

under the microscope

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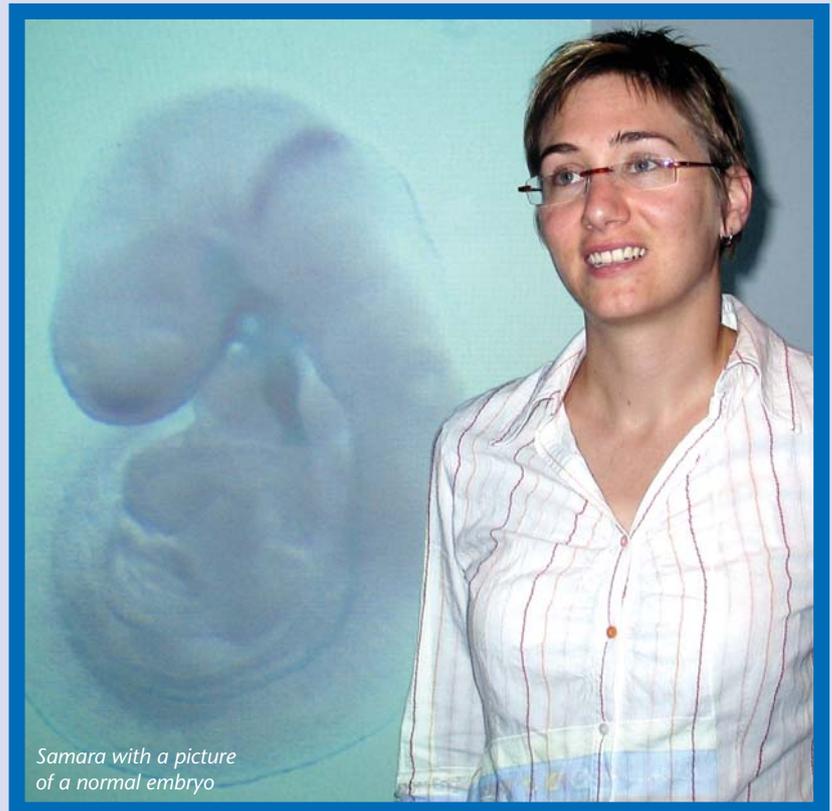
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Forging A-head

"Two genes (*Dkk1* and *Gsc*) act together to ensure normal head formation."



Samara with a picture of a normal embryo

Dr Samara Lewis of the Embryology Unit is ahead of most of us in understanding how the head is formed during development of the embryo. By studying gene interactions in the laboratory mouse, she has found two genes (*Dkk1* and *Gsc*), which act together to ensure normal head formation. Since mouse and human development are so similar, the genes involved in mouse head development are likely to work in the same way in humans.

In embryos, signalling (or cross-talking) among the cells is required to choreograph the formation of the body plan and the laying down of different tissues or organs in the right places. The *Dkk1* gene is known to block a signalling process, which needs to be tightly controlled for head formation. This type of signalling is found in mice and humans and is altered in cancers, which proves that this is a very important process.

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**CHILDREN'S
MEDICAL
RESEARCH
INSTITUTE**

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Director's desk



The Children's Medical Research Institute (CMRI) has recently completed the process for renewing its triennial accreditation as an independent research institute with the National Health and Medical Research Council. A new element of the accreditation process is that institutes are now required to have written policies regarding their engagement with the community. This led me to reflect on how extraordinarily important community involvement is, and always has been, to the CMRI. From its very beginnings, the community-based committees that support, encourage and inspire us have been at the very core of our activities. Many, maybe most, of the committee members have joined the organisation because they or their family have been touched by a major childhood disease. They are the reason for our existence and also our committed supporters. The culture of the Institute is strongly shaped by the women and men who meet with us every year and tell us amazing, sometimes humorous, and always humbling stories of how they raise funds for our research endeavours and the camaraderie that they enjoy in the process. We welcome the crowds who come on frequent occasions to hear about the latest research from the scientists who are doing the work. It's simply impossible to imagine CMRI without the great community to which we belong.

A handwritten signature in white ink on a red background, appearing to read 'Roger Reddel'.

