

# LYSOSOMAL DISEASES NEW ZEALAND

April Newsletter 2007

## New Zealand Lysosomal Storage Diseases Support Group

John Forman  
+64 4 566-7707

Jenny Noble  
+64 7 544-8868

[www.ldnz.org.nz](http://www.ldnz.org.nz)



## Our mission

To improve contacts, information sharing and support among affected people and their families, within New Zealand and Internationally.

To advocate for and support accelerated research into the causes and treatment of Lysosomal Storage Diseases.

To advocate for and support improvements to the clinical care of affected people.

## Table of Contents

Kypho's Corner	2
Late Breaking News / LDNZ host Charity Dinner Fundraiser	3/4
Announcement of Australian Batten Conference	5
Exploring novel ways to understand and treat Lysosomal storage disorders	6
Expanded New Born Screening	7
Government Announces Carer Strategy	8
Prof. David Palmer wins NIH funding for Batten Disease	9
LDNZ involved in Access to Medicine Coalition	10
LDNZ's wonder diet for Stress	11
Donations / thank you	12

## UPCOMING EVENTS

- ◆ **LDNZ Fundraising**  
Charity Dinner— 29th September 2007  
**See page 3**
- ◆ **UK MPS and Related disease conference**  
29th June—1st July 2007
- ◆ **ISM RD - 2nd International Scientific Conference** 26th—28th July 2007 Michigan
- ◆ **4th Australian Batten Conference**  
26th—29th October 2007 NSW

**Life is like a taxi. The meter just keeps a-ticking whether you are getting somewhere or just standing still.**

*-Author unknown-*

## Kypho's Corner



**G**reetings once more to all those with some connection to Lysosomal diseases.

It is not that often we get good news, so when it does come by it is important to celebrate it, and there are several bits of good news for LDNZ from the past few months. In this issue you will see information about a successful bid by LDNZ to hold the next Australasian conference in NZ next year. Jenny Noble put a lot of work into making a case for this and I'm delighted she has succeeded. Jenny has also done a huge amount of work leading a small team to get our Charity Dinner set up later in the year. More news about that in this issue too.

Additional cause for celebration came with the news that Dave Palmer, who is a Trustee of LDNZ and a researcher into Batten disease, has been awarded a full Professor post in recognition of his work on the important sheep model of Batten disease. Congratulations Dave for the work you have done and for your recent substantial research grant from the National Institutes of Health in the USA.

The development of a Carer Strategy is more good news. LDNZ has been working closely with 40 other not for profits to get this on the government's policy agenda and the recent announcement by Disability Minister Ruth Dyson that a strategy will be developed in partnership with the Carers Alliance, is a major step forward for all of us. An article in this newsletter gives more detail about this important development, which is certain to lead to better recognition and support for those who care for ill or disabled family members. Public consultation will begin in late May/early July and we hope you will all take the opportunity to make your voices heard.

On a different note, a major piece of research work was recently completed by the Access to Medicines Coalition and formed the basis of a submission to the Ministry of Health on the development of a medicine strategy for New Zealand. This is a very important piece of work for LDNZ and all families affected by Lysosomal diseases. LDNZ was a key driver in persuading government that a medicine strategy was needed, and we provided leadership in getting the research funded and a very substantial submission prepared. The final submission certainly has LDNZ's thumbprint on it. There is a strong focus on medicines for rare diseases, close scrutiny of Pharmac's decision making process, discussion of the ethics of treating rare diseases with high cost therapies (versus spending the money on lower cost treatments for larger numbers of people), and a case study of the Gaucher treatment problems.

Whether the medicines strategy will actually deliver improved access to therapies for our diseases is yet to be determined, but the great success we achieved was to work closely with 26 other not-for-profits to produce a document that is of critical importance to us, and which could not have been produced by us on our own. This certainly seems to be a case of the power of one multiplying hugely when in coalition with 26 other groups.

Yes, it certainly is a time for us to stop and (briefly) celebrate the progress that has been made in recent months, before we launch ourselves back into the fray to keep advocating for our families individually and collectively. Despite these recent successes, we know the beneficial outcomes will be some time away and a lot of continued effort will be needed to deal with the ongoing day-to-day problems our families face. With the very best of wishes to all of you.

