

# under the microscope

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## From strength to strength

**S**heer grit and determination allowed one child to overcome his inherited muscle weakness disorder. Greg Cremen, 45, who suffers from nemaline myopathy, is now a competitive cyclist.

"My father always taught me that my condition was a challenge and that I should not give up pushing myself even further," says Greg.

By exercising Greg has been able to make his muscle fibres grow larger and therefore stronger, effectively treating himself and reducing his weakness.

Now a muscly mouse has shown Dr Edna Hardeman, head of CMRI's Muscle Development Unit and Associate Professor Kathryn North of the Children's Hospital at Westmead a way to help many children benefit from this simple natural approach.

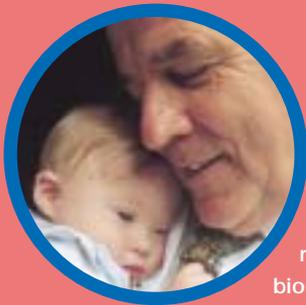
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*Pictured at top: An isolated muscle fibre showing genes switched on in the nuclei (blue). Photograph by Steve Robinson, Muscle Development Unit, CMRI.*

*Cyclist and nemaline myopathy sufferer Greg Cremen*

# Director's desk



Seeing the bigger picture...

The university teaching hospitals of the USA have long been responsible for producing much of the world's high quality biomedical research. Many of the individual departments within these institutions with their high quality clinical and research staff and access to state of the art research tools and financial resources constitute research institutes in their own right.

In Australia we operate on a smaller scale with the result that gifted individuals are often isolated in their efforts to pursue their research goals. The resulting frustration leads to them abandoning their research careers or pursuing them overseas.

Given the administrative and financial constraints placed upon our hospital system it is imperative that we organise our resources more effectively if we are to retain a place in the international system. This may entail some sacrifices on the part of those who are involved in empire building, but unless some consensus can be achieved in ensuring that adequate resources can be provided for the support of talented staff, we are doomed to failure. This is the general message delivered in the context of the Wills and Batterham reports on Australian science. There is the temptation that, in order to gain financial advantage many pseudo alliances will be formed to create the impression of collaborative research ventures. Eventually such schemes will fail but the cost will be great not only in terms of dollars but also with respect to the credibility and integrity of the world of Australian science.

Coupled to this is the huge emphasis being placed on the necessary economic relevance of research. This has the capacity to stifle the creativity of research, which has always been a hallmark of Australian science. On the Westmead Campus, the CMRI has the opportunity to build credible alliances with the Children's Hospital at Westmead, the Westmead Millennium Institute and Westmead Hospital, in a cooperative research environment. It is still unclear how this will be achieved and what form these will take, but we owe it to future generations to work towards this goal.

Professor Peter Rowe

Photo courtesy Wagga Advertiser

## From Strength to Strength

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Nemaline myopathy mouse with its muscly counterpart

Nemaline myopathy is a recessive genetic disorder in which children inherit a faulty muscle protein gene from both parents. The disease causes generalised muscle weakness and the severity of the disease varies in different children. In extreme cases the muscles that control breathing are affected and the child dies of respiratory failure soon after birth.

"In the past we have questioned whether exercise might be too much for the muscle to handle, but Greg has demonstrated that this is not the case," says Dr Hardeman.

Using a mouse model of nemaline myopathy, Drs Hardeman and North have shown that mice, like Greg, are able to effectively cure themselves of the disease by increasing the size of their muscle fibres. This phenomenon, called hypertrophy, occurs in humans when they exercise, so increasing their muscle strength. But in humans the effects of exercise can differ and some children with nemaline myopathy are too weak to exercise.

Using clues from people like Greg and the mice, Dr Hardeman and Assoc. Prof. North believe they have found the key to produce an effective treatment. The disease only

becomes a problem in the mice when they age and the muscles begin to shrink.

By exercising the mice on motorised treadmills they hope to confirm the beneficial role of exercise and hold off this age related muscle atrophy.

*"The question now is what is enough exercise to overcome the disease, and what are the pharmacological alternatives for children too weak to exercise."*

DR HARDERMAN

They will also try to mimic this effect with a number of pharmacological agents.

A promising candidate drug is a natural growth factor involved in muscle development, called Insulin-like Growth Factor I. Dr Hardeman is also testing other hypertrophy promoting compounds, which are the subject of a patent application, in collaboration with Emory University in the United States.

"Depending on the results that we achieve with the mice, we hope to be able to use these treatments on children with nemaline myopathy," said Dr Hardeman.

Jeans for Genes® Day is rapidly approaching. This year we have many new aspects to the campaign. There's our new baby image "Katie".

We also have new \$10 and \$5 badges and \$2 temporary tattoos. The \$10 badges are modeled on two Jeans for Genes artworks: Shania Twain and Nick Farr Jones by artists Paul Newton and Garry Fleming.