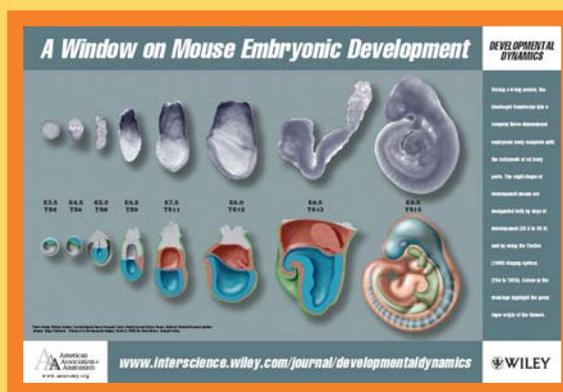
Roger Reddel
Acting Director

Painting Embryos

Kirsten Steiner of the Embryology Unit has spent the past few months painstakingly photographing microscopic mouse embryos. The photographs are for an educational poster on mouse development, which features alongside an invited review by Samara Lewis and Patrick Tam in the journal *Developmental Dynamics*.

The information in this poster is expected to be used by embryologists the world over as a reference for the way the mouse develops, and to guide researchers who are unfamiliar with mouse development.

"Three different colours have been used by an artist to represent the different layers of the embryo," explained Kirsten. "Blue represents cells that will later form skin, neurons and the brain; red indicates cells that will become the bones, muscles, connective tissues and the heart; and green shows cells that will develop into the cells of the gut."



"The fact that we can now trace the history of development of these cells in the embryos over time is a major achievement," said Dr Tam, Head of the Embryology Unit. "Other researchers will use this information to understand which part of an embryo is missing or which part is wrong when development is abnormal."

"I'd also like to thank Axel Neumann of the Cancer Research Unit, who provided valuable advice on the photography of these tiny specimens," said Kirsten.

For more information, please visit www.cmri.com.au/cmri.php?news=17

Forging A-head

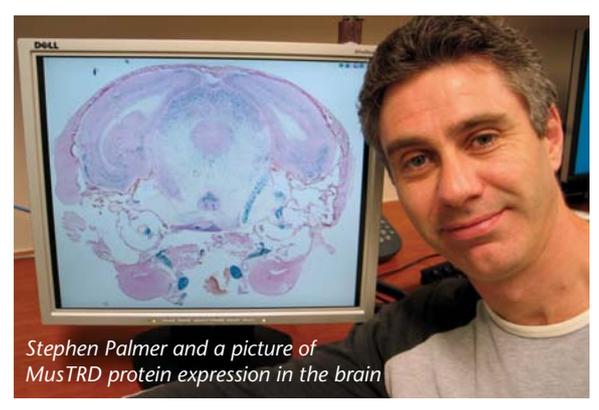
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Samara and the team have also discovered, for the first time in mice, the earliest role for the *Gsc* gene. This gene is thought to block the same signalling process as *Dkk1*, but how it works is not fully understood.

Dr Patrick Tam, Head of the Embryology Unit said, "Finding out that these two genes can work together is a step towards understanding development in humans and may be useful to identify the cause of birth defects affecting head formation in the future."

These results were published in *Mechanisms of Development*.

The picture in the "O" (see article on the front page) depicts part of a 7 day mouse embryo. The green staining pinpoints the presence of protein called β -catenin in the cells, which is important for signalling; the nuclei of the cells are stained red.



Stephen Palmer and a picture of MusTRD protein expression in the brain

William's Syndrome and MusTRD

Dr Stephen Palmer of our Muscle Development Unit is interested in a gene called MusTRD, because one of the two copies of this gene is missing in people with a condition known as William's syndrome.

William's syndrome children display an outgoing, talkative or gregarious personality, but also have very specific phobias and anxieties. Unfortunately, they can suffer from a life-threatening heart disease, low mental ability and most will never live an independent life.

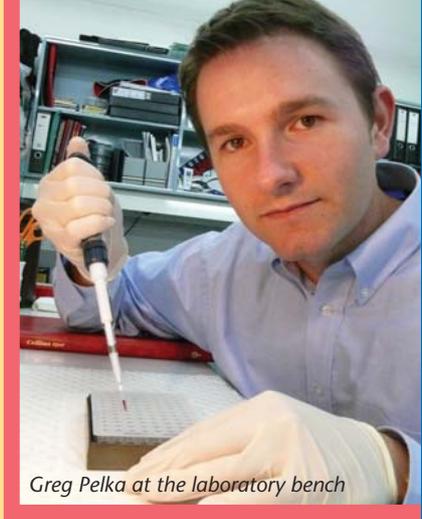
In addition to MusTRD, about 19 other genes are deleted in William's syndrome. Identifying which gene is responsible for the behavioural features of this syndrome will help us to understand the genetics of normal behaviour.

The MusTRD gene codes for a protein, which is thought to control other genes, turning them 'on' or 'off' or changing the way they are expressed. In the quest to discover which genes are controlled, scientists need to determine which cells of the body make MusTRD protein.

