



Lysosomal Grape Vine

Newsletter

May 2011



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

- Editorial and another call to action.
- LDNZ Annual Report.
- Behind the Scenes.
- What's in the news – latest research.
- Reports from Patient meetings.
- International Batten conference announcement.

LDNZ's Charities Commission Registration
CC24962

Sadly we mourn the loss of

Tasman McKinnon – Feb 2011

Billie McKinnon – May 2011

**Who lost their battle with
Metachromatic Leukodystrophy**



When you feel Lonely

When a person you love passes away
Look to the night sky on a clear day.
The star that to you, appears to be bright,
Will be your loved one,
Looking upon you during the night.
The lights of heaven are what shows through
As your loved one watches all that you do.
When you feel lonely for the one that you love,
Look to the Heavens in the night sky above.

Author unknown



Hello to our Lysosomal network – **and another call to action!!**

In the September 2010 issue of our newsletter we put out a call to action to all Lysosomal families asking you to meet with your local National MP to highlight the issues we have with access to enzyme replacement therapies for our diseases. We believed the time was right for this, given that the Minister was due to announce in December 2010 how he intended to fund highly specialised medicines. Thanks to all of you who responded to our call and arranged meetings.

Unfortunately the Minister didn't make the expected announcement. Instead he relied on Pharmac's review of the Exceptional Circumstances Scheme as the pathway to a solution. LDNZ joined with NZORD and the Access to Medicines coalition to carefully analyse the consultation documents and we concluded that despite the Minister's indications, Pharmac were in fact proposing a tougher regime for exceptional circumstances. We submitted on important changes that would need to be made to the proposed exceptional circumstances scheme, if patients needing medicines for "orphan" diseases were to have a reasonable chance of getting them.

In parallel with this work, in December 2010 two applications were submitted to Pharmac for exceptional circumstances review; one for continued funding of ERT for Jack who has Hunter disease and the other for one of our adults with Pompe disease. The Pompe case was clearly also a test case for the other three affected adults. **However in March the Pompe application was declined and Jack's application disappeared somewhere in the system and was not decided upon.**

It is fair to say we felt completely gutted by the decision regarding ERT for Pompe disease. It is funded in 43 of 45 comparable countries, including some Indian states, but Pharmac declined funding, even after noting there would be an expected modest improvement followed by stabilisation of this degenerative disease. It is particularly concerning that Pharmac addressed cost issues thoroughly in the advice to the Board, but did not once mention the other important issues of equity and community values that are contained in the Medicine Strategy. They noted there was no other treatment for this life threatening condition, yet still declined funding.

We are also extremely concerned at what seems like "game playing" by Pharmac in relation to Jack's application. The company supplying the ERT was giving free supply towards the end of the second year of his treatment while negotiations were going on. Pharmac simply chose not to deal with the application so they could get more free medicine supplied. LDNZ

considers this a serious breach of the integrity expected of public sector agencies, especially in light of the enormous distress their game has imposed on Jack's family.

Six years of working through the system, engaging with Ministers, officials, and submitting on consultations has got us practically nowhere with access to these highly specialised medicines. Recent meetings with Pharmac staff clearly indicate they are very unlikely to grant approval to these treatments in the foreseeable future. The one gain we got when Jack's treatment was approved in 2009 is now in jeopardy. Ministers have refused to meet with us to discuss the issue. We are backed into a corner with little option but to make this a high-profile media and political issue.

We are now building a campaign to seek a solution to the problem. However Pharmac will not be our main target, despite many difficulties we have with their approach to these medicines. **Our campaign will be focussed on the need for government to put in place a policy framework that gives an equitable opportunity for patients requiring highly specialised medicines like our ERTs, to get funding allocated.** A similar system applies in Australia, and some other countries have access policies regarding treatments for "orphan" diseases. Such a policy framework would give different criteria for Pharmac to consider and improve our chances of success.

Our campaign has already commenced its preliminary phases with legal advice being sought, meetings and regular briefings of affected patients, and discussions with a number of experienced politicians and media experts. We have gained the support of the Boards of LDNZ, NZORD and the Muscular Dystrophy Association. (Pompe disease is also as form of muscular dystrophy as well as a lysosomal disease).