John Forman  
Chairperson, LDNZ



## *LATE BREAKING NEWS !!*



### *Kiwis to host ANZAC Conference*

In the true spirit of the ANZAC tradition New Zealand and Australia are working together to improve scientific knowledge and clinical care for Lysosomal diseases. LDNZ will host a joint New Zealand and Australian conference in Christchurch in 2008. The first joint conference to be held in New Zealand will take some of the load off the Australian MPS Society which has run these conferences for scientists, clinicians and families every two years for the past few decades, with New Zealand families often attending the Australian events.

In a statement released today by LDNZ Chairman John Forman it was confirmed that LDNZ would organise the event and host their Australian cousins in Christchurch for the next biennial conference. "What makes it extra special is that it will be the **25<sup>th</sup> anniversary** of the formation of the Australian MPS Society. LDNZ will be honoured to recognise their important anniversary at this prestigious conference in New Zealand" says John. He went on to say "It is also a testament to the fine work done within the LDNZ back office lead by Jenny Noble. Jenny did the hard yards getting the proposal for this conference prepared and making the important connections that will turn this idea into a reality".

The conference is due to run over three days in **November 2008** and will include 25<sup>th</sup> birthday celebrations for the Australian Society, and guest speakers from New Zealand, Australia and the UK. "This is really important for LDNZ families as it will make access to such respected speakers as Professor Sillence, Dr Wraith and Professor Hopwood who are some of the worlds leading experts in Lysosomal Diseases, as well as focus on the significant contribution New Zealand scientists have made to knowledge of Lysosomal diseases over many decades." says Jenny.

## **LDNZ Announces Major Fundraiser 29th September 2007**

There has been a lot of brain storming, excitement, stress, fun, working lunches, bottles of wine, and laughter in putting together our first ever Charity Dinner.

Planning is well advanced, we have secured our guest speakers, our MC and auctioneers and sold 6 tables already. We are now working towards sponsorship and Auction items.

What started out as an idea is well developed and is going to be a fantastic night out with LDNZ raising much needed funds to help support the Australian conference in 2008 and research initiatives.

We are very proud to announce that we have the support of the Vodafone Warriors and hope to have some of their cheerleaders present.

Please visit our website for more information about the dinner. Check out Jack's meeting with Brendan Pongia. Jack had a wonderful day with Brendan.



Jo

Jenny

Kirsty

**Charity Dinner team**

## 'I'M A LITTLE WARRIOR, AND I'M BATTLING FOR MY LIFE'



### WE NEED YOUR HELP

Lysosomal Diseases New Zealand is holding it's first ever charity dinner to raise much needed funds for **research to help find treatments and therapies for our children** affected by these extremely rare and life threatening genetic diseases.

Date: 29<sup>th</sup> September 2007

Venue: The Sebel Trinity Wharf,  
51 Dive Crescent, Tauranga

Time: 6.30pm for 7.30pm start

Cost: Corporate table of 8 \$1,000  
Single Tickets \$120

Dress Code: Black Tie

Guest Speakers: MC - Duncan Garner (TV3)  
Guest speaker - Brendon Pongia (Good Morning host & Dancing with the stars)  
Guest speaker - Dean Bell (First ever Warrior & first Warrior Captain)  
Auctioneers - Frank Vosper and John Lamarson (Sponsored by REMAX )



Proudly supported by

#### **Mystery Prize – Swap your tickets for a key**

- Corporate tables of 8 will receive 2 keys per ticket
- Single tickets will receive one key.

One of these keys will unlock the mystery prize.

**Come and join us, have some fun and be in with a chance to win!**

## 1st Notice of Australian Batten Conference October 2007

The Australian Chapter of the Batten Disease Support and Research Assoc. Inc. *presents:*

4th Australian Batten Disease  
Family Conference



*When:* 26<sup>th</sup> October to 29<sup>th</sup> October 2007 (9am- 5pm)

*Where:* Outrigger on the Beach at Salt, Kingscliff, Northern NSW

*Accommodation:* The Outrigger holds a group booking rate per attached booking form for attendees (further viewing of the venue at [www.outrigger.com](http://www.outrigger.com))

**A Project of Hope....** The Chapter hopes that families, friends, doctors and researchers will come together in a wonderful and extraordinary way. Families travel from all states of Australia and parts of New Zealand.

