



Lysosomal Grape Vine

Newsletter

October 2010



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.

In this issue

- **A call to Action**
- Progress report on LDNZ's campaign for Enzyme Replacement Therapy
- LDNZ letter to Minister of Health and his reply
- Living with Pompe disease – by Laurie Hill
- Report on International MPS and Related diseases symposium – Adelaide June 2010

LDNZ's Charities Commission Registration
CC24962



Dr Ed Wraith receives

Life time MPS Award see page 12

Oliver Lodewyke at the entrance of the Gala Dinner see conference report.





A CALL TO ACTION FOR ALL LYSOSOMAL FAMILIES.

In recent months Jenny and I have been putting together ideas for a special report to cover the first ten years of LDNZ's existence. In fact we have been around a little longer than that, but it was 10 years ago that we were formally registered as a charitable trust.

At times like this it is tempting to focus just on the positive things we have achieved, either on our own, or in partnership with other rare disease groups. There have been real gains in winning support for the establishment of the Metabolic service, improvements to Genetic services, and some important progress in improving disability support and home help for a number of our families. We also made a significant contribution to the Royal Commission on Genetic Modification and other major policy discussions, ran some very successful conferences, helped lots of our families to get to overseas meetings about their disease, helped the drive for a Carers strategy and a Medicines strategy, and put a lot of work into trying to improve access to specialist services for families across the country.

But the one thing that has not progressed so well has been the task of winning funding for the new therapies for Lysosomal disease patients. True, we did succeed in getting Pharmac to improve dose levels for some Gaucher patients several years ago, and last year we had a significant breakthrough when they agreed to fund enzyme replacement therapy for Jack Peacock who has Hunter disease.

But if we only trumpeted loudly the positive things, we know we would be in denial of the major prize that has escaped our grasp so far – funding of the new therapies that are now available for another four Lysosomal diseases; Hurler-Scheie, Fabry, Pompe, and Maroteaux-Lamy. We know this is the prime goal of all the affected patients and their families, and is strongly supported by those who don't yet have a therapy, but where a therapy is being developed for their disease.

So instead of concentrating on a celebration of the past decade of LDNZ, we are issuing a **call to action from all Lysosomal families**. The timing is right for this. The election manifesto of the National Party promised to work with stakeholders to investigate ways to improve access to high-cost highly specialised medicines. Government appointed a panel to review this topic, and although the panel's recommendations are disappointing (see story on page 3), further work is continuing on this. The Minister of Health has made public comment that he intends to make decisions about funding of such medicines before the end of this year. Tony Ryall is also quoted as saying: "The Government is determined to see New Zealanders get better access to medicines, and I will be interested to see the feedback from consumers, as well as the health and pharmaceutical sectors." He has also asked Pharmac to review the exceptional circumstances scheme which deals with medicines that are not currently on the subsidised pharmaceutical schedule.

LDNZ believes the time is right, and that there has never been a better time, for all of us to get active in approaching Members of Parliament to push for improved access to medicines. In particular, we should be aiming to visit every member of the National Party caucus to make them aware of the importance of this issue, and press them to support Tony Ryall's initiative to improve access to medicines. We also want them to know what sort of things should be included in the criteria for exceptional circumstances consideration, so that our patients and families are not left out by tight criteria and unreasonable rules.

Between now and the end of November we'd like every one of you to visit your local National MP, or, if your local MP is from another party, a National list MP who provides some representation for your area. We think it is important to concentrate on National MPs because it is their Party that made the election promise to improve medicine access, their Minister who has responsibility for the health portfolio, and their administration that controls the budget. You are very welcome to brief other MPs from other parties on this as well, but we strongly urge you to ensure National MPs are lobbied first.

Jenny and I will prepare a summary of facts and figures about our campaign. We are keen to hear from you about the specific meetings you plan to organise, and we are happy to help you sort out any questions about MP contact details, etc. Please start preparing for this lobbying campaign. The health of those affected by Lysosomal diseases, now and in the future, depends on success with this campaign.

