

under the microscope

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Genes and behaviour: the forgetful mouse

"More research in this area is urgently needed as Rett syndrome is the second most common form of severe mental impairment in girls after Down syndrome in our country."



Dr Gregory Pelka of the Embryology Unit

Dr Gregory Pelka of the Embryology Unit at the CMRI has recently made a significant contribution to our understanding of the learning difficulties and memory loss associated with Rett syndrome. Using mice which model Rett syndrome to analyse the activity of more than 20,000 genes, Dr Pelka has also identified 13 genes which may be involved with the disease and hopes that these new findings will add to our understanding of this devastating condition.

The results from this research have just been published in the highly regarded journal, *Brain*.

Rett syndrome is a genetic neurological disorder that predominantly affects girls and is found in 1 in every 10,000 births. Following an apparently normal period of development for the first 6-18 months, these children then display a progressive loss of speech and coordination of movement. Cognitive development can be dramatically hampered and many patients become wheelchair bound.

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Director's desk



The Institute is a charitable trust committed to the conduct of high quality bio-medical research. It is also, however, a complex scientific and business operation, which demands constant monitoring and surveillance to ensure that we not only operate efficiently but that we

comply with the rules of multiple regulatory authorities. The Board has completed a Risk Assessment Analysis conducted by Wyndarra Consulting. While it would appear that our overall standards of compliance are high, the Board acknowledges that we must always look to the future. One of the issues that remains to be addressed is the relevance of both our constitution and our Board membership in relation to modern business standards. This will be of increasing importance as business and commercial partnerships evolve. Charitable trusts are under continuous government scrutiny and we must ensure that we are always in a position to satisfy the standards set by regulatory authorities.

Another risk area identified is the security of our fundraising activities. The Board commissioned Anita Batho of Torch to undertake an analysis of our approaches in this area. Many of you, both staff and supporters, were involved in this process. While her focus was on the relationship between the Jeans for Genes appeal and the Institute, she had to necessarily explore the full range of appeal activities. Her report has been accepted by the Board and it will be up to us to use her guidelines in creating a plan for the future.

The Board is looking at an expansion of our present building as it is clear that we are in need of research and administrative space. The architects for our present facility, Ancher/Mortlock/Woolley, have completed a schematic plan for a possible development on our forecourt area. Although we will of course be seeking external support for this venture, it is a tribute to the successful efforts of the Board that we are in a position to even contemplate such an expansion programme.

The Finance and Audit Committees of the Board have been extremely effective in the oversight and development of our resources and the Institute and its supporters are grateful for the commitment of the Board members.

It is evident that 2006 is already an extremely big year!

Professor Peter Rowe

Successful scientists awarded prestigious grants



Dr Roger Reddel Head of the Cancer Research Unit

Head of CMRI's Cancer Research Unit, Dr Roger Reddel has just received a large Cancer Council NSW grant for the third time. A total of two million dollars spread over five years was awarded.

The award will fund seven major ongoing areas of research into ALT, a method that cancer cells use to multiply in human cancer cells.

Dr Reddel said, "With the help and commitment of the Cancer Council, we are in a strong position to complete the journey from initial discovery of a previously unknown cancer mechanism all the way through to development of treatments designed to combat it."

CMRI Eye Genetics Research Group Leader, Dr Robyn Jamieson, has been awarded a grant from the Ophthalmic Research Institute Australia to support research into inherited eye conditions, including glaucoma and eye malformations.

"We have genetic samples from affected patients, so this grant allows us to study these conditions in more detail," said Dr Jamieson. "Due to the limited number of granting bodies in vision research it is a real honour to receive this grant," acknowledges Dr Jamieson.

These grants come as welcome recognition of the excellent standard of research conducted by CMRI's scientists and of the importance of their work.



Dr Robyn Jamieson, Chris Willcock and Marija Mihelec of the Eye Genetics Research Group

New Talent New Faces

The international reputation of CMRI and its researchers attracts young and talented researchers from across the country and globe.



Dr Tony Cesare of the Cancer Research Unit

Dr Tony Cesare will be at the CMRI at least two years, supported by a post-doctoral fellowship from the American-Australian Association. Tony will be looking into the mechanism by which cells respond to damaged DNA and telomere dysfunction. He wants to see if there's a link between this and the ALT mechanism, one of the methods that cancer cells use to maintain an indefinite lifespan. "This is a good chance for me to get an excellent scientific education, the cancer lab here is one of the best" says Tony of his new position.

