

# LYSOSOMAL DISEASES NEW ZEALAND

December Newsletter 2006

## New Zealand Lysosomal Storage Diseases Support Group

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## 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.

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## Seasons Greetings



The Team at LDNZ wish you all a very  
Merry Christmas





## Kypho's Corner

Hello to all LDNZ families and supporters. We are pleased to send you the third LDNZ newsletter for 2006. In past editions we have called this section the editorial. Jenny thought that sounded a little boring so has chosen the name "Kypho's Corner". Kypho is the name we gave to our kiwi symbol at the LDNZ conference in Auckland in 2003. The name is associated with the kyphosis, or curvature of the spine, that is often a symptom of Lysosomal diseases as well as a distinctive feature of the kiwi too. We hope this gives a nice aspect to our mascot's name, and help make LDNZ a bit more human and friendly.

I'm writing this note just as we have completed our third and final family gathering for the year in Christchurch. This completes a very successful series of meetings that have enabled an average of 8 families at each of the three meetings to make better contact, learn from each other, and find support in our shared experiences. We are very pleased that these meetings have proved so useful to families. Just like the support we provided to get families to conferences overseas, they are another example of the ways we have been able to improve families' knowledge, reduce their isolation and help them become experts in managing their condition.

Providing direct support to families in this way has been a major focus of our work during 2006 and it is a good complement to the work we have done on some of the big picture issues like medicines strategy, working with the Carers Alliance, and lobbying Pharmac. Our ability to give this support to families is entirely dependent on grants from charitable funders and we are grateful for the significant support they give us.

A sobering reality is that so many of our families are having continuing difficulties with issues like disability support, welfare benefits and access to the right medical services. We are planning to convey these problems in a group to the appropriate agencies to see if we can get a better solution to the systemic problems that seem so persistent for us. We are very fortunate that we have such an energetic and efficient Secretary in Jenny Noble. Her work ensures we can uncover these problems and convey the information to the right people, as well as continue all the other important networking, publicity and policy work for LDNZ.

Check out our website at [www.ldnz.org.nz](http://www.ldnz.org.nz) to see the latest annual report online, along with new reports from families on their conference attendances, and some more personal stories. I hate to be the one reminding you that the holiday period is not to far away, but once you get over the shock of shopping for gifts and catering for special gatherings, make sure you take some time to relax and refresh yourselves for the year ahead, and may 2007 be better for you.

**Best wishes, John.**  
**Chairperson, LDNZ.**

### UPCOMING EVENTS

- ◆ **LDNZ Fundraising**  
Charity Dinner—April 2007  
Phantom Tea Party—  
**(Watch for more details)**
- ◆ **UK MPS and Related disease conference**  
29th June—1st July 2007
- ◆ **ISMED - 2nd International Scientific Conference** 26th—28th July 2007 Michigan

### NEW FAMILIES

**We warmly welcome the following families;**

Emma Graham whose stepson Marcus has Cystinosis

Ra Timms who's children Jordan and Brad have Batters Disease

Karla Donovan and Anthony Stroud who's children Tia, Willow and Saige have Gaucher disease

Heather and  
Allan Turner

## Brisbane MPS Meeting 28th Sept - 1st October



Twenty six adults, children and affected adults attended the 11th National Australian MPS meeting in Brisbane.

The theme for this conference was strengthening partnerships. There were many thought provoking workshops where families discussed the partnerships they had built over time with their Drs, researchers and support groups, and how these partnerships worked together for the good of the affected person.

The New Zealand families had easy access to all the presenters and they were able to spend time with the Drs talking through the many health issues their family members have. They came away with new information and a much better understanding of what they needed to do to improve the clinical care of their children.

One family said that they would now make the effort to attend future conferences as they found it incredibly useful in understanding what was happening to their son.

The affected adults also got a better understanding of their disorders and learnt that they needed to have careful management of their spinal issues – they stated no one had ever told them this.

Another mum said just being able to talk to another parent who had the same disease was incredibly beneficial, she said she no longer felt alone in the world.