John Forman
Chairperson LDNZ

Progress report on LDNZ's campaign for Enzyme Replacement Therapy

Here's a summary of progress with our ongoing campaign to win funding for enzyme replacement therapies for Lysosomal diseases. We have been pushing hard for this for many years now, and there has been some limited progress, getting a medicines strategy in place in December 2007, and an extra boost to medicine funding in 2009. These should have resulted in improved chances of ERTs being funded but success has been limited to just one additional Lysosomal disease, Hunter disease, being funded last year.

Our work over the years did help win a promise from the National Party to review access to high-cost highly specialised medicines (HCHS), and in 2009 Health Minister Tony Ryall appointed a panel to advise him on how to improve access to these medicines. Our hopes for a quick solution were dashed when the panel's report a few months back looked at the "big picture" of medicines funding, and to some extent the entire health system, and suggested that improved access to HCHS medicines is most likely to come from improved efficiencies across the board. While that may be true in the long term, it did not provide us with a solution for now. However the panel did suggest that a revised Exceptional Circumstances scheme would best serve this group of medicines.

LDNZ was very frustrated with the outcome of the panel's review. It had some useful commentary but the review did not adequately address the issue of how to fund access to these medicines. We wrote to the Minister (see letter page 4) and also conveyed to government that the issue needed a solution such as:

- To focus on the narrow group of medicines that are difficult to assess by standard funding criteria, rather than looking as widely as the panel did. A narrower focus will mean quicker action to solve these problems.
- If the exceptional circumstances scheme is used it needs a better set of criteria and obviously a budget so the intention to provide for these patients can be carried through. An alternative could be the second tier approach taken in Australia, whereby if a new medicine fails to meet standard cost utility analysis (CUA) criteria it goes for consideration in the life-savings drugs programme, but an improved exceptional circumstances approach may be quicker and easier to implement.
- The need for ethics, equity, community values and fairness to be more specifically addressed.
- Criteria for consideration of high cost medicines should not focus so much on their price (that is a useful short-hand description but the policy approach needs to be more refined).

We have suggested a set of criteria that could be used in deciding if certain medicines should get such special review, with the combination of these factors helping to decide what is exceptional for the particular medicine.

These would be:

- Rarity of the disease
- Small patient numbers to be treated
- Complexity/severity of the disease
- Complexity of drug discovery and manufacture
- Initial CUA(cost utility analysis) puts it well beyond expected range or typical drug costs per patient
- Difficulty in getting high-grade evidence of benefit, meaning alternative evidence levels may be used

Tony Ryall's reply advised that Pharmac will now do a review of the exceptional circumstances scheme. He also said that he intended making decisions about funding for such medicines before the end of this year. Are we in the home straight? We might be, and that is why we are asking all Lysosomal families to join in a campaign of lobbying National Party MPs about the issue. We want maximum effort between now and the end of November to help secure the sort of policy and funding that is indicated by the Minister's statements.

Our letter to health Minister Tony Ryall about the Panels report on high cost highly specialised medicines.

Rt Hon Tony Ryall
Minister of Health
Parliament Buildings
Wellington

Dear Minister Ryall,

Funding of specialised therapies for patients with Lysosomal diseases.

You will be aware of issues with access to enzyme replacement therapies for this group of diseases. LDNZ is very pleased to note that our representations to you and your colleagues did result in young Jack Peacock starting treatment in July last year. Your approach to this treatment and funding of other medicines has demonstrated a willingness to look at issues wider than narrow cost-utility factors. We are delighted that there has been a change of heart about total medicine funding available and also about patients getting a chance to access these specialised medicines.

For the second half of last year we held back from any major lobbying about the issue of access to these specialised medicines for rare diseases, because we were aware of the panel you had appointed to advise you on how to improve access to such medicines. We directed our energy towards their work in the hope they would find the practical solutions you sought from them.

However we are writing to say we do not see much help in the panel's report to you, of practical ways to ensure improved access in the near future. We expected the panel would focus on the narrow group of medicines that are difficult to assess by standard funding criteria, rather than looking so widely at the whole health system, as the panel did. A narrower focus could have meant quicker action to solve these problems.

We are concerned that the proposal to use the exceptional circumstances scheme does not carry with it any recommendations for a better set of criteria or guidance on budget issues so the intention to provide for these patients can be carried through. An improved exceptional circumstances approach may be quicker and easier to implement but it must have more realistic rules and a budget to make it work for the medicines and patient groups it would be intended for.