As we are in the first stage of planning and it is very important that we know about you coming as:  
 We can allocate out funding to assist with your expenses;  
 The venue needs to secure you a room to best meet your requirements (per the attached booking form);  
 Our new group of volunteers (to be advised to you soon) want to begin organising the Kids Program, carers and guest speakers.

**Please help us to make the best arrangements for you by initiating your booking to the venue then we can liaise with you on the rest.** If we haven't received word from you by the end of February, it will be difficult to include you in funding assistance opportunities. A lot of voluntary organising is undertaken to put this together.

The Project aims to provide families and their friends, teachers and medical staff, affected or involved by this Disease with information, education and social interaction in various aspects of Batten Disease.

◆ The conference duration is Saturday and Sunday

Together with our BDSRA USA group, we organise for worldwide researchers to come here to share their knowledge, updating us on research and possibilities and hopes of trial treatments. A number of excellent speakers will travel from USA and Australia-wide to give each of us a greater understanding of this disease. The families' and carers from all fields, have a need for help in the form of knowledge, hope and practical ways of managing. Hence our second aim is having guest speakers who can form a panel group on the appropriate methods in which to provide the best care for these special children.

- ◆ The social time is at Friday night's orientation and Saturday night's dinner with entertainment for all.
- ◆ The Children's Program is operated during the conference period with packed activities of fun as a way for the children to interact in the care of volunteer carers and volunteer bereaved parents.
- ◆ Monday is an optional day for spending time with family and newly formed friends for a 'family day'.
- ◆ Sunday evening we hold a 'Remembering Service' in a nice location near the beach.

**This Conference welcomes the public with an interest in Batten Disease. You may wish to share this information with teachers, family and carers in case they would like to attend.**

**For more information please visit [www.battens.org.au](http://www.battens.org.au)**

## Amicus Therapeutics: Exploring novel ways to understand and treat Lysosomal storage disorders

Amicus Therapeutics is a biopharmaceutical company developing novel, oral therapeutics known as pharmacological chaperones for the treatment of a range of human genetic diseases. Pharmacological chaperone technology involves the use of small molecules to restore or improve biological activity in cells by selectively binding to misfolded proteins caused by genetic mutations. Amicus is initially targeting Lysosomal storage disorders, which are severe, chronic genetic diseases with unmet medical needs.

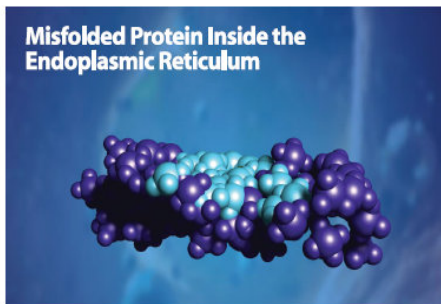
Historically, Lysosomal storage disorders (LSDs) such as Fabry, Gaucher, and Pompe have been described as genetic conditions in which enzymes that are needed to breakdown substrates in the Lysosomes of cells are missing or are only partially active. As a result, substrate accumulates in the Lysosomes. In most LSDs, this substrate accumulation is manifested clinically as an increase in the size of affected tissues and organs. Current treatment options are designed to address this understanding of the disease by replacing the missing enzyme or by reducing the amount of substrate that accumulates.

Recent evidence suggests that Fabry, Gaucher, and Pompe are part of a growing list of conditions that may be further classified, and understood, as disorders of protein misfolding and amenable to pharmacological chaperone therapy.

In a normal cell, specialized proteins called enzymes work to digest, or break down, substrate in the Lysosome. Normally, an enzyme is made of a specific sequence of amino acids that assumes a 3-dimensional shape by being twisted, bent, and folded. Enzymes are folded in a part of the cell called the endoplasmic reticulum (ER). Once properly folded, enzymes move out of the ER to other parts of the cell where they can perform their intended function. Certain types of mutations, or changes, in the gene that encode the enzyme allow the enzyme to be made but it is unable to fold properly. These types of mutations are called missense mutations. Misfolded enzymes are unstable and they remain in the ER where they may accumulate, clump together, and cause cell damage. The cell damage caused by accumulated enzyme in the ER is thought to add to the damaging effects created by storage of substrate in the Lysosome that is caused by the reduction or loss of enzyme.