From closer shores we welcome Vanessa Franklin who has joined the Embryology Research Unit as a Research Assistant. Vanessa will be investigating genetic factors that potentially contribute to cleft lip and palate. In particular she is looking "to get a sequence of events that might be causing cleft palate".

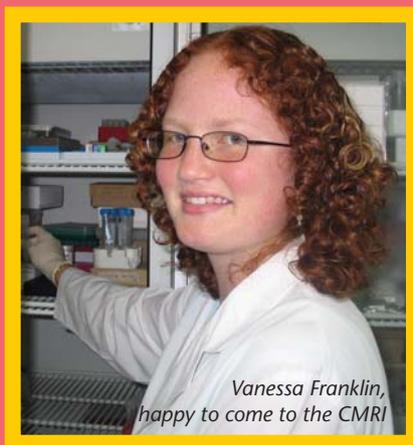
"I think research on cleft palate is very interesting. I've always thought that I'd really love to be someone who found a cure for something and I think this is the best place to do that" says Vanessa, who is keen to settle into her work at the CMRI.



Dr Megan Fabbro returning to Sydney

Another new face is Dr Megan Fabbro who started in Cell Signalling this month, she is supported by an NHMRC Peter Doherty Fellowship. Megan has joined us from Brisbane where she was researching the final stages of cell division, and its implications in breast cancer.

Megan became interested in the research carried out at the CMRI when she attended a seminar at which Dr Phil Robinson spoke. "I was really excited about Phil's area of research and his enthusiasm for science" says Megan about her decision to move to the CMRI.



Vanessa Franklin, happy to come to the CMRI

The forgetful mouse

Continued from page 1

Rett syndrome is predominantly caused by mutations in a gene known as *MECP2*, which is located on the X chromosome. By creating mice which lack this gene and studying their behaviour, Dr Pelka discovered that the affected mice were less active and not very good at learning and remembering simple behavioural tasks. The activity of a number of other genes was also found to have changed in an area of the brain associated with memory and learning. It is likely that these genes may be linked to the development of Rett syndrome and shed some light on possible therapeutic treatments.

'More research in this area is urgently needed as Rett syndrome is the second most common form of severe mental impairment in girls after Down syndrome in our country' says Dr Patrick Tam Head of the Embryology Unit.

The research on Rett syndrome is an ongoing collaboration between the Children's Medical Research Institute and the Western Sydney Genetics Program based at The Children's Hospital at Westmead. The Embryology Unit has recently set up a collaboration with Dr Anthony Hannan (a former PhD graduate of the CMRI) of the Howard Florey Institute in Melbourne to analyse aging Rett syndrome mice for abnormal changes in their behaviour. Dr Hannan, an expert in this field, recently won two Eureka Science Awards in 2005 for his research into Huntington disease.

Dr Pelka's innovative work has attracted a US\$100,000 grant from the Rett Syndrome Research Foundation (USA), to study the role of the *Mecp2* gene on behaviour.

“Be a Gene – ius” and help support the Children’s Medical Research Institute in 2006.

“Music is Better Than Rugby. Or is it?”

In a world of passions and tribalism two great creative disciplines stand out – Music and Rugby. With this in mind, the Royal Automobile Club of Australia have organised an entertaining debate, with celebrity speakers. The event will be held on Wednesday 31 May at the Sydney Conservatorium, to be followed by a sumptuous supper at the Royal Automobile Club of Australia. The amazing Sydney Symphony Quartet, will perform for guests. For further details and tickets, please contact Lynne Wilkinson on 02 8273 2319.



Kids for Life Gala Ball

For those wanting to go to one of Sydney’s most exciting events: the Kids for Life Gala Ball “Black Tie with a touch of Sparkle” is the one to go to. It will be held on Saturday 25 March 2006. Funds raised will support the Children’s Medical Research Institute and the Sydney Children’s Hospital Randwick Intensive Care Unit. Go to www.kidsforlife.org to buy your tickets, or give a donation.

Don’t forget **Jeans for Genes Day** is on Friday August 4, 2006.
This year is going to be BIGGER than ever.