We were also very disappointed by the lack of quality discussion in the panel's report about how issues like ethics, equity, community values and fairness could be more specifically addressed. Their commentary on ethics was a very superficial overview, but even then they reached no practical conclusion about it.

It is worth noting that in many cases the total cost of one of these novel medicines to the health system may not be very large at all, despite high individual price, because of small numbers of patients.

All LDNZ wants is a fair go for the patients we represent. The system has not produced a fair go for Lysosomal patients in the recent past and while we are delighted to see that Jack certainly has got his fair go, we don't see a path forward from the panel on how this fair go can apply to the rest of the Lysosomal patients who are waiting for treatment

To get this we think there should be criteria to trigger a special review of medicines that are well outside the usual range of medicine prices, for those that also have very low numbers, require complex development processes, apply to complex and serious diseases, and for which there is limited clinical experience.

And Pharmac's decision criteria should be changed to explicitly include fairness as a matter for consideration. That would require them to specifically address the non-monetary and non-clinical decision criteria set out in Medicines New Zealand, but which have never been specifically acknowledged or addressed by Pharmac.

We thank you for the work you have done in your time as Minister to address some of these issues, and urge you to continue this with a decisive solution to this long-standing problem.

Yours sincerely,

Jenny Noble
Secretary
Lysosomal Diseases New Zealand

**Office of Hon Tony Ryall**

Minister of Health
Minister of State Services

16 AUG 2010

Ms Jenny Noble
Secretary
Lysosomal Diseases New Zealand

Ref. no. 10001351

jenny.noble@nzord.org.nz

Dear Ms Noble

Thank you for your letter of 18 June 2010 about funding for specialised therapies for patients with lysosomal diseases.

By and large, PHARMAC is successful at achieving value for money in medicines spending and improving access to medicines. PHARMAC considers nine decision criteria when making funding decisions, and several of these are considered when the cost-effectiveness of a pharmaceutical is determined. PHARMAC's criteria include health need, the availability and suitability of existing therapies, and clinical benefits and risks.

The High Cost, Highly Specialised Medicines Panel Review Report suggested improvements to the funding of all medicines. The panel thoroughly considered the issue of high-cost highly specialised medicines, and concluded that while there are no easy solutions, there may be an opportunity to make beneficial changes to the Exceptional Circumstances (EC) Schemes. I note that current EC funding is not only for high-cost, highly specialised medicines, but for pharmaceuticals (high-cost or otherwise) where the patient's circumstances were not taken into account for funding under the Schedule and they meet other relevant criteria for EC funding.

I recently announced the Review of PHARMAC's EC schemes and encourage you to respond to its discussion document. More information is available from PHARMAC's website (www.pharmac.govt.nz/ecreview). The review will be completed in early 2011.

You also mentioned enzyme replacement therapies (ERTs) for lysosomal diseases. The Pharmacology and Therapeutics Advisory Committee (PTAC) is an advisory committee that considers PHARMAC's nine decision criteria when advising the PHARMAC Board on funding decisions. PTAC's assessment includes considering benefits and risks of treatments for patients from a clinical perspective. I note that PTAC has reviewed ERTs on several occasions and concluded that they have limitations. In relation to lysosomal diseases, PTAC has commented that ERTs do not compare well to other pharmaceuticals in terms of clinical evidence, as well as cost effectiveness.

Yours sincerely

Hon Tony Ryall
Minister of Health

Living with Pompe Disease

By Laurie Hill

Being diagnosed with any disease is incredibly devastating, but when it is an orphan disease it offers so many more complications. It appears there are no real benchmarks; there are not dozens of specialists with an interest in the disease, support groups, or a multitude of books written by knowledgeable experts, sufferers, or those that have struggled and survived. Most of the time it feels like you are entirely alone with the struggle. Mourning the continued loss of what used to be and now never will. Too afraid to open conversation with family members as they too struggle with the debilitating effects of this relentless disease. There is no way to gauge how well you are doing with the fight just the realisation each day that the fight must go on. Sometimes it is hard to know why.

