



Magazine of the Harry Perkins
Institute of Medical Research

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"one day..."

I'm a young mother and a teacher.
The next minute I'm on the floor
crying after hearing the words that
I had triple negative breast cancer."

Natalie



"one day..."

We're going to defeat breast
cancer. I lead a team at the
Perkins who are convinced we
will find a treatment that is much
better and less damaging than
chemotherapy."

Associate Professor Pilar Blancafort



Bringing
Tomorrow
Closer.

"one day..."

I hope to be able to look my patients in the eye and say I have the answer. One day I hope to be able to tell people with a diagnosis of cancer that we have the treatment that will stop their cancer in its tracks.



From the Director

As a doctor, as head of the Laboratory for Cancer Medicine here at the Perkins and as the Director, I have many "one day's".

All of my colleagues at the Perkins have their own one day hopes and dreams that they are relentlessly working to make a reality – for people with heart disease, with diabetes, with neurogenetic disease and with the hard to treat cancers.

Our one day dreams are the reason we come to work every day. We know that if we're good enough, one day we will find the answers we have spent our entire professional lives searching for ... but we aren't doing it alone.

This month, almost one thousand cyclists helped to make their one day dreams come closer when they raised \$4 million for our research by participating in the MACA Ride to Conquer Cancer.

The dedication and commitment of our long term partner MACA and every rider, crew member and supporter who contributed to this wonderful result were an inspiration.

Here at the Perkins we have world-class facilities and teams of internationally renowned, highly skilled researchers and doctors. We are uniquely positioned to fast track the development of new treatments, quicker diagnosis methods and tests that enable doctors to select the best approach for each patient. We believe that one day with the support of our community and corporate partners we'll be able provide better health outcomes for everyone.

Help bring one day closer by continuing your support of the Perkins in the new year.

Professor Peter Leedman

Director, Harry Perkins Institute of Medical Research

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Stan Perron
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Thank you to everyone who has supported the Perkins this year.

one day... that changed my world

Natalie Mathews was 43 years old with no noticeable lump and no family history of breast cancer when her GP sent her for a precautionary mammogram.

Before the mammogram, Natalie's days were spent as a busy secondary school teacher, a wife and a mother of two young boys.

"I was called with the test results in the middle of a maths class. From the caller's tone of voice, I knew right away that something was terribly wrong and my whole world shattered," Natalie said.

"I was told I had grade 3 triple negative breast cancer. It's not hormone driven, it's this rogue breast cancer."

The tumour was too big to remove, so Natalie underwent chemotherapy. Her oncologist told her there was only a 50/50 chance the triple negative cancer would respond to the anti-cancer drugs, as it is notoriously difficult to treat.

"I was on intense chemotherapy for six months, it was tough but we could see the tumour was shrinking so we stuck with that."

While recovering, Natalie spent her days sleeping – a stark change from the busy, active woman she had been.

"Fear comes up when you feel ill. Fear comes up when you have an ache or pain. It can stop you from living, so you have to forget that and try to be positive and proactive."

"Now my husband and I keep a big eye on research. We're very passionate and interested in how medical research is moving towards better treatments, prevention and earlier diagnosis."

Natalie recently visited the Perkins and met researchers including Dr Benjamin Garcia-Bloj and Dr Anabel Sorolla, both of whom are making strides towards improved treatments for triple negative breast cancer.

"I have joined support groups to give back to other women and I've signed myself and a big group of friends up for the Hawaiian Walk for Women's Cancer."

"I went through this awful experience, but I'll make sure it wasn't for nothing. I'm devoted to doing everything I can to help others."



Natalie and her son in the Lotterywest BioDiscovery Centre



Natalie and triple negative breast cancer researchers at the Perkins

You can make a difference too by joining Natalie in the Hawaiian Walk for Women's Cancer on 6 May 2017.

Learn more at walkforwomenscancer.org.au

Natalie tells her moving story in a video alongside Perkins Laboratory Head, Associate Professor Pilar Blancafort, whose team is focussed on finding better treatments for triple negative breast cancer. **Watch the video now at perkins.org.au**



MACA Ride to Conquer Cancer raises \$4 million

A dedicated group of 905 riders raised an impressive \$4 million to support ground breaking cancer research at the Perkins, through the 2016 MACA Ride to Conquer Cancer.

Perkins Director, Professor Peter Leedman extended his thanks to every rider, crew member and supporter who helped make the MACA Ride to Conquer Cancer such an inspiring success.

Professor Leedman particularly thanked title sponsor, MACA, for their extraordinary commitment to the Perkins.

"This year MACA have raised \$1.2 million, bringing the total raised during their five year partnership with the Perkins to over \$6.6 million," Professor Leedman said.

