

Under the microscope

NEWS FROM INSIDE THE INSTITUTE FOR OUR SUPPORTERS



AUTUMN EDITION 2015



Single Cell Power Professor Patrick Tam

A tremendous Thank You to everyone who contributed to our Christmas Appeal and helped us raise \$67,677 towards the purchase of a Single Cell Sorting Machine. This machine is a powerful new technology that will allow us to study the genes at work in a single cell, helping us pinpoint exactly where errors occur and how they impact on the cellular functions that cause abnormal development. For clinicians – and families affected by rare diseases – this is vital information.

This machine is a major priority for the Institute and we are working hard to raise the remaining funds. Overseas research using this technology is progressing very quickly. CMRI is at the forefront of worldwide embryology research; this equipment will help keep us there. *Thank you again for your contribution.*

Healthier kids, brighter futures



From the Director
Welcome to the Autumn 2015 Edition of "Under the Microscope". CMRI's recent research achievements are too numerous to cover in detail, but I hope you will find the brief summary in this newsletter both interesting and exciting. There have been many "behind-the-scenes" developments which are substantially enhancing our research capabilities. CMRI's Biomedical Proteomics facility, which underpins our research programs developing promising new treatments for cancer and epilepsy, has now moved into its new custom-built home on level 6 of our new building. The facility is attracting international interest, and later this year several scientists from Europe and other parts of the world are coming to a conference at CMRI that will help highlight the facility's cutting-edge technology. Another new research technology centre we are setting up is a 'Vector and Genome Engineering' facility, which will assist and speed up the work of all of our research teams and especially our gene therapy research. This newsletter also contains a Q&A with Associate Professor Robyn Jamieson, who is currently undertaking pioneering work on inherited blindness, and some striking photographs taken by our researchers. I hope you enjoy these insights into what is happening at CMRI.

Best Wishes,

Professor Roger Reddel

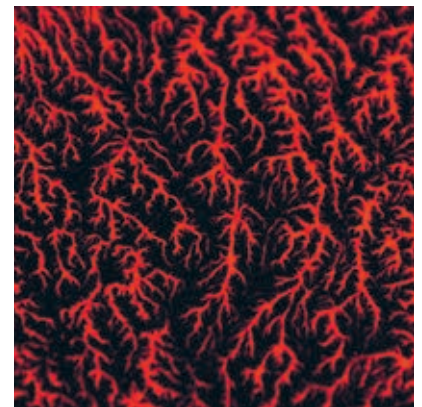
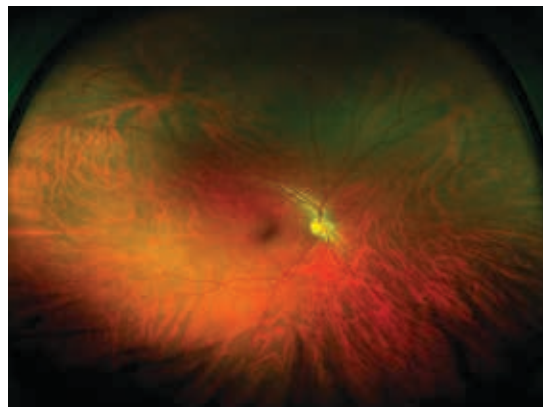


Aha Photography Award Winner, Christine Napier received, 1st prize for an image taken 'under the microscope'.



Research Impact

Research is a long process, but every day, month and year, progress is made. In 2014, CMRI researchers leaped ahead in their understanding of cancer, infectious disease, development, and eye genetics.