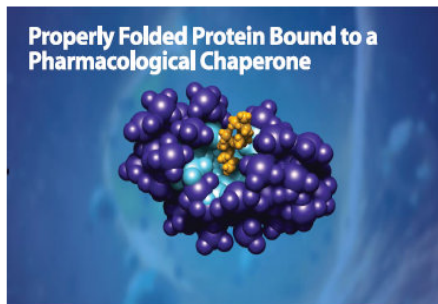
Pharmacological chaperones are small molecules designed to attach to the misfolded enzymes in the ER, to stabilize the enzymes so that they can fold into the correct 3-dimensional shape. Once the enzymes are folded properly, they can travel normally in the cell and reach their intended location. Once the stabilized enzyme arrives at its cellular target (which is the Lysosome in the case of LSDs), the pharmacological chaperone detaches, which is thought to allow the enzyme to perform its proper function. Amicus is exploring this novel approach as potential therapies for Fabry, Gaucher, and Pompe. Amicus is currently conducting Phase 2 clinical trials for its lead compound, migalastat, for Fabry disease, has completed Phase 1 clinical trials of AT2101 for Gaucher disease, and is conducting Phase 1 clinical trials of AT2220 for Pompe disease. Phase 2 clinical trials of AT2101 for Gaucher disease are expected to commence in Spring 2007.

More information about pharmacological chaperone technology and opportunities for clinical trials is available on the Amicus web site at [www.amicustherapeutics.com](http://www.amicustherapeutics.com).



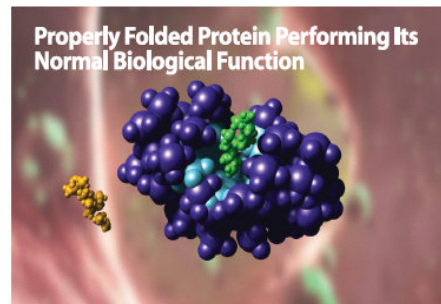
**Misfolded Protein Inside the Endoplasmic Reticulum**

Proteins, including enzymes, are formed in the cell's endoplasmic reticulum (ER), where they fold into a specific, three-dimensional shape required for the protein to work properly. Genetic mutations can prevent proteins from folding properly, causing the ER's quality control system to "flag" these misfolded proteins and send them to be degraded.



**Properly Folded Protein Bound to a Pharmacological Chaperone**

A pharmacological chaperone selectively binds to and stabilizes a mutant protein in the ER and helps it fold properly, thereby allowing it to successfully pass through the ER's quality control system.



**Properly Folded Protein Performing Its Normal Biological Function**

After leaving the ER, the protein-chaperone complex can be trafficked to its final destination in the cell, where the chaperone is displaced and the protein is now free to perform its normal biological function to reduce accumulated substrate.

## Expanded New Born Screening

**A major milestone was reached on 1 December when the Newborn Metabolic Screening programme added over twenty new tests to the seven previously performed on the Guthrie card blood spot taken from most newborn babies. This is a significant advance in improving the health of babies, with the new tests predicted to save the lives of about 3 to 5 babies each year, and significantly improve the outcomes for a similar number of others. The full programme will now produce life-saving interventions, deliver significant improvements to health and wellbeing, and avoid major disabilities, for around 40 to 45 babies every year.**

Dr Dianne Webster from the National Testing Centre tells us more:

New Born screening is done by a fancy instrument called a tandem mass spectrometry to measure many different markers (amino acids and acylcarnitines) in dried blood spots collected from newborn babies in order to detect inborn errors of metabolism. Until December 2006 testing was for seven conditions, now an extra 20+ disorders have been added.

This is possible due to the Starship Foundation gifting the tandem mass spectrometer or 'tandem'. Australia and the United States have been using the technology since 1998. The tandem allows New Zealand babies to be tested for the same disorders as Australian babies.