I was first introduced to Pompe Disease in 1990 when after a barrage of tests and doctors I was informed I would be lucky to survive the year. A few years later I realized the doctor passing on this wonderful news was in fact confusing the adult late onset, that I had, with the infantile strain.

At the time I had two young children and a desire to be treated as 'normal' - well at least not to be totalised by a disease therefore losing my own identity. I decided that only those that had to know would know and I would continue life as 'normally' as possible for as long as possible. After about 12 years (so much for only surviving 1 year!!) I was contacted by a family member who had heard John Forman from LDNZ speak on the radio, and thought that maybe I would want to hear more about what he had to say. We looked him up and followed the links from the website and discovered there was an International Pompe Association and that they were having a conference to discuss a range of possible treatments. For the first time in over 10 years I felt the slightest twinge of hope. Although the conference was in Germany we managed, with a great deal of help to attend and the news of enzyme replacement and gene therapy was so exciting. Also to be around other people who Pompe had attacked was unbelievable. Finally I felt I was not alone in my struggle – even though I had to cross the world to gain that feeling.

A major outcome of that trip was the discovery of an internet group (GSD Net) that specialized in Pompe Disease and whereby patients, doctors, and families can post findings, ask questions, or just generally support each other in this battle. I came home with a new attitude of support and a lot more knowledgeable about what to expect in my future. The biggest hope was that there would be a treatment - it was just a matter of time!

About 4 years ago the FDA and other drug authorities around the world approved a drug called Myozyme and a large part of the Pompe population began treatment. The GSD Net became swamped with stories of who was getting treatment and what difference it was making to their lives. Understandably people were extremely excited and hope for a different and better future became the general topic of conversation. Patients began posting comments like:

"Four years ago I added Myozyme (aka Lumizyme) to my treatment. Since that time, my pulmonary function has stabilized, and in combination with all my physical therapy I have gained strength. Myozyme is not a cure, but over the long term I feel like it has made all the other work that I do much more beneficial. I even started skiing again!" - Hillary

Children diagnosed with Pompe were previously told they would not make 7 years but now their parents are being told with the help of Myozyme that they should start saving for college

In New Zealand however, the news is not so great. Whilst Myozyme is an approved drug there is no way the government will assist with the cost of treatment. What once was a time of hope for my family, friends and I, that treatment would arrive and at the minimum halt the progress of the disease, has now turned to one of frustration and disappointment. It is hard to know that for no fault of my own I have to watch my body slowly die when there is a way to prevent this happening available but for economic reasons Pompe sufferers cannot access it. I find myself thinking about the idiots that get drunk and wrap their car around a pole and require a few hundred thousand dollars worth of treatment to save their lives. Naturally there is no hesitation to do this, there is no choice. But for me to obtain the same care to save my life is not possible – because there is a choice!

The GSD Net that once provided a feeling of comfort, care and advice I now struggle to open and read. The daily postings of those who speak with excitement of how their life has changed as a result of treatment now haunt me. Not with any malice towards the writer but their words serve as a constant reminder of what could be but by all accounts never will be. I am forced to appreciate how quickly my body is deteriorating as theirs is improving. The group I once felt part of now separates and distances me by the recognition that they travel towards life and renewed hope whilst I do the opposite.

I struggle to understand how a country I am so proud of can decide to allow this disease to continue to inflict its cruel and painful onslaught on my body. If there was no hope, no possibility of change - as sad as this would be, it would be manageable. The fact that due to financial reasons my country is telling me that it will not offer me the chance for a better future I cannot believe. It is the hardest thing to understand how one person can be told they are worth saving whilst another is told they are not.

Now each day I try my hardest to focus on dealing with the challenges I must overcome to enable myself to be able to contribute to the many New Zealanders I work with. I try not to think about how much more I could do with the assistance of a treatment that has proven itself to be the salvation I once only dreamed of. Now I appreciate it may never be more than just a dream as I had the misfortune to not only be born with a disease but also to be born into a country that does not care enough.



LDNZ wishes to thank our sponsors for their very generous support.



