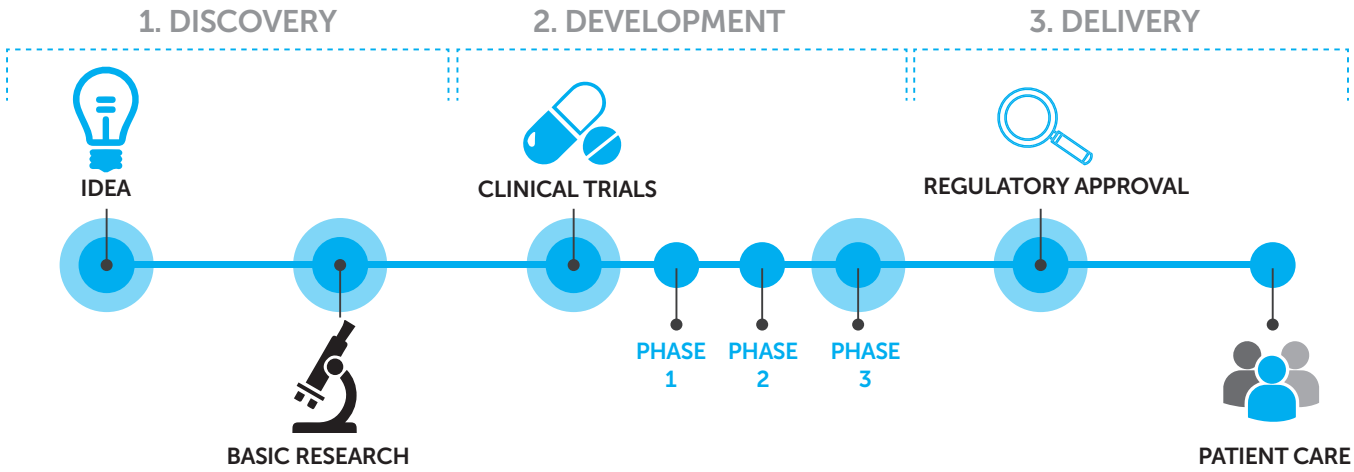
"While Linear runs a lot of trials with patients seeking access to world-first treatments, we also support trials involving young healthy individuals who are prepared to give up their time to help evaluate the effects of a brand new drug," Dr Winlo said.

"My aspiration is that one day we will be able to bring a potentially beneficial trial to any patient with an advanced stage of a disease to effectively improve their condition and give them a better quality of life."



Staff and patients at Linear

BENCH TO BEDSIDE



Engineering the health care of tomorrow

A new class of engineers are joining forces with scientists and surgeons to design tools that could revolutionise the way we treat disease.

The Biomedical Engineering @ Perkins program was launched in 2016, bringing together electrical, mechanical, chemical and biomedical engineers to design and develop innovative solutions for important medical problems.

Dr Brendan Kennedy, who leads the Bioimaging Research and Innovation for Translational Engineering Laboratory (BRITElab), says the 16-strong team of engineers, split across two laboratories, hope to offer practical solutions for doctors and patients.

"We're looking at healthcare programs from an engineering stand point, you need to match the problem with the solution and we've been developing new tools that could be more effective than existing options," Dr Kennedy said.

The BRITElab team made international news with their progress towards the development of a small probe that could be worn on the end of a surgeon's finger during operations. The device helps ensure no microscopic bits of cancer are left during breast cancer removal surgery, which could regrow if left in the body. The device is soon to be tested in clinical trials.