The new disorders are all inborn errors of metabolism due to inherited enzyme deficiencies. In all cases there is either a build up of a toxic enzyme substrate and/or a deficiency of the enzyme product, and the conditions are treatable with diet (including avoidance of fasting); metabolic precursors and/or vitamins. The new conditions are -

### 1. Disorders of amino acid breakdown – examples include:

Citrullinaemia  
Glutaric acidemia  
Methylmalonic acidemia

In these conditions waste products such as ammonia build up to harmful levels causing life-threatening complications. They are treated by special diets including extra vitamins. About 1:12,000 babies will have one of these disorders.

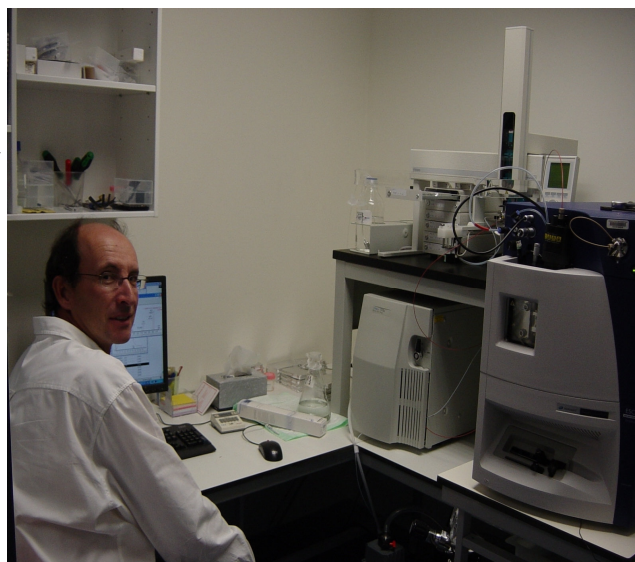
### 2. Disorders of fatty acid oxidation – examples include:

Medium chain acyl-CoA dehydrogenase deficiency (MCADD)  
Carnitine transporter defect

In these conditions energy cannot be used from fats. Without the energy from fats the body can run out of energy. The conditions lead to brain damage and life-threatening complications. The treatment is to ensure babies and children are fed regularly. We expect about 1:12,000 babies will have one of these disorders.

The screening programme has been very fortunate to recruit Detlef Knoll to run the tandem. Detlef is a scientific officer from South Africa with extensive metabolic disease experience.

Lysosomal storage disorders are inborn errors of metabolism. Research is occurring in some countries about using a tandem to screen for Lysosomal disorders in order for early treatment but so far only single-disorder screening e.g. Pompe disease is in pilot status. Lysosomal disorders have many different markers and it is complex making a test for all of them which finds most cases and has an acceptable retesting rate. Another tandem would be needed for NZ to screen, and we would also have to consider whether to screen when treatment is not always available, either because it isn't available worldwide, or not funded in NZ.



Detlif with tandem mass spectrometer

## Government Announces Carer Strategy



***John and Jenny attended the Carers Summit in Wellington on 12th—13th April and once again took our diseases out into the community. It was incredibly exciting to hear the Government finally announce that they will develop a carers strategy for those of us who are caring for our children and loved ones who have a disability or illness. LDNZ will be watching this strategy develop with interest.***

### Charities Support Government Focus on Family Carers

National charities have welcomed the Government's announcement that it will develop a Carers Strategy for the 15% of New Zealanders who provide care for sick or disabled family members. Non-profit Carers NZ established a coalition of almost 40 national charities two years ago to call for a Carers Strategy. LDNZ and the NZ Organisation for Rare Disorders are both actively involved in the Carers Alliance.

At a recent Carers Summit at Te Papa, Government Minister Ruth Dyson pledged the Government's commitment to a Carers Strategy, and announced a public consultation process to be coordinated in partnership with the Alliance. Carers NZ director, Laurie Hilsgen, says a review of existing policy and legislation confirmed what Alliance non-profits already knew: that the focus of New Zealand health service delivery is on the person needing support, without adequate regard for the needs of the wider family and those in 24/7 caring roles.