Keep an eye out for more news on this issue and expect a request from us for you to get involved in our campaign. This is scheduled to commence in the first week of June.

Warm regards at the start of a chilly winter,

John Forman
Chairperson
Lysosomal Diseases New Zealand



Lysosomal Diseases New Zealand – Annual Report

This report covers the financial year July 2009 – June 2010, and includes some commentary up to the date of our meeting in February 2011.

Overview: 2010 was again a difficult year for fundraising. We have found LDNZ is having difficulty fitting into the new criteria set out by several of our traditional funders and we have lost several grants usually obtained in past years. However a surplus from the Adelaide conference did provide us with a good return and this helped save the situation for us.

Arrangements for the International MPS and related diseases symposium which was held in Adelaide were a major task during 2010. I want to commend the efforts of Jenny Noble who put in a very substantial effort to ensure the success of this meeting, and Wendy Boon of the Australian MPS Society who contributed significantly.

Throughout 2010 we continued to advocate for Access to Medicines for our diseases. This is proving to be quite a challenge for us, but in December 2010 two applications were submitted through Exceptional Circumstances for Pompe and Hunter syndrome. We had high hopes that we would see some progress through these two applications. At the time of our trustees annual meeting we were waiting anxiously for a response from Pharmac.

Finances: Raising funds via charitable grants has become very difficult for LDNZ. We have normally received grants from Lottery Grants board, J R McKenzie Trust, Todd Foundation, Pub Charity and other such groups. However most of these funders have less money to spend and some have changed their funding criteria, meaning we are receiving considerably less from these traditional sources. Without the wonderful fundraisers that we have held in the past we would be in a very difficult situation.

Income for the financial year ended 30th June 2010 was \$99,803 with expenditure of \$124,008 giving us a deficit of \$24,205. However we were able to maintain financial viability thanks to the Charity dinners and Charity golf functions that have been held over the last few years as this has left us with funds in the bank of \$89,658 at the end of June 2010. The surplus from the Adelaide conference came in after the end of the financial year and will help maintain our activities into the future, however even with those additional funds, the Trust has identified the need for fundraising a minimum of \$40,000 to keep us going for the next two years.

If you would like to have a closer look at our accounts please ask Jenny to send you a copy, or you can go to the Charities Commission website www.charities.govt.nz, search the register using the word Lysosomal, and this will bring up links to all recorded information about LDNZ, including links to audited accounts.

New Families: Over the last 12 month a total of 10 new families have been identified and although we would prefer to not to see new families diagnosed with these diseases we do welcome all them all to our Lysosomal community. The disease groups for new families are: Batten 2, Fabry 2, Gaucher 1, MLD 1, Pompe 1, Tay Sachs 1, Krabbe 1, Sanfilippo 1

Publicity Materials: We spent the last year giving LDNZ a fresh new look and would like to acknowledge the grant given to us by Genzyme to help make this all happen. We now have a beautiful new A4 flyer with new information that we hope will be very helpful for new families. We have taken this flyer to several professional meetings where many copies have been snapped up by those present.

10 year report: During the last 3 months of 2010 we started work on our 10 year report. It has been like a trip down memory lane, looking back at all the things we have done as an organisation over 10 years. We had hoped to have this project completed prior to Christmas 2010, but we have been waiting for government decisions on the funding of specialised medicines and the review on Exceptional Circumstances which is being done by Pharmac. With the delay in both of these areas we have put the report on hold, as we are keen to include detail of progress on this issue in the report. It is, after all, the most pressing issue we face. We are hopeful that we will have this completed before Christmas 2011.

Advocacy within the health system: We have continued to keep the pressure on both Pharmac and the Minister of Health, in regards to the funding of Enzyme Replacement Therapies. The Minister did not make his expected decision on the funding of specialised medicines at the end of 2010, instead he asked Pharmac to review the EC application process as a possible way of improving access to these medicines.

We are very concerned about the consultation document put out by Pharmac and believe in its current form we will see great difficulty getting applications for ERT accepted. We have made a formal submission to Pharmac and joined with a number of other groups in making representations to Pharmac and to government about this issue. We expect this issue will take up a lot of our time in the coming year.