Using a mouse developed by Dr Enoch Tay, the team was able to identify which cells in the body have the MusTRD protein. Importantly, MusTRD was discovered in specific cells of the brain, which may explain the behavioural differences observed in these patients. The protein was also found in parts of the developing face, eyes and ears, which could explain the 'elfin-like' face, and the abnormal eye and ear development typically found with William's syndrome.

"Finding a link between the MusTRD gene and the clinical features of the disease is very exciting," said Dr Stephen Palmer, "as it is the first step to fully understanding what is happening in William's syndrome patients."

Our Eye Genetics Research Group and the Oncology Research Unit at The Children's Hospital at Westmead also contributed to this work, published in *Gene Expression Patterns*.



Greg Pelka at the laboratory bench

Young Scientist Awarded Top Research Grant

Dr Gregory Pelka of the Embryology Unit has been awarded the prestigious Peter Doherty Biomedical Fellowship for four years by the National Health and Medical Research Council. He is investigating the *MECP2* gene, necessary for movement, balance and learning. This is important as faults in this gene contribute to the symptoms of Rett syndrome, which affects one in 10,000 girls and is the second most frequent form of severe mental disability after Down syndrome.

MeCP2 controls the expression levels of various genes, which in turn affect the levels of proteins in a cell. Dr Pelka's project aims to identify the precise regions of the brain where MeCP2 is absolutely indispensable. Despite the complexity of the disorder, Dr Pelka hopes his research could eventually contribute to an effective treatment, to lessen the burden of Rett syndrome on patients and their families.

Tomorrow's Scientists

A new scholarship program has been initiated to attract the brightest, keenest minds to take on the challenge of a research project at CMRI. In October of this year, top university graduates interested in an exciting career in medical research were invited to apply for PhD scholarships.

A short list of highly promising candidates was selected, interviewed and four of them were awarded the CMRI scholarship. All have accepted the award and we now look forward to welcoming these trainee scientists to the Institute in February. They will have the opportunity to gain valuable research experience, work alongside our top research scientists, contribute to scientific knowledge and will hopefully take up exciting research careers in the future.

Dr Phil Robinson, Convenor of the Scholarships Award Panel said: "It is due to the extraordinary generosity of Claire Yass, the Judith Hyam Memorial Trust Fund for Cancer Research and Douglas and Lola Douglas that we are able to fund Australia's future generation of research scientists in this way. The estate of Claire Yass and the Judith Hyam Memorial Trust Fund for Cancer Research have been providing financial support for student scholarships for a number of years, which together with the new support from the Douglas estate is vital to the continuation of this program."

Jeans for Genes®

Jeans for Genes has Expanded!!!

Due to the success of Jeans for Genes over the past few years, the Board of the Institute noted that it was time to employ extra staff - to help raise more funds for the facility and support the ever increasing Genie base.

CMRI is pleased to welcome three new staff members. Christine McGee, our Special Events Manager, Lynda Dave our Genie/Volunteer Coordinator, and Melissa Nassif who will replace Kelly Morgan while she is on maternity leave.



New J4G girls
Chris, Melissa
and Lynda

Jeans for Genes Update

To date Jeans for Genes 2006 has raised nearly \$3.9 million with still more funds coming in. We do expect to reach \$4 million before the end of the financial year. Once again, thank you so much for your fabulous support. If you still have not sent in your donation or monies please do so as soon as possible.

Jeans for Genes Sponsorship Opportunities

Jeans for Genes 2007 is going to be a big year. We are looking for key sponsors for some of our major national events including: 'Jamm for Genes®', 'Do it with Denim®', and the new 'Jeans for Genes Golf Day,' which will be held in May. If you would like us to send you a proposal, please contact Christine McGee 02 9687 2800 or cmcgee@cmri.com.au

Charity Gift Wrapping - a Huge Success

Weber BBQs, Casio keyboards and iPods are just a few examples of the many Christmas gifts that over 200 Genies wrapped and made beautiful for MYER customers in Bondi Junction, Sydney City and Parramatta.

Despite volunteering long hours, our Genies smiled and chatted with shoppers with enthusiasm and passion for the CMRI. They staffed the charity gift wrapping stations from early in the morning until late, raising a fabulous \$28,384. A big thank you to MYER for this wonderful opportunity.



Gift wrappers Trudi Arnold and Melanie Trevor

Our wonderful Genies included staff from: Westpac, NRMA, AAMI, Hunter Hall International, Cundall, Origin Energy, The Good Guys, Coverforce, Kimberly-Clark, KPMG, The Australian Tax Office, Yahoo, and IAG; university students and people just wanting to help in their spare time. If you would like to register as a Genie, please contact Lynda Dave 02 9687 2800 or ldave@cmri.com.au

N.B. For your diary – Jeans for Genes Day – Friday August 3 2007.