Julijana and Kelly from Jeans for Genes, fighting over a pair of Hugh Jackman’s jeans

Jeans for Genes Art Auction – 2006

This year the Jeans for Genes Art Auction will be held on Thursday 20 July. Some fantastic, personally signed jeans have been donated by Australian celebrities such as: Nicole Kidman, Hugh Jackman and Layne Beachley. The theme of “Be a Gene-ius” will carry through to the dinner auction, where all of the celebrities are gene-ius’s in their own field.

STOP PRESS . . . Let’s put our toes together and thank Dancing with the Stars

On behalf of the Children’s Medical Research Institute and Jeans for Genes we would like to thank Channel 7 and Chris Bath who raised \$15,583 on Dancing with the Stars last year. The support and publicity has been absolutely fabulous.



Exactly what we want!

Final year PhD student, Dr Jeremy Henson has been selected as one of ten researchers to receive a one year Cure Cancer Australia Foundation grant. His research application focuses on cancers that rely on the ALT mechanism for unlimited cell growth.

Dr Henson is particularly interested in a protein produced by many cancers that provides protection against certain chemotherapy drugs. During the course of his PhD research Dr Henson found reduced levels of this protein in cancers that use the ALT mechanism. This may mean that cancers using ALT are more susceptible to treatment by these chemotherapeutics.

In further work, Dr Henson discovered several other proteins whose interaction with the ALT mechanism may prove to be novel targets for cancer therapy. These proteins could be targets that will allow cancer cells to be killed without harming normal cells.

“ALT is an example of how understanding the inner workings of cancer cells could allow medicine to specifically kill the cancer and not harm the patients’ normal cells – as often happens with conventional chemotherapy,” says Dr Henson.

Kate Christian from Cure Cancer Australia says that “Jeremy’s application was ranked number one, out of 53 applications, by the medical grants assessment committee and all the peer reviewers. The aim of Cure Cancer Australia is to promote the work of young researchers and novel research, and Jeremy’s application fits both those categories, which is exactly what we want.”

The picture in the “O” on the front cover depicts colourful structures (yellow), sometimes found in the nucleus (blue), which identify cancer cells that use ALT, (Photo by Jeremy Henson).



Dr Jeremy Henson at work in the Cancer Research Unit laboratory

The Amazing Human Body

Exactly how does smoking affect lungs?
What are the effects of high blood pressure?

These are just two of the questions you can find answers to at the up-coming exhibition "The Amazing Human Body" to be held at the Sydney Superdome, Sydney Olympic Park from February 3 to April 2 this year.

This is no ordinary exhibition as it contains 20 real human bodies and over four-hundred specimens, preserved through a process of 'plastination'.

As a charity partner the Children's Medical Research Institute will receive 1.5 % of all ticket sales throughout the exhibition's national tour over the next two years.

Take this special discount coupon to receive two adult tickets, normally \$20 at the concession rate of \$16, available with tickets purchased at the door.



Dr Samantha Ginn with Noel Dowling's daughter Barbara Beynon

Generous Support

The Noel Dowling Memorial Fellowship was set up in 2002 and it has continued to generously support the research of Dr Samantha Ginn, a member of the Gene Therapy Research Unit (a joint venture of the CMRI and CHW).

Dr Ginn has focused her research on improving gene therapy approaches, using DNA as medicine, for the treatment of the genetic disease X-linked Severe Combined Immunodeficiency Disorder (SCID-X1). Sometimes referred to as "Bubble Boy Disease," SCID-X1 is a rare disorder which affects the white blood cells of infant boys. These boys battle life-threatening infections and rarely survive past two years of age without treatment.

The Gene Therapy Research team have recently been involved in a gene therapy trial in conjunction with a research group in Paris. Results from the trial suggest patients can be cured of their disease by gene therapy. However, three children in Europe have developed a leukaemia-like illness as a result of their treatment.

Dr Ginn is examining new gene delivery methods for patients with this condition in the hope of providing safer and more effective treatments in the future.



Prevention is better than cure...

And the key to prevention lies in research.
It's as simple as that!

There are over 10,000 known genetic disorders. To correct these and many other disorders research is needed to find the causes.