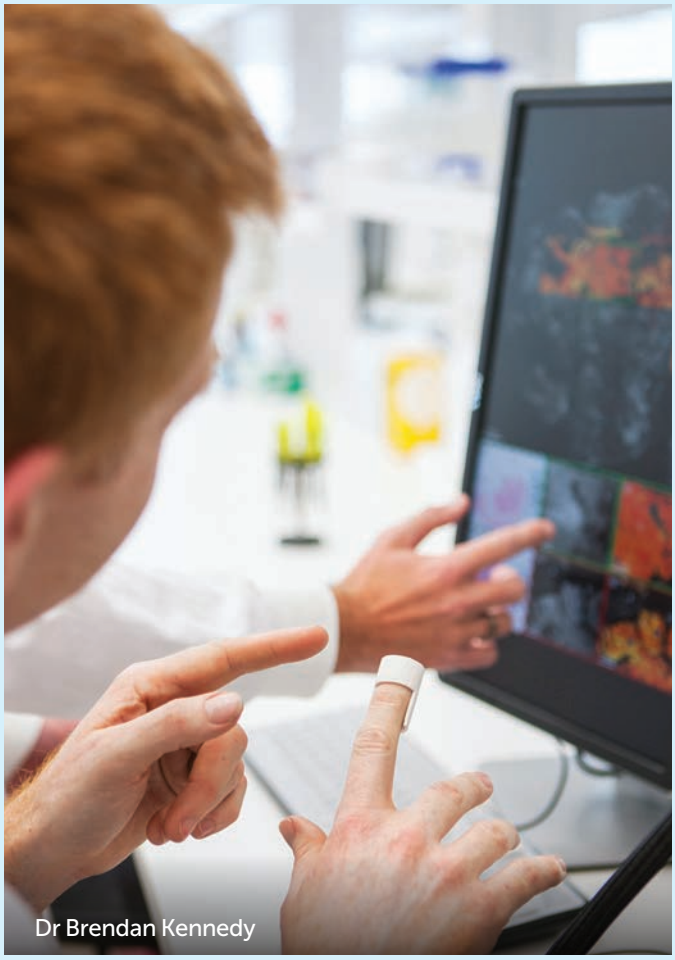
While Dr Kennedy and his team are focused on finding solutions to benefit breast cancer patients, the other half of the Biomedical Engineering @ Perkins program is looking for ways to combat cardiovascular disease, the world's biggest killer, which takes one life every 12 minutes in Australia.

The Vascular Engineering Laboratory (VascLab), headed by Dr Barry Doyle, says some of their current research projects involve aneurysm rupture risk assessment, coronary artery disease, type B aortic dissection, placenta biomechanics and using 3D bioprinting to create new medical devices.

Dr Doyle said the aneurysm rupture project was helping researchers predict the exact location that an aneurysm will rupture.

"We do this by combining CT and MRI scans to create highly accurate 3D reconstructions of the aneurysm and applying a solid mechanics approach to determine the stresses in the vessel. We are currently testing this approach in a study of 350 patients with aneurysms," Dr Doyle said.

"Once we optimise the system to correctly predict the likelihood of rupture in 100 per cent of cases we hope to do a worldwide trial involving thousands of patients across many research centres before the system can be rolled out in hospitals. We are now attempting a similar project with aortic dissection, which is a particularly challenging condition for surgeons."



Dr Brendan Kennedy

"No matter what disease we are examining, we try to use as much patient-specific detail as possible to determine the risk profile," Dr Doyle says. "For patients with aneurysms, this means we calculate the likelihood of their aneurysm rupturing, and not based on the likelihood of the general population."

"That is the beauty of what engineering can bring into medicine, it takes away any ambiguities and it focuses right down on that specific patient, helping to make better informed clinical decisions."

The Biomedical Engineering teams aren't limited in their scope – other projects include the use of soft materials with cells for bioprinting applications. This enables both a better understanding of cell behaviour in three dimensions, as well as the possibility of 3D bioprinting new devices.

"If you wanted to print a living structure with several different materials and multiple cell types, each material often requires its own engineering specification to work with," Dr Doyle says. "It is a huge interdisciplinary challenge for engineers and material scientists, but one we are actively working on."

Team Blister Sisters in the 2017 Walk

Family take Hawaiian Walk in their stride

The Hawaiian Walk for Women's Cancer is a family affair for Maria Clark and her daughter-in-law, Amanda Clark.

The women will be taking on the one day, 30km or 42km challenge for the second year in a row on Saturday 5 May 2018.

Inspired by their efforts, Amanda's three children Seth, Tia and Nadine have signed up to join them in 2018.

Maria said she registered to take part after being diagnosed with breast cancer and undergoing a double mastectomy and reconstruction.

"I am walking because I am a breast cancer survivor, I am able to walk and able to raise awareness along with much needed funds to find a cure," Maria said.

"My Mum has had breast cancer twice. In February this year a beautiful person, Lisa, whom I had the privilege to work with passed away from cancer. A couple of days after this year's walk my daughter-in-law lost her sister at age 38 from cancer."