11th International MPS and Related diseases Symposium Adelaide 24th -26th June 2010



The day the 2010 committee had been working towards for 2 ½ years finally arrived, with delegates from 38 countries arriving in Adelaide to take part in the 11th International MPS and Related Diseases symposium.

The symposium began with a Taste of South Australia welcome reception which saw the exhibition hall themed to showcase South Australia. The food and wine was supplied by various companies throughout the region. The delegates were tantalised with Kolas, a snake and a cockatoo. Delegates were able to touch and have their photos taken with the Kolas and snake. It was a wonderful opportunity to catch up with old friends and the scene was set for what was the beginning of three long days of conferencing.

Thursday 24th June. The symposium began with a Joint scientific and family session. Mark Haskins from the USA began the session with his topic of Looking forward from looking back but in his usual style changed the title to ***Back to the Future***. David Oliver from the Australian MPS Society followed Mark with the same topic from a family perspective. David's talk brought tears to a few eyes, the presentation is reprinted below:



[Setting the Scene – Looking forward from Looking back](#)

If life were a road map, then the MPS road would be one of those faint lines, dotted, twisting and turning, travelling over mountain peaks and descending deep into gullies and ravines. It would split into side roads, cross over highways and visit remote towns, seldom visited. It would have warning signs and obstacles across its path. It would definitely be the road less travelled.

For MPS families this scenario fits very well. Nearly all of us have found ourselves on the MPS road, not by choice, but by chance. This is the chance and lottery that genetics brings to our lives. We are here because someone we love, a child, a sibling, a grandchild, friend or relative inherited something that we have previously known nothing about. I am just one traveller along that road. My wife Christina and I have been very fortunate to have three fantastic children. That our youngest child is MPS affected was completely unexpected, and took us on a detour off the highway of life.

This gathering is one of the towns on that road map. We are all MPS travellers. We all have stories to tell of how we arrived here and where we think we are headed. Some of us are guides, and some are guided. I would like to welcome all of you and thank you for travelling (some of you a very long way) to be here. It is an honour to be up here before some of the legends of our community from families to professionals and scientists.

It has been a long and difficult journey in the development of the family networks that exist within MPS. The Australian Society is now only just over 25 years old, as are the UK and USA Societies. The early days of our Society now seem primitive in their ability to provide services to families and professionals. Throughout the last 25 years the ability of societies to meet our families' needs has developed enormously. As a society most families are able to reach assistance simply by a single phone call. The development of the internet and email over the last 10 years has provided our Society with a means to be accessible to anyone in our country or around the world. The ability to communicate with other societies around the globe has gone from fragmented and tenuous contact, to being fully engaged.

This symposium provides a unique opportunity for us all to gather together, to learn and to support each other, and celebrates the strength that is in each of us. The strength to care, the strength to study and think, the strength to persevere, and the strength to endure and sacrifice. We are all here because we love that person that is affected. That

love is the fuel in our tank. Love is the fuel that keeps us running along the road to what ever destination we arrive at. We are all on the road less travelled, and for most of us it is the journey, rather than the destination that gives us rewards.

After such a poignant start to the symposium the meeting broke for an extended morning tea break giving everyone time to chat and mingle. Moving forward through the next few days the symposium broke into two meetings of scientific and family programs.

[The scientific program on day one looked at:](#)

Diagnostics

- The importance of accurate diagnostics and the changing scene.
- The diagnostic delay in MPS 1 patients – The Latin American picture.
- Screening patients referred to a metabolic clinic for lysosomal storage disorders.
- Identification of potential urinary biomarkers for the management of patients with Fabry disease,
- Towards proteomics to identify biomarkers for MPS 1

Prognostics

- Prognostication of clinical course after early diagnosis: do we have the proper tools
- Discovery of biomarkers and improved diagnosis, as illustrated by Gaucher disease

Population Screening

This topic looked at the different ways to screen for Lysosomal disorders and was followed by a strong debate on are we ready to screen newborns for lysosomal storage disorders?

[Family program:](#)

The big picture: We welcomed Jim McGill (Australia) to the podium to open the family program and set the scene for the next few days with his presentation of an Overview of MPS and highlighting the key challenges for the future, which were the nervous system including the brain and bone.