Our work with the National Health Board has progressed well and late in 2010 they confirmed that Genetic services and Metabolic services will be national services that are centrally planned and funded. Work is continuing to get these new services set up by 1 July 2011. LDNZ expects this will result over time in better clinical care services for families, faster diagnosis, and thus better outcomes for all of us. There will still be a need for advocacy for our families as we do not expect all the problems regarding referrals from District Health Boards, transition to adult services, and so on, to be resolved overnight. But the new services will have specific responsibilities that have not been clear in the past. We are optimistic that improvements will occur.

Natural History Study for Glycoprotein Storage Diseases. The Scholarship won by Jenny Noble from AMP in 2008 was used during 2009/10 to include 15 patients from New Zealand and Australia into a Natural History Study for Glycoprotein Storage diseases. This was an exciting opportunity for our families to be involved in this International Study and another good excuse to bring this group of families together again. We continue to support this project and will be working with Dr Cathey in 2012 as she extends the study to include the 9 diseases that ISMRD (The International for Glycoprotein Storage Diseases) supports.

Sanfilippo Research: In Jan 2010 Jenny and John were invited to Massey University to meet with Bob Jolly to see some of the results from his research project that LDNZ supported during 2009. Bob has been looking at how they can get Enzyme replacement therapy into the brain via intrathecal injection into the cerebral-spinal fluid, using the Huntaway dogs that naturally have this disease. He was able to show that they can get some enzyme into the brain, and was able to show that positioning of the dog during treatment was an important factor in the flow of enzyme into brain cavities. Bob presented his work in Adelaide at the International MPS family meeting. We will continue to follow his work as he moves onto considering Gene Therapy for Sanfilippo disease.

Trust Accountability: The trustees met in February 2011 for their Annual Meeting and to plan activities for the year ahead. Activities based on that plan have been carried out mainly by Jenny with regular reporting to the chair of the Trust, with additional issues being referred to the trustees by e-mail for decisions. This continues the pattern from past years where we meet face-to-face once a year for a full day of review and planning, and deal with all matters in between times by phone and email.

Keeping our books in order is very ably supported by Tim Hannagan through his accounting firm and this help is much appreciated, especially as the volume and complexity of transactions has grown in recent years.

My grateful thanks to trustees Dianne Webster, Dave Palmer, and Philip McKinstry for their support throughout this time, and in particular to Jenny for the significant commitment she makes to ensure the success of LDNZ's day-to-day operations.

**John Forman
Chairperson
LDNZ**



BEHIND THE SCENES

Jenny Noble
Field Officer | Administrator

What a terrible start we have had to 2011 with the Christchurch earthquake. It was devastating seeing the damage on the TV, but even more devastating for our Lysosomal families, who live in this region. It took us several days to touch base with everyone and thankfully you are all safe. However two families have had significant damage done to their homes. There were significant issues around medical access and disability support for the families as they continued to try and keep life as normal as possible. I am very pleased that from Tauranga I was able to sort out many of the issues and give much needed support during this very difficult time.

I want to take a moment though to thank the Hannagan family for offering their holiday home in Wanaka for families to have some time out. The Denovan family took up this offer and had two weeks free from earthquakes. **Thank you, Marianne and Tim.**

It is the behind the scenes work that I want to talk about here. You can see from my title above the board of trustees have re-defined my roles for LDNZ as much of the work I do is supporting families, whether it is helping you work through the mine field of access to Health and Disability services, or helping you to find up to date information on your particular disorder, connecting you to other families both here in New Zealand or overseas, or just a listening ear at the end of the phone.

So what is it that LDNZ does behind the Scene?

- Helps you to negotiate appropriate allocations for Carer support and Personal Care hours.
- Attend Needs Assessment meetings with families when necessary.
- Supports families to meetings on their particular disorder when funds allow us to do so.
- Provides information and support to New Families.
- Organises Family Education meetings in New Zealand.
- Advocates for Access to Enzyme Replacement Therapy.
- Works with the Ministry of Health on improved clinical services for our diseases.
- Attends meetings around New Zealand and hosts display booths on our diseases.
- Helps with submissions such as the current review for Exceptional Circumstances.
- Helped to develop Rare Disease day for New Zealand and encourages involvement by LDNZ families.