### Further comments by our families

Heather and I attended our first MPS conference in Brisbane this year. We were very pleased to be able to go because it was our first chance to meet other people in our situation and to talk about things of common interest. It was great to meet these people and to swap email addresses so that we can keep in touch. It's very helpful to have other people to talk to and learn from and nice to be able to encourage them.

It was worthwhile meeting with the doctors and researchers dealing with these conditions. It was very informative to hear about the latest research and know what treatments are becoming available. It increased our appreciation of the important work that the Lysosomal Storage Diseases Support Group does in promoting awareness and advocating for action.

#### Heather and Allan Turner

We thought the doctors were once again just wonderful compassionate people who were so very accessible to everyone. A really positive outcome from the conference was probably putting our faces in front of the doctors again and hearing some up to date research info. It was also really wonderful seeing some of the New Zealand people.

#### Tim and Marianne Hannagan

*I would like to thank the New Zealand Lysosomal group, and the Ministers Discretionary fund for making it possible for me to attend the Australian MPS Conference in Brisbane. This is the first conference I have ever been to.*

*I am a foster mum to Blake who has Sanfilippo Disease. The information I gained from this conference was just fantastic, it is going to make caring for Blake so much easier. It was wonderful to learn that I am not alone in this journey with Blake. I made some new friends and now have many people to talk to when things get difficult. So Jenny and John what can I say but a very big thank you to you both.*

**DIANNE ROBERTSON**

Stacey Humphries  
and Jo Palairge





**The Gang**

Ready for some fun with the Australians on the sports night.



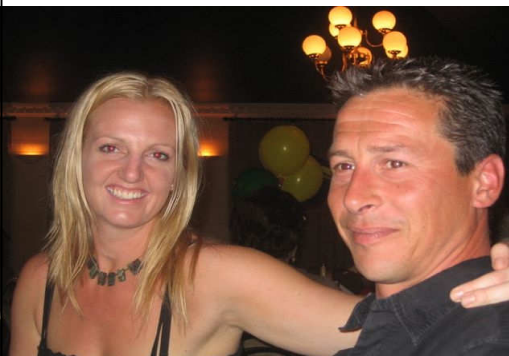
Evan Standish—painting with the children



**Rachel and Adam Lodewyk**

**Dr Ed Wraith and Jenny Noble**

**Front: Dianne Robertson, Marianne and Tim Hannagan  
Back: John Forman ,Allan Turner,**



## First International Education Conference on Batten Disease –Sweden 3rd—6th May 2006

**By Sharon Noble**

There were a lot of positives things coming out of this conference, especially in terms of the information gained and networking with other Batten families. It was a bonus to attend a conference that focussed solely on Batten Disease.

The main objective of the conference was to examine educational approaches for bringing optimal independence to students with Batten Disease. It addressed the need of consensus in the understanding of the educational needs and methods used in working with children and adolescents (and in some cases Adults) with Batten Disease.

There were also good opportunities to exchange ideas and experiences for development in the field of education

The presentations of research and use of technical solutions for use of children with various disabilities was interesting. Especially the talking cane and the lap top computers with shell program by SRRIDA Communication Centre.

Jonathan Cooper, Sarah Mole, and Paul Uvebrant gave clear descriptions of research being done in the area of Batten Disease. Gene therapy and enzyme therapy, although useful possibilities are being explored for other Lysosomal storage disorders, these do not appear to be useful in the treatment of Juvenile Batten disease.

Real progress towards a therapy for Juvenile Batten disease is frustratingly slow, but they have learnt some important lessons about where and when the brain is affected. There is some thought that the brain of Juvenile Batten Disease patients is leaking and that there may be a problem with the auto antibodies. Jonathan thought by combining this type of information with an understanding of precisely how brain cells are impacted by the disease will be the key for understanding how Juvenile Batten Disease may finally be treated successfully. It's just a matter of time. Unfortunately for most of our children time is not a friend!

A number of parents spoke on the affect the disease had on their child and what treatments or strategies had been best suited to the child and how they have coped with living with the disease. Moving towards dependence rather than independence. As expected, because each child, their disease progress, and family was unique to them, ideas of exactly what was done and when they did it varied, but one thing was very clear at some point all the child began to follow the same pattern and path.