Jim was followed by Lorne Clarke (Canada) who presented Unfolding clinical events following a diagnosis of MPS.

- This presentation provided an overview of the clinical events that unfold after a diagnosis of MPS with particular emphasis on disease heterogeneity and the factors that underlie various potential disease complications.
- Dr Jules Leroy (Belgium) talked about the Genotype Phenotype correlation in the MPS's using MPS 1/ Hurler Scheie and Mucopolidosis as examples. He talked about the Mucopolidosis experience of 63 ML patients who went through gene mutation diagnosis and Natural Histories to understand the natural course of the disease and to understand the phenotype correlation of ML II and ML III.
- Simon Jones (England) talked about the therapeutic approaches for MPS: Stating that the last 10 years have been dramatic for the MPS disorders. Until 2003 and the licensing of Aldurazyme for MPS 1, the only treatment hope was bone marrow transplantation. While BMT is the standard of care for severe MPS1 patients it seems less effective for MPS II, III and IV. He went onto to talk about the success of both ERT and BMT in these disorders. He said there is still much to learn about BMT and why it didn't work for MPS II or III. He said the currently licensed ERT's for MPS I, II and VI are all clearly effective at improving endurance and lung function testing in the clinical trials. Subsequent clinical experience has demonstrated efficacy in a variety of other tissues such as airways and soft tissue around joints. Outcome for bones and joints remain less clear with both ERT and BMT. Other modalities of treatment are at a less advanced stage with the prospect of gene therapy trials in MPS IIIA and B likely this year.

- Joanna Wilson-Smale gave a wonderful presentation about her experiences of living with MPS I and being treated with ERT. Joanna began ERT at the age of 17 years and saw immediate benefits such as a decrease in the size of her liver and spleen, increased mobility and a dramatic reduction of pain throughout her body. She said that with benefits there is usually a price to pay. For Joanna it meant travelling 200 miles and 5 hours for her infusions and all this was done during her A-levels. Joanna now receives her treatment at home giving her family the flexibility to try and lead a normal life.

Managing the Skelton

The big issue for many patients with MPS and related diseases is skeletal problems of the spine to hands, hips, knees, feet, ankles, including carpal tunnel and trigger fingers.

- Prof David Sillence (Sydney) set the scene by talking about the progressive and characteristic skeletal pathology which reflects storage in cartilage cells, impairment of cartilage growth and repair, bone remodelling and bone structure. He talked about Mucopolysaccharidosis and its destructive bone pathologies which have been demonstrated to result from abnormal sensitivity to the bone remodelling effects of parathyroid hormone. He said this pathology possibly affects some older patients with MPS II and MPS VI. He said the cartilage involvement in MPS also increases the risk of various processes such as aseptic necrosis (death) of bone in, for example, hips, which is seen in increased frequency in Sanfilippo. He talked about strategies to manage skeletal complications which depend on high level awareness, regular imaging studies and the use of selective medial and surgical approaches.
- Dr Andrew Cree (Australia) talked about the surgical management of cervical and thoracic lumbar spine.
- Bruce Foster (Australia) talked about limb deformity. In particular he spoke about bow legs and knock knees and how they respond to correction by limb alignment with the corrective osteotomies (breaking and setting of the bones). He did say that the rates of recurrence of deformity during the growth periods are unfortunately high.
- Damian Clarke (Australia) spoke about Managing Carpal Tunnel and Trigger fingers outlining that early diagnosis is important to achieve good outcomes.

Breakout sessions:

After a very full and intense day the family session broke down into 3 workshops. Each workshop had three presentations from families and professionals and set the scene for discussion. These sessions were very informative and gave parents the opportunity to ask questions and gain much needed information.

Although our day closed at 5.30 the breakout sessions continued until well after 6pm. It was wonderful to see the professionals who were chairing these meetings give of their time and ensure that the families were able to ask their questions and share their experiences of travelling the MPS road.

Friday commenced with New Therapies and the management of Mucopolysaccharidoses which covered all those difficult areas of Dental Care, Heart Disease, Anaesthetics, and Ophthalmic complications. The afternoon saw us looking at the Challenges of the Central Nervous system and Therapies: something old something new.