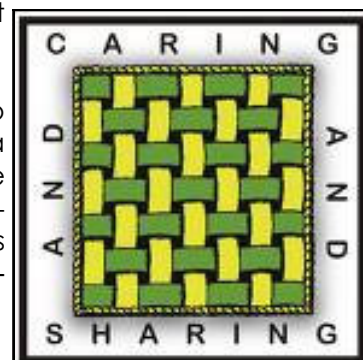
"There are good reasons why New Zealand also needs to care for its carers. The population is ageing, more of us are living for longer in the community as we grow older, and modern medicine is allowing us to better manage conditions and disabilities that once caused early mortality. These trends mean that New Zealand is relying more on family carers to provide unpaid care for loved ones. While caring is a traditional family value, making the choice to care can have lifetime consequences," she says.

"Often carers are unable to fully participate in the workforce. They do not nationally consistent training to undertake basic home health procedures. There is inadequate recognition of the needs of children and young people in caring roles. There is also a need to better understand that caring can impact on your own physical and mental wellbeing, and is an independent early mortality risk for older carers."

Legislation to achieve thoughtful support for carers is in place in the United Kingdom, where a strategy was championed by Prime Minister Tony Blair in 1999. Blair's father had a stroke when he was a child, and the experience influenced his support for progressive family care policies in the UK. In February the British government announced a review of the 1999 Carers Strategy, and new government measures to deliver improved services and support to carers. Australia is also progressive for carers, with legislation and services in place nationally and in all states; services for carers include access to free counselling, and programs for young carers.

Carers Alliance participants are all national non-profits which collectively support tens of thousands of families. "Our social fabric is woven from the principles that will underpin the Carers Strategy," says Laurie Hilsgen. "Helping families cope with trauma, be it for a short time or for a lifetime, is something New Zealand prides itself on. The Carers Strategy is a missing piece of public policy. We hope it will be supported by every political party and health agency, because sickness, injury, and disability don't discriminate. We can all expect to have caring experiences during our lives."

Carers Alliance non-profits will work with the Ministry of Social Development to organise a menu of ways New Zealand families and organisations can have a say in the Carers Strategy. LDNZ and NZORD recognise that these issues are very important to our groups, and we will be actively engaged in the consultation process. We look forward to ensuring the experiences of Lysosomal families are fully included in the discussions and have a positive influence the final strategy.



## Prof. David Palmer wins NIH Funding for Batten Disease

**LDNZ is delighted to announce Prof. Palmers success in gaining an NIH Grant to continue his work in Batten Disease.**

The United States National Institutes of Health (NIH) have awarded Professor David Palmer of Lincoln University a major grant, US\$705,000 to continue his studies of Batten disease for another three years. The proposed research is centred on a form of Batten disease in sheep and continues work that has been on-going for the last 30 years. The disease in these sheep is very similar to the CLN6 variant late infantile human disease and studies on them have proved invaluable in understanding the biochemistry and preclinical pathology of the disease. They have a human-like brain anatomy, physiology and genetic organization, and display similar neuropathology and a similar development of clinical disease. The overall aim of this project is to complete understanding to a point where trials of viral vector gene therapies can begin in combination with a test of the benefits of suppression of inflammation. We have defined the gene product required for correction and its sub cellular site of residence. Neuron cultures will be tested with gene therapy vectors to determine their suitability for introducing the corrected gene into sheep brain cells.

Prenatal and preclinical neuropathology studies have indicated that abnormal inflammation like cell activation begins early in brain development and has a primary role in the disease. Studies are aimed at determining the critical steps in the cascade of inflammation, and thus determine the most effective point for intervention. The effectiveness of chronic treatment with anti-inflammatory drugs on the development of the disease in sheep will be tested in vivo.

Prolonged genesis and migration new brain cells from a particular area (the subventricular zone) in the brains of affected sheep was also indicated in the pathology studies. The extent of this will be established by labeling of new cells in vivo and tracing their migration within the brain. Finally the usefulness of this migration for carrying a gene to the areas where it is required will be determined by targeted injection of the preferred vector carrying a reporter gene into affected sheep brains and detection of the dispersal of gene expression. These studies are aimed at providing all the requirements for directly testing gene therapy on the sheep, in combination with suppression of inflammation.