I would like to thank LDNZ and the Ministers Discretionary fund for their financial support towards the cost of travelling to this very special conference.

To Denise Laraman a big thank you for the tremendous effort she put into organising the Comedy Night fundraising event and for both Mike King and Andrew Clay for the generous donation of their time and talent.

I will provide LDNZ with the resource booklets I have collected from the conference. This should be of value to other Batten's families and LDNZ to use when helping these families.

**Thank you once again,  
Sharon Noble.  
(Parent of a Juvenile Batten Disease child – Antonia Noble)**



## Natural History Study begins for Mucopolidosis Type II and III

By Denise Crompton  
USA

**Dr. Sara Cathey called it serendipity. We called it an answer to many prayers.**

A variety of efforts made by dedicated members of the medical community and dedicated parent advocates created the circumstances that led eight individuals affected with Mucopolidosis II & III and their families to the Greenwood Genetic Centre in South Carolina.

Since 1974, the Greenwood Genetic Centre has been a leader in the delivery of genetic services, diagnosing genetic conditions; counselling individuals and families; researching the causes of birth defects and genetic diseases; and educating health care providers, teachers and the public about genetics in the United States.

The lab at the Centre has started to compile the first ever Natural History Study for Mucopolidosis, having completed extensive testing on gene mutations of ML II & III. This will help the GGC, families, and future researchers to better understand the manifestations of this condition. Now, they want to analyse the information more closely to determine if specific gene changes are associated with specific clinical features.

Since the gene for ML II & III, known as GNPTA, had recently been isolated, this Centre believed that an in depth study with as many affected individuals as possible was important to undertake. We were all happy to participate. Once our lab work was done, and they established the markers in each participant within the GNPTA, they sent us questionnaires to be completed. The next step was for them to see Mucopolidosis II & III individuals in order to take photos, x-rays, and family histories.



**Front row:** Spencer, Gina, Robin, Jenny, Sergio, and Zachie

**Back row:** Kelley, Autumn and Brenda (Zachie's Mom)

The GGC staff were most gracious and accommodating to all of us. Everything was well planned, although we did run into a glitch with X-rays. We laughingly assured the doctors that we were accustomed to running into such problems.

We were treated to lunch and presentations by doctors explaining about the concept, origin, and growth of the Centre, as well as plans for expansion. GGC works with the South Carolina Department of Disabilities and Special Needs to provide diagnostic services, treatment, and prevention programs to reduce the risk and severity of disabling conditions. They are breaking new ground in clinical service, laboratory and diagnostic testing, and providing hope for every family at risk of genetic disease.

Dr. Mike Friez, Director of the Molecular Diagnostic Laboratory, used charts and analogies to assist us in understanding his overview of the concept of DNA. Some of us took notes (the human genome contains 30,000 to 40,000 genes), and later joked with each other about our

hopes that we wouldn't be tested. A tour of the labs helped us to more fully comprehend the way in which the genes were mapped. We were assured that they don't want to compete with the popular TV show *CSI*. It really does take much more than an hour to complete all the lab work!

This study of GNPTA is a genotype (genetic makeup of an individual), and phenotype (external appearance of an individual and their characteristics) produced by that individual's genotype interacting with the environment. In our private meetings with staff, we learned how very complex this condition is. This is the reason that we see a variety of ways in which it is manifested in different individuals.

It was difficult to say goodbye at the end of our gathering, particularly so for our precious children, who have quickly bonded with others like themselves. Sincere promises to meet again in the future were exchanged, along with warm hugs. We left knowing that a second group like ours would be meeting the following week to have an unforgettable experience in Greenwood, and that our world of ML II & III holds so much more promise for the future than it once did.

**Denise Crompton**

Author of *Kelley's Journey: Facing a Rare Disease With Courage*

**Sarah and Hayden Noble Mucopolidosis type III also took part in this study which came about due to collaboration with the Greenwood Genetic Centre USA and ISMRD of which John and Jenny are board members.**





## Living with Maroteaux-Lamy

By Stephanie Standish

**Stephanie writes about her life with Maroteaux-Lamy the pain she suffers from and her beautiful Art.**

As adults with MPS we are always told it 's ok you have the mild form of MPS 6 what does that really mean and does any child or adult with a Lysosomal disease have a mild form? I don't believe this is correct. Since I started going to conference's about MPS, I have noted that medical advances are helping children live longer with new medicines and now the all important gene therapy or enzyme replacement therapy.