Sir Norman Gregg's Legacy

Internationally renowned Australian ophthalmologist and one of the CMRI's founding fathers, the late Sir Norman Gregg discovered the connection between rubella in pregnancy and blindness in the 1940's. In memory of this ground-breaking eye research the Sir Norman Gregg Post-doctoral Fellowship was established, with a bequest from Sir Norman's daughter Sheila.

This prestigious fellowship has been awarded to Dr Yongjuan Chen, who recently joined the Embryology Unit. Dr Chen, conducted research into eye development for her PhD study with Professor John McAvoy of the Save Sight Institute at the University of Sydney. She will be working with Dr Robyn Jamieson, who leads the Eye Genetics Research Group, to investigate the genetic causes of congenital eye defects.



Dr Yongjuan Chen
with Dr Robyn Jamieson

Committee Power

COMMITTEES

Quirindi Committee Race Day was a fabulous event and Wendy Cudmore received a CMRI life member badge for over 30 years of dedicated fundraising.

The **Hills Committee Race Day Luncheon** was an overwhelming success. We would like to thank John Lees and John Stanley for their valuable contributions.

The **Racquet Committee Melbourne Cup Day Luncheon** on the veranda was a fabulous event enjoyed by 90 guests.

Gerringong Committee has raised an amazing One Million Dollars for CMRI after 47 years of committed fundraising. A powerhouse of support since 1959, the Committee celebrated at the **Annual Gerringong Committee Quilt Show**. Pharmacist Mr Glenn Steele generously donated the last \$1000.

Gosford Committee Garden Party was a huge success. Guests enjoyed an inspiring Christmas table decoration demonstration by Bobbie Shailer, stalls and a fabulous afternoon tea.

The **Wagga Christmas Fair** had the usual huge turn out of Christmas bargain hunters. This year's Annual Fair was full of fun, in spite of the difficult rural conditions. Thank you for your generosity Wagga Wagga.

At the **Canberra Committee Government House Luncheon** snow on the mountains did not cool the spirits of well over 500 guests, who relocated from Government House to the warmth of the beautifully decorated old bus depot. It was a roaring success.

A perfect summer evening was enjoyed by a packed audience at **Tamworth Committee Carols in the Park**. The event was skilfully MC'ed by Kevin Anderson, who introduced many talented performers.

OTHER SUPPORTERS

At the **Glen Innes Open Garden Scheme** Rhonda March opened her garden for CMRI. In spite of drought, snow and torrential rain 'Briarsdene' looked magnificent and \$3,000 was raised thanks to the generosity of the Glen Innes community.

Many thanks must go to **Hunter Hall International** for their very generous donation as part of their Charitable Giving Program. We are delighted that CMRI is regarded as another good investment.



Top to bottom:

Wendy Cudmore with her life member badge and Rhonda March at 'Briarsdene';

Hills Committee President Patti Payne (centre), with CMRI's Jan Mullin and Libby Brooks;

Guests at the Racquet Committee Melbourne Cup Luncheon;

Gerringong Committee's Kate Quinn, Rhonda Bailey, Dorothy Hambridge, Margaret Weir and Jenny Bolden with Glenn Steele at the Gerringong Quilt Show (Photograph courtesy of Illawarra Mercury photographer Andy Zakeli);

Cathy Johnson Corporate Relations Manager for Pernod Ricard Pacific, who generously donated the wine, with Canberra Committee President Celia McKew.

A Sad Farewell to Olive Drayton

Sadly we recently lost a very dear friend of CMRI, Olive Drayton. Olive was the driving force of the Cessnock Committee of CMRI raising almost \$100,000 from the local community. She was a very well-loved Cessnock identity and will be sorely missed for her devotion and endless fundraising for CMRI.



Dates for your Diary

Monday 30 April. The **Racquet Committee Card Day and Luncheon** will be held at the St Ives Bowling Club. Bring along your bridge four and enjoy this social event. Contact Prue Kellaway 02 9974 4197.

Friday 11 May. At this year's legendary **Hills Committee Mother's Day Luncheon** MC John Mangos will chat to celebrity Australian chef and author **Kylie Kwong** and her mother Pauline, Hilton Hotel, Sydney, 11.30am. Contact Patti Payne 0418 867 784.

Wednesday 4 April. The **Allan Research Trust Race Day** at Sydney Turf Club, Canterbury Park will be a major fundraising event for CMRI. Contact Janys Allan 0407 987 473 to be a sponsor with naming rights, signage and to sash the winning horse...