"So many of us have been touched by this terrible disease. The impact is not only on the person who has it but their families and friends too."

"I walk for those who are currently fighting, for the memory of those who have not survived and in honour of those who have survived."

Amanda said she didn't hesitate in signing up to walk 30kms again in support of cancer research.

"I'll be walking along side an incredible bunch of women in my team 'blister sisters', which has gone from 6 to 17 team members in just one year," Amanda said.

"I knew I wanted to do more to help the Perkins. My family alone has seen more cancer diagnoses than we should have. My sister lost her fight with pancreatic cancer in May and I have lost my dad, Nana and cousin to cancer just recently, with more family members still fighting the disease."

"There are too many lives affected and every little bit helps," Amanda said.

Maria's husband and Amanda's father-in-law, Kevin, also supports Perkins research, having recently ridden in the MACA Ride to Conquer Cancer and raising over $10,000 for cancer research.

Register now at walkforwomenscancer.org.au

hawaiian
WALK FOR WOMEN'S CANCER
Perkins

Doctor to journey from Perth to Patagonia

Mosman Park GP, Dr Jamie Prendiville, is on a mission to push himself to his limits and raise funds for melanoma research.

Dr Prendiville, along with his son James Prendiville, and friends Matthew James, Scott Murphy and Jon Kissick will be competing in a 250km, 7-day ultra-endurance race across Patagonia.

The group will cross the lower end of the Andes Mountains in the intensity challenge, with Dr Prendiville and the team also setting a formidable goal to raise $50,000 for the Kirkbride Melanoma Centre.

The Kirkbride Melanoma Centre aims to improve the diagnosis and treatment of melanoma - a disease commonly referred to as Australia's cancer.

Scott Kirkbride was a patient of Dr Prendiville's and after he passed away from malignant melanoma at the age of 27, the centre was set up in his honour.

"My family and I have been raising donations for the Kirkbride Melanoma Centre since 2005, including swimming the English Channel and undertaking several ultramarathon races in various deserts across the world," Dr Prendiville said.

"We are also focused on melanoma awareness and intend to run events to promote melanoma awareness in younger, 20-40 year old, people later this year."

"Recently, we have funded a Prendiville Family Scholarship which supplies top-up scholarships for PhD candidates to undertake melanoma research at the Perkins," Dr Prendiville said.

"The endurance marathon will be hard, but it's nothing compared to what people with cancer go through."

Dr Jamie Prendiville

Australia and New Zealand have the highest melanoma rates in the world, with melanoma killing more young Australians aged 20 to 39 years than any other single cancer.

Research is turning the tide on melanoma, with new, targeted treatments extending the life expectancy for people with advanced disease - but with 14,000 Australians expected to be diagnosed with melanoma in 2017, further research is vital to save lives.

Learn more or donate at: https://perth-to-patagonia.everydayhero.com/au/team-perth-to-patagonia

How you can help

1. DONATE

You can help bring the medical miracles of tomorrow closer by donating directly to the Perkins.

2. FUNDRAISE

Challenge yourself to run, cycle, walk, climb or even shave your hair to make a difference to people facing illness and disease.

3. SPREAD THE WORD

Help raise awareness for ground breaking WA research by sharing Perkins news with friends and family

New way to defend against Type 2 Diabetes

Researchers from the Perkins Centre for Diabetes Research have identified a substance that protects cells from triggering Type 2 Diabetes.

Type 2 Diabetes affects 1.4 million Australians and is caused when insulin-producing cells stop functioning properly.

Diabetes researcher, UWA Associate Professor Fang-Xu Jiang, said it had been established that people who develop Type 2 Diabetes often have low levels of vitamin D3.

Numerous international clinical trials have tested the benefits of supplementing vitamin D3 in pre-diabetic patients without success, and the underlying mechanism that leads to Type 2 Diabetes is still unclear.

"Our research team began searching for potential drugs that could act like vitamin D3 and restore the function of failing insulin producing cells," Associate Professor Jiang said.

Perkins researcher, Abraham Neelankal John, said the study ultimately discovered a substance that protects insulin-producing cells from shutting down and leading to diabetes.

"Our investigation uncovered a compound that appears to prevent the onset and delay the progression of pre-diabetes and early diabetes," Mr John said.