It's hard to see parents going through the pain of diagnosis and the understanding of what their children will suffer as they go through the disease process. It is particularly hard to see friends and children with these diseases die.

Having had a child myself and loosing him to SIDS or cot death which I still can't quiet believe is the real reason he died I understand the parental issues better than most.

Having been told in my teens that **40 was our expected life expectancy**, this year has been a big one because I turned 40 and I am alive and well to my surprise and amazement. So to celebrate this year I decided to have an art exhibition of my works. I have been doing a diploma in arts and creativity for the last 3 years and loving it.

One of the major issues I have with MPS 6, has been the constant chronic pain that had not responded to the many drugs I have tried for pain relief. Some days I am so tired I just don't have the energy and want to sleep my life away. It is very hard to stay focused. If I go anywhere I have to make sure I have extra pain meds and be prepared to sleep a lot when I get home to recover. I find myself not wanting to go far these days.

Everything has a cost and my goal is to not feel too much pain and have time just to enjoy life and living without pain. However; I have been on Pamidronate now for 1 year which has turned my life around. Before taking Pamidronate I use to spend most of my days in bed. I most certainly would not have considered taking on my Art course. I am the first MPS 6 patient in the world to use this drug. My brother Evan is about to commence treatment he also suffers from much pain.

On June the 9<sup>th</sup> six days before my actual birthday the local Salvation Army Hall held my art exhibition and birthday combined. It gave me something to plan and aim for. I showed 24 paintings and prints and 3 small white clay sculptures and sold 6 pieces with a deal for 2 more and interest in some that were on show. I was happy to see my mothers pride in me.

This is a piece of carving on burnt rimu that I did for my younger brother to help him celebrate his Maori heritage.



These two paintings are my representation of what I would like to feel and look like but I am not, so I paint my ideas of the perfect womanly shape and figure.

So what are my thoughts and feelings - life is to short to be scared of living it. That doesn't mean it is going to be easy by any means but it does mean you are true to yourself.

If anyone asked me now— would I have had Ben my son if I had had any real understanding of the physical damage he did to me, I don't think if I had the knowledge I have now, that I would have gone through with that pregnancy. I was always worried about him seeing me go backwards never did I think or plan that he would go before me.....

***But I am glad I got to be some ones mum***

## Special thanks to Kirsty Peacock and Genzyme

LDNZ would like to send a great big thank you to Kirsty Peacock who has spent many hours with John and Jenny working through the designing process for our publicity package.

Kirsty also runs her own business Oncue Graphics and is the mother of Jack who has Hunters disease. Jack is seen on the bottom of our banner.

Our publicity package now consists of a banner as you can see here, new family handouts, brochures for Drs and other medical professionals, and a poster showing the Pattern of Inheritance. Along with all the publicity material we also have letterheads and business cards to complete the package.

We are currently working on a poster showing our National and Global connections and early next year we will start work on the science posters showing how animal models are also affected by Lysosomal Diseases and how they correspond with human disease.

We gratefully acknowledge the financial support of Genzyme. Without this support we would not have been able to complete this project and take our diseases to the Medical Profession in New Zealand.

If you want to help LDNZ lift our focus and would like some of our handouts and brochures please contact Jenny by phone or e-mail.

Phone 07 544-8868

e-mail [jenny.noble@xtra.co.nz](mailto:jenny.noble@xtra.co.nz)

**LDNZ**

**SUPPORTING FAMILIES WITH LYSOSOMAL DISEASES**

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

**WHAT ARE LYSOSOMAL DISEASES?**

- Inherited genetic defects
- Physical and/or neurological symptoms
- Serious life threatening diseases
- Approximately 100 New Zealanders are affected

**HOW DOES LDNZ SUPPORT FAMILIES?**

- Improves knowledge and information which enables families to become 'the experts'
- Family meetings
- Practical support / legal advocacy
- Contacts families in NZ and internationally