# Jeans for Genes

## What's happening?



Dr Patrick Tam

## Promoting excellence in science

"Truly exceptional" and "a real pioneer" were the phrases used to describe Dr Patrick Tam, head of CMRI's Embryology Unit, in response to his application to the top level of the prestigious Research Fellowship of the National Health and Medical Research Council (NHMRC).

The Fellowship scheme supports outstanding scientists allowing them to continue contributing research that is of major benefit to both their field and Australian science. Dr Tam has rapidly progressed through the ranks of NHMRC fellowships in just four years and has now been promoted to Senior Principal Research Fellow. This is the highest level of appointment possible and is awarded only under exceptional circumstances. The Fellowship Committee was unanimous in its decision to approve the promotion, stating that, "It is clear that Dr Tam is an exceptional scientist, and a credit to Australian science."

"This is a significant milestone of my scientific career," said Dr Tam. "It is a recognition of my achievements as a medical researcher. The Fellowship also provides substantial funding for the research program of the Embryology Unit at the CMRI.

The CMRI is proud to count Dr Tam as one of its own.

"For Me" magazine is running a Jeans for Genes® Blue-jean Babies hunt for Australia's most adorable jean-clad baby under three. The lucky overall winner will receive a \$500 Target voucher, a \$1000 Lamaze toy pack, a \$500 Johnson & Johnson baby hamper, a \$500 Innox pack and a dapple grey life-like rocking horse from Rocking Horse Australia. These rocking horses are individually handcrafted to a fine and lustrous finish, with real horsehair for the tail and lashes. Maybe you know a baby you would like to enter, if so buy a "For Me" magazine and enter the competition.

The Celebrity Art Auction for 2001 will be held at the Regent Hotel, Sydney on Saturday, 28 July, and will have a black and white movie theme. It promises to be a dazzling night, with celebrities, great entertainment, a fabulous raffle and a wonderful cause. The artworks are looking fantastic too! Call Mary or Amanda for more information on (02) 96872800.

We have been making many personal visits to both regional NSW and interstate genies this year. The Jeans for Genes® team think it very important to thank our genies first-hand and let them know that we value all their hard work, and it's so good to meet these wonderful people.

The Jeans for Genes® team welcomes Mary Donnelly who has been with the CMRI as Personal Assistant to the Director for eight years. She has job-shared in this position for over seven years and has recently gone full-time with the rest of her time spent as Special Projects Manager in the PR Department. Mary has been involved with the Jeans for Genes campaign since its inception, but this is her first official year as part of the campaign team. She will sit on the Celebrity Art Auction committee and oversee the implementation of an automated messaging and invoicing system at our outsourced Jeans for Genes® call centre.

Amanda Eaton is the Assistant Campaign manager of Jeans for Genes. She has a Bachelor degree in Communications and has been working on the campaign for one year. Amanda's role is to meet the needs of all the major retail badge outlets and coordinate badge selling at railway stations. Amanda also sits on the Celebrity Art Auction committee.

Call us on 1 800 677 260 for information and to order your badges, and don't forget to wear your jeans on Jeans for Genes® Day Friday 3 August.



From top: Baby Katie, \$2 temporary Tattoo, Amanda and Mary, one of the prizes in the 'For Me' Jean Clad Baby Competition.



CMRI Cancer Researcher Lyndal McLure with the new Mass Spectrometer

# a MASSive advance

*"For many years researchers have been able to find proteins, but we could only dream about their function," Says Dr Phil Robinson, head of CMRI's Cell Signalling Unit. "It used to require years of work to identify a single protein and determine from which gene it originated. Now, we will be able to do it in 2 days, and can do 100 or more every day!"*

Thanks to CMRI and Jeans for Genes® supporters and a generous donation from Mr James Fairfax, the half million dollar investment in a MALDI Mass Spectrometer will bring the CMRI to the forefront of current protein research.

Only the third of its kind in NSW, the Mass Spectrometer will allow the CMRI to benefit from and build upon the wealth of information generated by the human genome project and the subsequent project to identify all human proteins, or the "proteome" as they are collectively called.

Proteins are the ultimate products of most of our genes. They are the tiny machines that carry out the functions of living cells. New generations of Mass Spectrometers are capable of measuring with incredible precision the exact mass of individual proteins and their fragments.

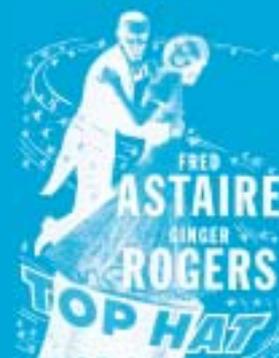
To identify a newly discovered protein the Mass Spectrometer is used to produce a mass

"fingerprint" of a fragmented protein, similar to a DNA fingerprint. The fingerprint is sent via the Internet to GenBank, the vast database of information generated by the human genome project. Computers compare the unknown protein to all the genes in GenBank and come back with a rapid identification for up to 90% of samples.

For the Cell Signalling Unit this means that they will be better placed to piece together the network of genes and proteins that help nerve cells in the brain communicate with each other.