**REVELANCE:** This project intends to gain knowledge to define realistic therapies in a large animal form of Batten disease that can be tested for possible human use. Much of the knowledge gained will be relevant to other diseases and other treatment strategies, such as Alzheimer's disease and stem cell transplantation.

For the last 25 years Dr Palmer has been studying sheep with Batten disease having begun at Massey studying with Professor Jolly. Much of that work being funded by a succession of NIH grants supplemented by funding from the Neurological Foundation of New Zealand the Batten Disease Support and Research Association (USA) and some funding from LDNZ. This work is centred at Lincoln but also involves a number of high quality collaborators; Professor Richard Faull and Drs Stephanie Hughes and Henry Waldvogel in Auckland, Dr Imke Tammen in Sydney, Dr Jon Cooper in London, Dr Jaana Tynnelä in Helsinki and Professor Thomas Braulke in Hamburg.

He has used sheep because they have a human-like brain anatomy, physiology and genetic organisation. The development of the disease in sheep and humans is also similar, including the clinical course and symptoms and neuropathology.

Batten disease is actually a group of closely related diseases. They cause severe brain atrophy, blindness and seizures of increasing severity that lead to premature death. There are no effective therapies and even palliative care is difficult.

Professor Palmer has discovered neuro-inflammation plays a primary role in the development of the disease. These findings open up the possibility that control of cellular inflammation may be beneficial to patients, especially as an adjunct to gene or enzyme therapy.



**Affected North Hampshire ram in his own environment**

"This will allow more accurate targeting of therapeutic suppression. Starting with widely used anti-inflammatory drugs, trials will be conducted on the effects of pharmacological suppression of inflammation on the development of the disease."

The research will also further define the process by which neurons are created to populate the growing brain. This process, called neurogenesis, is a normal event in brain development, and drops to almost nothing in the mature normal brain. In the brains of mature diseased sheep it continues, perhaps in a futile attempt to generate replacements for the neurons dying in the affected cortical regions.

As well as being important for Batten disease, the research findings may also have relevance to other neurodegenerative diseases such as Alzheimer's disease, and other storage diseases such as Sandhoff disease.

NIH funding is prestigious and difficult to obtain, particularly foreign grants which require a unique opportunity to lead research world-wide. The NIH have determined that the research being undertaken into Batten disease by Dr David Palmer of Lincoln University is ground breaking enough to meet this high standard.

## LDNZ involved in Access to Medicine Coalition

### ***LDNZ joins 25 other not-for-profits in calling for an overhaul of New Zealand's medicine funding system***

Lysosomal Diseases New Zealand, in partnership with the New Zealand Organisation for Rare Disorders, played a lead role in the recent work of the Access to Medicines Coalition to win government agreement to develop a medicine strategy, and in the work involved in putting together a very substantial submission from the ATM Coalition on the medicine strategy consultation document prepared by the Ministry of Health.

ATM combines the voices of 26 non-government organisations advocating for increased access to medicines in New Zealand. Members of the coalition are all disease-specific groups that provide support, information/education, health promotion or clinical services to their constituent groups.

The Coalition has called on the Ministry of Health and government to completely overhaul the way medicines are funded in New Zealand, including stripping PHARMAC of some of its roles. ATM has criticised Pharmac's multiple roles as very confusing and a likely cause of many unsatisfactory outcomes in managing medicine access in New Zealand. Pharmac's conflicting roles were contrasted with the Justice system where there is widespread understanding and acceptance of the need to separate the roles of defence, prosecution and judge and jury. ATM submitted that unless there is clarification and separation of roles in medicine access, we will continue to have tension and confusion leading to ongoing frustration and an unsatisfactory medicine funding system.

ATM spokesman John Forman says a complete overhaul is necessary because fine tuning to the current system will only result in continued failure to ensure access to medicines for all people who need them. "It is inappropriate, for example, for PHARMAC to be in charge of assessing both whether a medicine is clinically effective and cost effective. A separate body should be set up for assessing the clinical effectiveness of medicines - as exists in Australia - as PHARMAC's primary focus is on cost at the expense of patient need."