Professor Leedman also thanked Team Woodside, whose fundraising through the Ride over the past five years has totalled \$1 million.

"It's not only the big companies, it's all the individual people whose lives have been touched by cancer and who decide to make a difference," Professor Leedman said.

"Each year I ride alongside so many inspiring and courageous people who have battled cancer or who have lost children, or mothers, or siblings to this insidious disease."

Five-time rider Sue Kingsley said her diagnosis with terminal breast cancer had strengthened her desire to give back.

"I ride because it makes me feel like part of a huge community and when I'm out there I don't think that I have a shortened life, I think how much I can do for others," Sue said.

Sue, who works at St John Ambulance, trains for the Ride between rounds of chemotherapy and long shifts at work.



In five years of fundraising for the event Sue has personally raised over \$17,000 for cancer research at the Perkins.

Funds raised through the Ride directly support innovative cancer research projects, specialist new equipment and ensure the best and brightest medical researchers remain in Western Australia.

Each year the two-day, 200km Ride brings together communities of survivors, cyclists and supporters with one common goal to make a difference.



Sue Kingsley taking part in the opening ceremony at the MACA Ride to Conquer Cancer

Perkins researchers turn on cancer killing genes

Dr Benjamin Garcia-Bloj, along with his colleagues from the Perkins Cancer Epigenetics Laboratory headed by Associate Professor Pilar Blancafort, have used an exciting new technology to successfully switch on dormant tumour suppressor genes in multiple cancer types.

The researchers used a gene-editing technology called CRISPR to 'wake up' key tumour suppressors to destroy the cancer from within.

In everyday life people often develop potentially cancerous cells, but there are systems that very efficiently recognize DNA damage and either repair it or self-destroy the problematic cell.

"Tumour suppressor genes are like police in the cell, they check that the cell doesn't have any damage or mutations and isn't growing uncontrollably," Dr Garcia-Bloj said.

"If the cell becomes badly damaged, the tumour suppressor gene will trigger a process to destroy the cell."

Dr Garcia-Bloj said when these processes fail and a cancer takes hold, certain anti-cancer genes can be switched off but they are still present inside the cell. We call the genes in 'off mode' silent or dormant genes.

"Scientists have used CRISPR technology to activate dormant genes before, but our team was able to boost the



Dr Benjamin Garcia-Bloj

genes 22,000-fold and stop the cancer growing in vitro," Dr Garcia-Bloj said.

"We tested the system on breast cancer, lung cancer and gastric cancer cells. Now we know we can strongly activate these genes and it has a big impact in cancer cells."

"This could be used for other repair genes that are dormant, you could reactivate the genes to start repairing other parts of the body."

Genes provide clue to frog's origin

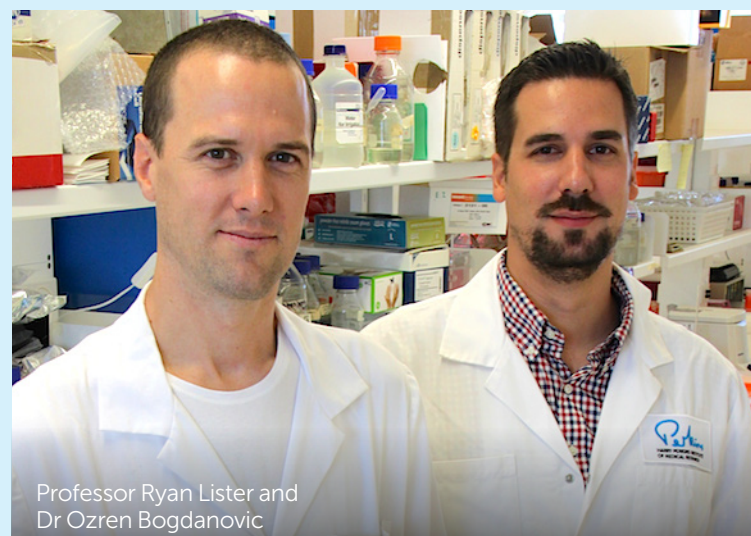
An international team of scientists, including researchers from the Perkins, have decoded the genetic sequence of the African clawed frog, an important model system for cell and developmental biology, and immunology.

The study, published in the journal *Nature*, revealed that the frog arose from the mating of two species, resulting in a "duplicated" genome. The frog carries genetic material from both species and, interestingly, certain portions of the "duplicated" genome appear to be evolving at different rates.

Dr Ozren Bogdanovic and Professor Ryan Lister from the Perkins Epigenetics and Genomics Laboratory contributed to the project by mapping the frog's epigenome.