Then we add to this list all the day to day administration that takes place, such as our newsletters, management of the annual accounts, grant submissions, website management, design and production of all our publicity materials etc. This is just a wee snippet of what gets done behind the scenes. This role for LDNZ is a part-time job which ends up being full time.

I want to acknowledge the work that Tim Hannagan does in getting our accounts compiled at the end of each financial year. He does an amazing job given that we are at different ends of the country and I know this can be very trying given the complex nature of the accounts when fundraisers take place. Thank you Tim for all that you do to help support this part of the Administration work for LDNZ.

Both John and I are always available if you ever need to talk with us other wise I am making every effort to go through our database and touch base with you all this year.



WHATS IN THE NEWS

B:OMARIN®

BioMarin Initiates Pivotal Phase 3 Trials for GALNS for the Treatment of MPS IVA (Morquio)

Novato, Calif., February 1, 2011 – BioMarin Pharmaceutical Inc. (Nasdaq: BMRN) announced today that it has initiated a pivotal Phase 3 trial for N-acetylgalactosamine 6-sulfatase (GALNS or BMN 110), intended for the treatment of the lysosomal storage disorder Mucopolysaccharidosis Type IVA (MPS IVA), also called Morquio A Syndrome.

“In under two years, we have progressed the GALNS program from Clinical Trial Application to initiation of the Phase 3 trial. We have received FDA feedback and have finalized the design of the Phase 3 pivotal trial,” said Jean-Jacques Bienaimé, Chief Executive Officer of BioMarin. “The study will be conducted at approximately 40 centers worldwide including Brazil, Japan, Taiwan, most Western European countries, Canada and the U.S. The trial is expected to enroll approximately 160 subjects and will be the largest enzyme replacement therapy trial conducted. There are no therapeutic options for MPS IVA patients who have a high unmet medical need. Initiation of this well-designed pivotal study is an important milestone for both the company and the MPS IVA community.”

The Phase 3 trial is a randomized, double-blind, placebo-controlled study to evaluate the efficacy and safety of GALNS in patients with MPS IVA. The study will explore doses of two mg/kg/week and two mg/kg/every other week for a treatment period of 24 weeks. The primary endpoint is the six-minute walk test, and the secondary endpoints are the three-minute stair climb test and urine keratan sulfate concentration.

Read more at: <http://www.bmrn.com/pipeline/morquio-syndrome.php>



BMN-701: IGF2-GAA for Pompe Disease

B:OMARIN®

Pompe disease, a lysosomal storage disorder, is a progressive degenerative disease of the heart muscle, diaphragm and skeletal muscle. It is caused by a deficiency in the lysosomal enzyme acid alpha glucosidase which leads to the accumulation of glycogen in myocyte lysosomes and results in cell death. The incidence is one in 40,000 births. There are two main forms of Pompe disease: adult onset with an incidence of one in 57,000 births and infantile onset with an incidence of one in 138,000 births.

The key attribute of BMN-701 is its ability to bind to key cell receptors that direct the enzyme to the cell's lysosome. Instead of trying to engineer cells to make GAA with higher levels of mannose-6 phosphate, which in the case of GAA lowers cell productivity and significantly increases manufacturing costs, BMN-701 takes advantage of the fact that the peptide IGF-2 also binds to the M6P receptor. Every molecule of BMN-701, a fusion of IGF-2 and GAA, can bind the mannose-6 phosphate receptor, be taken up into cells and trafficked to the lysosome where it can degrade the glycogen that causes Pompe disease.

Editor's note: LDNZ understands that BioMarin is close commencing a clinical trial for this therapy.

Read more at: <http://www.bmrn.com/pipeline/igf2-gaa-for-pompe-disease.php>

Oregon Health & Science University (OHSU) begins a Phase Ib clinical trial sponsored by StemCells, Inc. for Infantile or Late Infantile Neuronal Ceroid Lipofuscinosis (NCL).