There isn't enough space to cover all the research impact we've made this past year thanks to your support, but here's a few highlights from our upcoming annual report:

- We've determined dynamin-inhibitors should be safe for treating cancer and epilepsy in humans
- we've learned dynamin inhibitors may be a potential treatment for Ebola
- we identified a new gene for aplastic anaemia and short telomere syndrome
- we uncovered a key anti-cancer target for ALT cancers, some of the most aggressive types, the biggest discovery in this field in the last 15 years
- we're gaining a better understanding of brain plasticity, which is important for learning, memory and Parkinson's disease
- we've discovered new genes causing retinal disease, cataracts, or glaucoma
- we published evidence that exercise is beneficial for Rett syndrome
- we've developed a new understanding of RNA editing, a vital process in all cells, that has implications for embryo development and normal cell functions
- we've gained new understanding of some triggers for changes in cell shape (tight versus loose) which is important for understanding how a solid tumour moves to other parts of the body
- we're learning how individual variations in our DNA (SNPs) predispose to ovarian cancer
- we're using the latest gene sequencing technology for early screening of infants prone to cataracts so their sight can be corrected early enough for them to have any hope of developing sight

You can also explore and learn more about our research areas online on our website. — Read more online at www.cmri.org.au/research

Committee members Beth, Nancy and Marie at the Strathfield Golf and Bridge Day on 27 January. A capacity crowd of 124 eager golfers signed up.



Professor Ian Alexander's research was recognised by two NHMRC grants and an ARC grant at the end of 2014. Congratulations!



Visit our new website to watch a new video on CMRI's completed research facilities – See more at: cmri.org.au/News/Videos

Associate Professor Robyn Jamieson is looking for the genetic causes of blindness and how to treat conditions like retinal disease which affects 1 in 3000 people.



Q & A

With Professor Robyn Jamieson

Q: Why do you choose to study eye diseases?

A: Blindness, especially in children, has a devastating impact on individuals, families and society in terms of lost potential, not to mention medical and disability costs.

Q: What hope do you offer patients?

A: For some cases of retinal disease, there are significant new advances in treatment, with improvement in vision reported after trials using gene and cell

based therapies. In order for patients to access these potentially sight-restoring trials, and for scientists and doctors to develop new therapeutic strategies, we need to know more about the underlying genetic error in patients.

Q: What are you working on now?

A: We're hoping to develop a cell-based treatment for retinal diseases, and have started the key groundwork needed. We're focusing on a new syndrome we've discovered, but what we develop will be applicable to other retinal diseases.

— To ask one of CMRI's scientists a question send an email to marcomms@cmri.org.au



Community news

Jeans for Genes is one of Australia's most recognized and loved charity campaigns and is a major fundraiser for CMRI. 2014 was the 21st Anniversary of Jeans for Genes, an occasion celebrated with enormous energy and enthusiasm across the country.

Over \$2 million was raised thanks to the dedication of an army of supporters, including more than 1,000 volunteers collecting donations and some of the country's most recognisable retail chains, including Big W, Outback, HCF, Lowes, Crazy Clark, Sam's Warehouse, Newcastle Permanent, Live Life Pharmacy, and Jeanswest contributing their support. Thousands of individuals sold merchandise and held events,

while a record number of schools donned their jeans and made gold coin donations.

2015 heralds a new era for Jeans for Genes as we continue to create innovative ways for the public to support our cause. This year will see the introduction of virtual Jeans for Genes badges, an exciting addition to wearing an actual badge on the day. Keep an eye out on Facebook for more information.

Many dedicated supporters have already registered their details even before this year's campaign officially opens and you can too at

jeansforgenes.org.au. We're looking forward to a massive year in 2015, and, with your help, we hope to set a new fundraising record!

— Read more online at jeansforgenes.org.au

Jeans for Genes Day 2014 volunteers



Dates for your diary

15/03 Vacluse Tennis Day

30/03 Port Hacking 50th Birthday Gala Dinner

30/03 Racquet Committee Card Day

08/05 Hills Mothers' Luncheon

27/05 Beecroft Card Day

29/05 AGM of Committees

25/07 Gosford Committee Trivia Night

07/08 Jeans for Genes Day

30/08 Jeans for Genes Gala Dinner

— Contact Jennifer Philps (jphilps@cmri.org.au) for more information on upcoming events or visit cmri.org.au/events