In its submission to the Ministry, ATM is also questioning PHARMAC's input into setting its own budgets. Budgetary constraints are often used as an excuse by PHARMAC for not funding needed medicines, yet PHARMAC themselves are involved in setting these budgets. Patients are being told the cupboard is bare when PHARMAC consistently reports coming in under budget year after year. The system has to change. ATM believes that PHARMAC's dual role is causing decisions on clinical effectiveness to be confused with a product's affordability. Separating decisions on clinical effectiveness and affordability will create more transparent decision making about the adequacy of the total budget, and generate greater confidence in the health sector.

For a copy of ATM's detailed submission go to: <http://www.nzordgroups.org.nz/cms/imagelibrary/100402.pdf> or read more background to the saga of the medicine strategy development on the ATM website at: <http://www.atmcoalition.org.nz>



## LDNZ's WONDER DIET FOR STRESS

***This is a specially formulated diet designed to help LDNZ Mothers cope with stress that builds with caring for our wonderful children.***

### **Breakfast**

- ◆ 1 Grapefruit
- ◆ 1 slice of whole-wheat bread
- ◆ 1 cup skim milk

### **Lunch**

- ◆ 1 small portion lean steamed chicken with a cup of spinach
- ◆ 1 cup herbal tea
- ◆ 1 Tim Tam

### **Afternoon Tea**

- ◆ The rest of the Tim Tams from the packet
- ◆ 1 tub Gina Ginelli ice-cream with chocolate topping

### **Dinner**

- ◆ 2 bottles of wine (red or white)
- ◆ 2 loaves of garlic bread
- ◆ 1 family sized Supreme Pizza
- ◆ 3 snickers bars

### **Late night snack**

- ◆ 1 whole Foodstuffs Cheesecake (eaten directly from the freezer)

***Remember "Stressed spelled backwards is.***

## ***"desserts"***

### **Him and Her**

- ◆ A man will pay \$2 for a \$1 item he needs.
- ◆ A woman will pay \$1 for a \$2 item that she doesn't need.
- ◆ A woman worries about the future until she gets a husband.
- ◆ A man never worries about the future until he gets a wife.
- ◆ A successful man is one who makes more money than his wife can spend.
- ◆ A successful woman is one who can find such a man.
- ◆ To be happy with a man, you must understand him a lot and love him a little.
- ◆ To be happy with a woman, you must love her a lot and not try to understand her at all.
- ◆ Married men lived longer than single men, but married men are a lot more willing to die.
- ◆ Any married man should forget his mistakes - there's no use in two people remembering the same thing.
- ◆ Men wake up as good-looking as they went to bed. Women somehow deteriorate during the night.
- ◆ A woman marries a man expecting he will change, but he doesn't.
- ◆ A man marries a woman expecting that she won't change, and she does.
- ◆ A woman has the last word in any argument. Anything a man says after that is the beginning of a new argument.
- ◆ There are two times when a man doesn't understand a woman: before marriage and after marriage.

## Feedback / Donations

Please send us your feedback, your request for further information or make a donation to LDNZ.

**Jenny Noble**  
**16 Woodleigh Place**  
**Ohauiti**  
**Tauranga**  
 E-mail [jenny.noble@xtra.co.nz](mailto:jenny.noble@xtra.co.nz)

Donations over \$5.00 made to Lysosomal Diseases New Zealand are Tax deductible.

### What happens to the funds we raise?

Funding of all administration expenses for our group.

- ✻ Supporting families wishing to attend Conferences.
- ✻ Advocating for families for disability support, health services and access to therapies.
- ✻ Lobbying the Ministry for improvements to diagnosis, screening and care.
- ✻ Keeping in touch with researchers and biotech companies on research progress.
- ✻ Supporting some research efforts here in New Zealand.
- ✻ Keeping you informed of progress with our mission.

*Thank you for your Support*

Our very heartfelt thanks go to the following organisations and families who have provided grants since our last newsletter.

*Tim and Marianne Hannagan, Mike and Trudi Fermor, Carol-Ann and Shaun Morris-Eyton, Heather and Jack Bellaney, Mrs Fay Holmes,*



**JR MCKENZIE TRUST**  
ESTABLISHED IN 1940



**genzyme**