Here is my gift:

\$100 \$75 \$50 \$25

Other \$

Please find enclosed my cheque

Please charge my credit card:

credit card number

cardholder's name

cardholder's signature

expiry date

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address

postcode

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Please send me:

- More information about CMRI
- Christmas catalogue
- Information on my nearest fundraising committee
- An annual report
- Information on how to make a bequest
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All donations are tax deductible



I would like to join the Automatic Credit Card Donor Program

Automatic Direct Debit Amount to be donated:

\$

Please complete the Credit Card Details at left.

Frequency: Monthly Quarterly Other

Commencement Date:

Please complete this coupon, enclose it with your donation in the envelope provided and mail to:

Children's Medical Research Institute
Locked Bag 23, Wentworthville NSW 2145 Australia
Tel 02 9687 2800 Fax 02 9893 9166

Thanks for your help.



Committee Power

Racquet Committee

"Happily Everafter" was the name of the popular Christmas pantomime performed by the Pymble Players! The proceeds were generously donated to CMRI.

Judith Hyam Trust Fund for Cancer research

The Christmas party celebrated 15 years of fundraising and a total of one million dollars of funds raised. Congratulations! Dr Reddel from the Cancer Research Unit remembered with great fondness a stalwart supporter of the Trust Fund, Mrs Roma Kennedy, who passed away late last year.

Circular Keys Chorus

Once again the talented Circular Keys Chorus sings its harmonic way to supporting the CMRI.



Dates for your Diary

Judith Hyam Trust Fund for Cancer Research – Musical Sunday at The King's School

The talented Opera and Arts group will be performing at The King's School, Parramatta over a scrumptious afternoon tea on Sunday 21 May at 2pm. For further details contact Diana Mahoney on 02 9630 5572.

Hills Committee

Don't miss the "Mother's Day" luncheon at the Sofitel, Phillip St, Sydney on Tuesday 2 May at 12pm. Georgie Parker, and TV personality John Mangos will MC the event. Entry is \$80, attendees receive a fabulous gift. Contact Patti Payne 0418 867 784.

Northern Beaches Golf Day

The Annual Golf Day is to be held on Tuesday 2 May at the Mona Vale Golf Club. Tickets are \$50, including lunch. For further details, please contact Cheryl Ewing on 0419 612 240.

Mudgee Committee

Sunday 2 April 8pm, celebrate 40 years of country fundraising in glorious Mudgee by joining us for a country luncheon at "Heaton Lodge". Contact Bronwyn Sams 02 6372 3254 for a special invitation.

Port Hacking

Mother's Day Fashion Parade, Wednesday 10 May at 7pm. The latest fashions over a delightful dinner with drinks, at "Doltone House", Sylvania Waters.

Vaucluse Committee

Join in for a social mixed doubles tennis day, or just for lunch and savour the magnificent view of Sydney Harbour from "Fairwater" in Double Bay. Sunday 26 March, contact Jan Madigan 02 9343 4331.

Racquet Committee

Annual Card Day and Luncheon at St. Ives Bowling Club, Killeaton St, on Monday 20 March at 10.30am. Tickets are \$25. Bring your friends and make up a table (for bridge, whist etc.). Contact Prue Kellaway 02 9974 4197.

QUT Urban Country Music Festival

In Caboolture Monday 1 May, six 'little piggies' will be racing round an obstacle course to raise money for Jeans for Genes. Contact Angela on 07 5420 0435 for more details.

Beecroft Card Day

The Beecroft Committee are holding a Card Day on Wednesday 31 May. Men and women are invited, so come along and play Bridge, 500 or Canasta and enjoy some morning tea and lunch. For more information, contact Mary Howell on 02 8850 1271.



Clockwise from far left:

The Circular Keys Chorus;

Pymble players get together for a post-performance chat with fans;

Little piggy and friend at the Caboolture Urban Country Music Festival. This photograph was kindly provided by The Caboolture Shire;

Stephen Ryall from CMRI with Des Kennedy, President of the Judith Hyam Trust Fund.

Moving Art Touring Exhibition

The Moving Art Touring Exhibition is calling for entries from emerging artists to join this major event, which is generously supporting the Children's Medical Research Institute.

Moving Art
touring exhibition & auction

For more details, contact Tricia Donovan on 02 6733 2062 or email tricialawsonmenzies@yahoo.com.au