The researchers mapped the precise genomic locations of an important biochemical signal, called DNA methylation, known as tiny chemical signposts that have the ability to switch genes "on" and "off" during embryo development and disease formation.

The study shows that DNA methylation played one of the most important roles in fine-tuning the levels of gene products



Professor Ryan Lister and Dr Ozren Bogdanovic

making sure that proteins are produced at the correct levels despite the duplication of the genome sequence.

"This is a very important step towards our understanding of how genomes evolve and how genetics and epigenetics shape life on earth," Dr Bogdanovic said.

Sudden infant cardiac death explained by gene mutation

Perkins researchers have contributed to an international project that linked a faulty gene to the deaths of seemingly healthy children whose hearts suddenly stopped.

Last year the Perkins Neurogenetic Diseases Laboratory received DNA from a family in Scotland whose four-month-old baby had tragically suffered a cardiac arrest and died.

With the family's permission, their doctor sent the baby's DNA to the Perkins laboratory and noted that the family was expecting a second child.

Perkins researcher, Dr Gina Ravenscroft, said the team analysed the child's DNA to find possible disease-causing genes.

"We found two genetic variants in a gene called *PPA2* that could be the cause of this disease, but we didn't have any other patients with variants in that gene," Dr Ravenscroft said.

Dr Ravenscroft entered the gene into a research database and found no other researchers had identified it as a disease gene.

Months later the Perkins team were contacted by genetics researchers at the French Institute of Health and Medical Research (INSERM) in Paris. The INSERM team knew of two families with similar cardiac death and mutations in *PPA2*.

The collaborators have been able to quickly translate their findings into a genetic screening test for future babies that might face this silent disease risk.

"The family we received the initial DNA from had their second baby and recently we screened the baby's DNA and found he only has one of the gene variants. To get the disease you need two copies and he's only carrying one so he won't get the disease," Dr Ravenscroft said.

"The screening test has also helped a family who lost two sons from sudden cardiac arrest linked to *PPA2*. Their two other children were screened and found to have two copies of the *PPA2* mutation, so they have been fitted with implantable defibrillators. If their heart stops the defibrillator will support their heart function."

Head of the Perkins Neurogenetic Diseases Laboratory, Professor Nigel Laing AO, said his research team is dedicated to finding answers for families.

“Our job is to find as many disease genes as we can, as rapidly as possible. We're filling in the map of known disease genes and this is another step in the right direction.”

Professor Nigel Laing AO



Professor Nigel Laing AO
and Dr Gina Ravenscroft

Perkins clinical research facility wins Industry Export Award

Linear Clinical Research, the clinical trials arm of the Harry Perkins Institute of Medical Research, has won a WA industry export award for the fourth consecutive year.

Linear was established six years ago, and since 2012 has consistently won government awards in the Business Services or Health and Biotechnology categories.

Linear CEO, Dr Michael Winlo, said he was delighted that Linear had been recognised with a *Business Services Export Award* but noted that Linear was more than a commercial venture.

"Linear works closely with local physicians to run clinical trials for therapies developed around the world," Dr Winlo said.

"The real motivation was to ensure that local patients could get access to new and innovative therapies that wouldn't otherwise be available."

Perkins Director and Chairman of Linear, Professor Peter Leadman, congratulated the team for the achievement.

"It is a credit to Dr Winlo and the whole team for the high quality that Linear delivers to the sector," Professor Leadman said.

linear

Learn more about Linear at linear.org.au

Proteomics International, which is housed at the Perkins headquarters in Nedlands, won the top honour *Exporter of the Year*.

The Australian Export Awards is a national program that recognises and honours Australian companies engaged in international business who have achieved sustainable growth through innovation and commitment.



Eureka Prize winners

A major international project, led by Perkins Professor Alistair Forrest, won a 2016 Australian Museum Eureka Prize.

The 2016 *Scopus Eureka Prize for Excellence in International Scientific Collaboration* was awarded at a gala dinner in Sydney in recognition of the FANTOM5 project.

The project involves researchers systematically examining the sets of genes used in most cell types of the human body and the data from FANTOM5 is already being used to identify genes involved in genetic diseases and to reprogram cells for regenerative and personalised medicine in the future.

Professor Forrest's work is funded in part by a Cancer Research Trust fellowship and the MACA Ride to Conquer Cancer.



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WALK FOR WOMEN'S CANCER

Saturday
6 May 2017



register now: walkforwomenscancer.org.au



Together we can make a significant difference for women facing breast and ovarian cancer by supporting vital research at the Harry Perkins Institute of Medical Research.

#WhyIWalkPerth



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