Saturday looked at Patient surveys, Registries, Natural History Studies and Home infusions, Getting the right clinical care and transition from Paediatrics to Adult Services. All these wonderful presentations will very shortly be available for you to watch on-line at <http://mps2010.eproceedings.com.au/index.html>

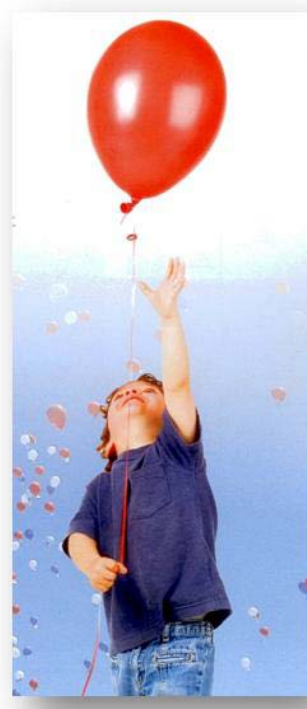
Memorial to our Special Children:

On Saturday afternoon we gathered on the banks of the Torrens River to remember the children who have passed away. The service was lead by Kimberlee Francis who is the parent of an MPS I daughter. We couldn't have asked for a better day the sun was shining and there was not a breath of wind or a cloud in the sky, but just as we were to release the balloons a small breeze started and as the balloons were released they floated gently into the sky. All our children who have gone before us were there watching over us.

The following poem was read during the service:

**Where I have gone, I am not so small.
My soul is as wide, as the world is tall.
I have gone to answer the call, the call
Of the one who takes care of us all.**

**Wherever you look, you will find me there –
In the heart of a rose, in the heart of a prayer.
On butterflies' wings, on wings of my own.
To you I'm gone, but I'm never along –
I'm over the moon, I am home.**

Gala Dinner and Life Time Awards:

The Gala Dinner was a spectacular grand finale of 2 ½ years of hard work done by LDNZ, MPS Society Australia and LDA. We combined both the Children's gala dinner and the Adults Dinner together and all were welcomed back to the convention centre one last time with our fire and Ice themed evening of entertainment, good food and good company.

There had been a lot of excitement building throughout the day. On arrival at the convention centre for the Saturday sessions delegates saw a white tunnel leading into hall H. There certainly was a lot of speculation as to what might be happening that evening. Wendy Boon and I had great fun telling everyone to wait and see.



**Oliver Lodewyke at the entrance to the
Gala dinner**



**Flame thrower: welcoming everyone to
the convention centre for the last time.**

Guests walking through the tunnel that lead them into the themed room



The evening was full of laughter, entertainment and dancing, but there was a serious side to the evening as well, as we presented Life Time awards to three very special people who have spent their lives researching, and caring for families who are affected by MPS and related diseases.



Dr Sly received a Life time award for his work in Research for Lysosomal Diseases

In 1973, he was the first to recognize mucopolysaccharide disease Type VII (now called Sly Syndrome) as an inherited metabolic disease; ground breaking research in medical genetics, cell biology and lysosomal storage diseases has led to new approaches to diagnosis of inherited diseases and possibilities of gene therapy.

Dr. Sly is recognized internationally for his work on inherited human diseases. Best known for research on lysosomal storage diseases, he is also known for biochemical genetic studies that revealed the molecular bases of human carbonic anhydrase deficiencies and of the inherited disease that causes the accumulation of a chemical misidentified as ethylene glycol. Most recently, Dr. Sly's pioneering research at St. Louis University School of Medicine in gene therapy has led to development of a genetic screening test for hereditary hemochromatosis, making diagnosis possible before symptoms appear. He received the 1999 Coriell Medal, given every other year, from the Coriell Institute for Medical Research.



Dr Ed Wraith received his life time award for his work in the clinic caring for all those affected by a Lysosomal Disease.

Consultant Pediatricians with a special interest in inherited metabolic disease. Principal Investigator for a number of pivotal phase III studies for Lysosomal Diseases.

Ed also has played a vital role here in New Zealand. He has attended our Lysosomal conferences and contributed valuable information to the International Bone Consensus meeting held prior to the 2008 Lysosomal meeting in Christchurch. Ed vast knowledge has helped us all better understand what is happening to our loved ones.