Abraham Neelankal John

"More research is needed but if proven in clinical trials, it would certainly become a drug for people at risk of developing Type 2 Diabetes or in the early stages of Type 2 Diabetes. Our ultimate aim is to regenerate the health of people who suffer from Type 2 Diabetes."

Faulty gene linked to obesity in adults

Groundbreaking new research linking obesity and metabolic dysfunction to a problem in the energy generators in cells has been published by researchers from the Perkins Mitochondrial Medicine and Biology Laboratory.


Professor Aleksandra Filipovska

The researchers found that reduced function of a gene that impacts the breakdown of fats resulted in adult-onset obesity and fatty liver.

Laboratory head, Professor Aleksandra Filipovska, said the study provided new knowledge that would enable the development of specific drugs and treatments to overcome the impact of obesity on normal body function.

"Our new model is helping us test drugs that can lessen the burden of obesity and fatty liver disease," Professor Filipovska said.

Lead researcher Kara Perks said that in normal metabolism, fat and carbohydrate in our diets was broken down to produce the energy that our bodies required by the cellular energy plants, known as mitochondria.

"We investigated the impact of mitochondrial dysfunction, in relation to a specific gene known as PTCD1, required for energy production and the breakdown of fats and carbohydrates."

"Healthy adults will have two copies of the PTCD1 gene, but we looked at what happens when there is only one copy, and we learned that PTCD1 is vital for the breakdown of fats and energy production. When one copy of this gene is lost, it results in obesity, fatty liver and ultimately heart disease."

Excess weight and obesity are major risk factors for cardiovascular disease, diabetes, some musculoskeletal disease and cancers. The prevalence of obesity among Australians and most western societies has been increasing for the past 30 years, and this research offers useful insight into one genetic cause of obesity.

BioDiscovery Centre – one of Australia's best

A NSW student on the hunt for an authentic science experience travelled to Nedlands to take part in a unique program at the Lotterywest BioDiscovery Centre.

Based at the Perkins, the BioDiscovery Centre is a specialised state-of-the-art teaching laboratory where medical researchers and science teachers offer hands-on activities to the community.

Sophie Bailey (16) travelled from the Sydney suburb of Clontarf to Perth, for the three-day immersion program, an engaging workshop for high school students to get a feel for the life of a medical researcher.

Sophie said she found the program online after seeing the Perkins on the news.

"I had been looking for over a year in Sydney for a program that let you work in the same environment as the research scientists, but I really struggled to find a program that focused on the practical side," Sophie said.

"When I read about how you could work in an institute that is leading in medical research I knew it was an opportunity I couldn't pass up."

Sophie encouraged others to learn more about the BioDiscovery activities available.

"It really offers an experience that is hard to find anywhere else in Australia."

Learn more at biodiscovery.org.au

Sophie Bailey and her mum Sandy.

Lotterywest
BioDiscovery
Centre

Corporate Christmas Party Packages

Package 1 - During School Hours

Get back into school uniform and join a science class for the afternoon. Look 'too cool for school' in your lab coat.

Help the boffins at the Harry Perkins Institute of Medical Research with their experiments.

Time: 3 hours **Cost:** $100 per head

Packed lunch and juice box included

Package 2 - After Dark

Sneak into the lab after dark and become a researcher for the night! Put on a lab coat, conduct experiments and have a private tour of the Perkins.

Create a signature DNA cocktail and enjoy some science inspired finger food.

Time: 3 hours **Cost:** $150 per head

Finger food & drinks package included

Packages can be tailored to your needs - Please contact 08 6151 0811 education@perkins.org.au

Thank you to all our supporters

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woodside

Ashok and Renu Kumar and family

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AUSTRALIAN CANCER RESEARCH FOUNDATION

Add these dates to your calendar!

Learn more at perkins.org.au/events

Wesfarmers' Harry Perkins Oration
– Tuesday 28 November 2017

Hawaiian Walk for Women's Cancer
– Saturday 5 May 2018

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All contributions to the Perkins go directly to advancing medical research. If you would like to support a specific area of research, please tell us.

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Donations over $2 are tax deductible and a receipt will be issued.

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1. perkins.org.au (click "Donate Here")
2. Phone (08) 6151 0700
3. Post this form to the Harry Perkins Institute of Medical Research PO Box 7214 Shenton Park WA 6008 or fax to (08) 6151 0701

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