**WE ADVOCATE FOR**

- Access to health services
- Funding of treatments
- Policy change for transition to adult services
- Improved access to palliative care
- Research funding
- Improved access to disability support and social services

**LDNZ SUPPORTS RESEARCH**

- On NZ's unique strengths with liposomal enzymes
- Through grants given to New Zealand researchers
- By advocating for more research funds

LDNZ held yet another successful Family gathering In Christchurch

## Family Gathering Christchurch

over the weekend of 10th—12th November. Seven families gathered at the Camelot Motor lodge for a weekend of sharing and learning from one another. We were honored to have Dr David Palmer from Lincoln University join us and present to the meeting his latest research on Batten disease and where they are heading in understanding this very complex disorder. David also talked about the success of a significant funding grant he gained from the National Institute of Health in the USA for Batten research here in New Zealand.

Cecelia Gillard from the Neurological society also joined the meeting. This give us the opportunity to discuss with her the neurological issues some of our Lysosomal children have. Cecelia found our meeting very useful and I am sure we will be collaborating more with this organisation in the future.

On behalf of all the families who have had the opportunity to attend one of these meetings we sincerely thank the Todd foundation for their very generous support. Without this funding we would not have been able to have these meetings and make the connections among our families .

Evan Standish—MPS6  
Sage—Gaucher disease



## ERT- Elaprase approved by FDA for Hunter Syndrome



Shire has announced that the US Food and Drug Administration (FDA) has granted marketing approval for idursulfase (ELAPRASE) an enzyme replacement therapy for the treatment of Hunter diseases, also known as MPS II.

Hunter disease is a rare, life-limiting, genetic condition, mostly affecting males, caused by the deficiency of the Lysosomal enzyme iduronate-2-sulphatase. Without this enzyme, cellular waste products accumulate in the tissues and most organs. Hunter disease comprises a wide spectrum of severity and some children with the sever form of the disease have progressive developmental delay and usually die before reaching their mid teens whilst others have an attenuated form of Hunter disease suffering little or no central nervous system damage and can live well into adulthood. In all cases, Hunter disease causes progressive cardiac disease and physical disability and makes it very difficult for adults to live independently.

We watch and wait here in New Zealand with interest. This is yet another therapy that Pharmac will withhold from our families and will require much advocacy on LDNZ's part.

### Hunter Diseases—Did you know?

Charles Hunter who gave his name to Hunter Syndrome first described the condition in two brothers in 1917. After graduating from the University of Aberdeen, Charles Hunter undertook postgraduate training in London and Berlin before emigrating to Canada where he settled in Winnipeg practicing at Winnipeg General Hospital and specializing in internal medicine.

During the First World War he served in Europe as an army medical officer and in 1910 was appointed to the Faculty of Medicine at the University of Manitoba. In 1928 Charles Hunter was appointed Professor of Medicine, However he was unhappy with the tedious administrative responsibilities and resigned the following year.

Charles Hunter who was regarded as the leading diagnostician in Western Canada continued teaching and retained his private practice until only a few years before his death in 1959 at the age of 82.



## LDNZ and NZORD attend Paediatric meeting - Nelson



LDNZ and NZORD (*The New Zealand Organisation for Rare Disorders*) attended the Paediatric Society meeting from 31st October - 3rd November. This was a wonderful opportunity to have a focus for our diseases.

It gave us the opportunity to ask Drs if they had seen or treated a Lysosomal disease,? Did they know about the Metabolic Service? Would they make referrals to the service? We had a power point presentation running during their break time with the faces of Lysosomal diseases, so thank you to everyone who sent us a photo of your child.

We had the opportunity to speak with Pat Touhy from the Ministry of Health and two Pharmac representatives and really get our issues out there.

We head to Rotorua to attend the Medical Sciences meeting 28th Nov—1st Dec. Both John and Jenny are presenting at this meeting and will have a booth to once again bring attention to rare diseases.



## 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.



*Courage is the art of being the  
only one who knows you're  
scared to death.*

**Harold Wilson 1916– 1995**



*Thank you for your Support*

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

**T G McCarthy Trust**



**genzyme**