The new machine will also assist the Cancer Research Unit to identify proteins that are present in cancer cells but not normal cells. Similar benefits will also be felt in most of the other CMRI Units.

Instead of crawling towards discovery of new drugs that can help treat brain disorders such as epilepsy, schizophrenia and cancer we can now start running towards this goal.



The Thumbelina Committee of the CMRI invites you to enjoy an afternoon of family fun and nostalgia, Sunday, 26 August 2001, 11am at the Hayden Orpheum Picture Palace, Cremorne. Tickets \$25

Entertainment by Helen Zerefos accompanied by the Orpheum's own Neil Jenkins on the mighty Wurlitzer organ will be followed by a screening of the classic movie "Top Hat" starring Fred Astaire and Ginger Rogers.

Free movie packs and lots of prizes to be won. Also a chance to win 'The Genes On Broadway' raffle...A week for two in New York AND tickets to a show on Broadway.

For CMRI preferential bookings please send a cheque (made out to the Thumbelina Committee CMRI) or quote your credit card details PLUS a stamped addressed envelope to: Genes on Broadway, PO Box 328 Lindfield 2070. Tickets will be issued at the beginning of August.

## Medical Research week

Medical Research Week is 4-9 June 2001. The theme of this year's event, organised by the Australian Society for Medical Research, is 'Medical Research – an Everyday part of life'.

The NSW branch of ASMR has organised a week filled with a variety of activities. A scientific conference kicks off the week, which also includes a public lecture and the presentation by the NSW Premier of the Amgen award for outstanding young researchers. The week culminates on Friday, 8 June with an Expo, held at the Customs House in Sydney.

The CMRI will have a strong presence at the conference, and at the Expo which will showcase key NSW medical research institutes and organisations. The Expo will provide a fascinating and interactive insight into the wealth of Australian medical research as well as a resource for young Australians contemplating science as a career. For more details go to [www.asmr.org.au](http://www.asmr.org.au)





*Left: The ladies of our Beecroft committee provided delicious homemade refreshments at Jazz in the Mountains. Seen here beside the beautiful 'Motherhood' statue by Tom Cooley. Right: Parliament House Luncheon – Hon Ian Sinclair and his wife Rosemary, seated, with Ross Cameron, MP and members of the Strathfield Committee. Bottom Right: Professor Rowe speaking at Fenton, home of Sir Lorimer Dods for much of his life.*



### Strathfield Committee

The Hon Ian Sinclair won over the appreciative audience with his warm, witty and pertinent speech at the Parliament House Luncheon in March.

A special thank you also goes to Trevor Thiele for years of support organising the Holden Dealers Golf Day in benefit of the Strathfield Committee of CMRI.

### Port Hacking Committee

The committee are as active as ever and must be congratulated on the highly successful South Pacific Lu'au. The Mother's Day 'Covers' Fashion Show brought mothers of four generations together for the dinner and show. The new season's fashions were widely applauded and a grand time was had by all.

### Thumbelina Committee

The committee yet again organised a fantastic Easter Fashion Show with a fantastic seasonal catwalk display provided by Jan Jolly Fashions.



# Fun and Functions

## Fenton

### *Looking back and looking forward*

A celebration was held on 7 March to commemorate the life of Sir Lorimer Dods, founder of the CMRI. Sir Lorimer was born 101 years ago on this day and died 20 years ago, also on 7 March. CMRI supporters, staff and friends and family of Sir Lorimer gathered at 'Fenton' his beloved home in Edgecliff, Sydney, to reflect on his achievements and those of the CMRI. Sir Lorimer's grandson, Rev. Simon Manchester, spoke about his grandfather with affection and humour, while Professor Peter Rowe spoke of the recent high profile successes of CMRI scientists and looked forward to many more years of excellence in striving to realise Sir Lorimer's dream. CMRI wish to express their gratitude to the Tudehope family, current owners of 'Fenton', who were so generous in opening their beautiful home for the event.

### Jazz in the Mountains

Many thanks to the Rotary Club of Beecroft for the magnificent event. The weather, the garden at 'Bisley' and the combination of traditional and modern jazz produced a perfect day.

### Griffith Committee

Mira Martinazzo, landscape architect and designer, delighted guests at the Rose Luncheon held in Griffith in April. The success of the luncheon is due to the enthusiastic members of the Griffith committee. For those in the Griffith area looking for another dimension in their lives contact Margaret on 6964 4039 to join this active committee.

## Dates for your Diary

### Allambie Committee

Round off Jeans for Genes® Day with a "Night at the Trots" on 3 August at Harold Park Paceway. Contact Jeanne 9982 6053 or Jenny 9451 9563.

### Blacktown Committee

Annual Car Rally. Start time 9-10am Sunday, 27 May Evans High School, Blacktown. BYO Picnic.

### Jeans for Genes Quiz night

Friday, 3 August 7pm at Blacktown Civic Centre. Cost \$10. BYO supper and drinks. Contact Gwen 9621 5202.

### Port Hacking Committee

Jeans for Genes Ball at Doltone House on 3 August. Contact Angela 9527 7655.