LDNZ is very proud to see such a prestigious award be presented to Ed for his life time work with these very rare diseases. Congratulations Ed it is well deserved.



Ros Smith received her award for her life time involvement with the Australian MPS society

Ros's daughter Adrienne, was born in 1974, and was MPS III affected.

In 1983, Ros was pivotal in creating the Australian MPS Society, and was the inaugural President. She held this position through to 1989, during which time the Society organised their first three family conferences. Ros also attended the UK Society conference in 1985, which cemented a bond between the Societies which remains strong today.

In 1992 Ros, and then President Denise Law, travelled to Austria to bid and secure the chance to hold the 4th International Symposium in Wollongong, Australia in 1996. This was a huge event for Australia, and did much to develop the organisation of the Australian MPS Society, and their relationships with Clinicians and Professionals in Australia. In 1997, in recognition of her work, Ros was made a Life Member of the Australian MPS Society. After 18 years service, in 2001 Ros finally retired from all duties on the Committee of Management and Board of Directors of our Society.

In parallel with her work for the MPS Society, Ros had been exploring other areas and needs in the Community.

Local to Sydney, Ros has long been involved with the Children's Hospital at Westmead, and worked closely with Professor David Sillence and others. This resulted in the establishment of the ConnecTed group, who provide assistance to all families being treated through the Connective Tissue Dysplasia Clinic at Westmead. Parallel with this at Westmead, Ros was also associated with the Innovative Therapies Group, which brought together the major support groups dealing with Westmead, such as Fabry, Pompe and Gaucher support groups. Working with the NSW State Department of Health, Ros continues to serve on the NSW Birth Defects Advisory Committee.

LDNZ Congratulates Ros on receiving an MPS Life time award acknowledging her dedication and life time commitment that has seen many improvements in service delivery and quality of life for those affected by these rare diseases.



It has been a real privilege to be involved in the planning, organising and hosting of the 2010 International MPS symposium and I want to take a moment to thank the Trustees of LDNZ for their support during the last 21/2 years. It has been a truly amazing experience, right from planning the meeting, raising the funds, and then finally hosting the meeting, and I know that families in our part of the world have benefited from having access to a meeting of this calibre. I also want to make special mention of Wendy Boon. We began this journey together with a 30 hour flight to Brussels to put in our bid for the meeting and through this journey we have become very good friends. I also want to make mention of our husbands. They have supported us both through this journey and were the gofers during the symposium. They took the children's food out to the venues each day, they went out for Velcro dots and other small items that we had forgotten to bring with us, and they made sure that Wendy and I ate and delivered to us a glass of wine or two at the end of each day. Thank Paul and Eric.

Jenny Noble
Secretary
Lysosomal Diseases New Zealand



Please help the work of Lysosomal Diseases New Zealand

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

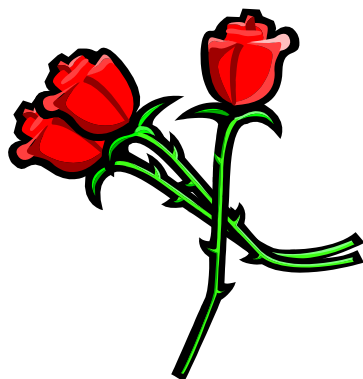
Funds raised by LDNZ cover the following areas

Funding of all administration expenses for our group.

- ✿ Supporting families wishing to attend Conferences.
- ✿ 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.
- ✿ Advocating for families for disability support, health services and access to therapies.

We gratefully accept donations that will enable us to continue toward our goal of a future free of the tragic consequences of Lysosomal Storage Diseases.

Donations can be sent to **Lysosomal Diseases New Zealand**
16 Woodleigh Place, Ohauti, Tauranga 3112



LDNZ Trustees

Chairperson: John Forman		Secretary: Jenny Noble
E-mail: john.forman@xtra.co.nz		Email: jenny.noble@xtra.co.nz
Phone 04 566-7707		Phone 07 544-8886

Trustees:

Dr Dianne Webster - Auckland
Prof. David Palmer - Christchurch
Philip McKinstry - Auckland