Phase Ib Trial in NCL – Initiated October 2010

Based on the favorable safety data from our first NCL trial, we have advanced to a second clinical trial to further assess the safety and preliminary efficacy of HuCNS-SC cells as a potential treatment for NCL. The Phase Ib trial is designed to enroll patients with less advanced stages of the disease than those enrolled in our first NCL trial, which we believe will enhance the prospect of detecting clinical benefit. Like our first NCL trial, this second trial is being conducted at OHSU Doernbecher Children's Hospital, a leading medical center with nationally recognized programs in pediatric neurology and neurosurgery.

In this trial, we expect to enroll six patients with either infantile or late infantile NCL. All patients will be transplanted with HuCNS-SC cells via a neurosurgical procedure, and will be immunosuppressed for nine months. Following transplantation, the patients will be evaluated regularly over a 12-month period in order to monitor and evaluate the safety and tolerability of the HuCNS-SC cells, the surgery, and the immunosuppression. In addition, MRI evaluations will focus on certain measurements to evaluate the impact of the HuCNS-SC cells on disease progression. These measurements will include assessing and tracking cerebral volume and unique neuronal metabolites, both of which are negatively impacted by disease progression. As we intend to follow the effects of this therapy long-term, a separate four-year observational study will be initiated at the conclusion of this trial.

Phase I Trial – Completed January 2009

Our Phase I trial in NCL, initiated in 2006, was the first ever FDA-authorized clinical trial of human neural stem cells. This landmark trial primarily focused on assessing the safety of our HuCNS-SC product candidate as a potential treatment for NCL. Data from this study, reported in June 2009, demonstrated the clinical safety and tolerability of the cells, and included evidence of engraftment and long-term survival of the donor cells. We continue to follow patients who completed this trial, some of whom are now more than two and three years' post-transplant



Zymenex enzyme in Phase 2 clinical trials in patients with the rare disease alpha-Mannosidosis

Phase 1 trial has just demonstrated that the enzyme is safe and well tolerated and the Phase 2 dose-finding clinical trial is now underway. The biotechnologically derived human enzyme product rhLAMAN (Lamazym™), which is produced by the Scandinavian biotech company Zymenex and developed for the treatment of patients suffering from the rare disease alpha-Mannosidosis, has successfully completed Phase 1 trials and has now entered Phase 2a clinical trials. The patients were recruited into the Phase 1 trial from around Europe, where the initial goal of demonstrating that the enzyme is safe and well tolerated, has been achieved. This now allows the 10 patients to be moved forward into a 6-month Phase 2a dose-finding clinical trial, where the aim is to identify the most optimal dose to achieve the desired clinical effect.

Read more at: <http://www.zymenex.com/PressReleases>

Three-Year Data from Phase 2 Trial of Genzyme Gaucher Disease Oral Compound Suggest Sustained or Further Improvement across all Endpoints



CAMBRIDGE, Mass. Feb 18 2011 Genzyme Corporation (NASDAQ: GENZ) today announced three-year follow-up data from patients enrolled in the phase 2 clinical trial for its investigational oral therapy for Gaucher disease type 1 known as eliglustat tartrate. Sustained or further improvements were observed across all endpoints, including bone disease, at the three-year time point. The results were presented for the first time this week at the Lysosomal Disease Network WORLD Symposium in Las Vegas, Nevada.

Genzyme previously reported that the eliglustat tartrate phase 2 trial had met its primary endpoint at one year, and that data demonstrated continued improvement through two years. The primary composite endpoint was a clinically meaningful response in at least two of three endpoints: improvements in spleen size, hemoglobin and platelet levels. The study has continued with 19 patients through three years. The extension phase of this trial is still ongoing.

Eliglustat tartrate continued to show robust clinical response through three years:

- Spleen volume decreased from baseline by a mean of 61 percent and liver volume decreased from baseline by 29 percent.
- Hemoglobin level increased from baseline by a mean of 2.6 grams per deciliter.
- Platelet count increased from baseline by a mean of 91 percent.

The study also analyzed the clinical response of patients in the phase 2 trial with respect to achieving therapeutic goals. Due to the heterogeneity of Gaucher disease, therapeutic goals were previously developed by experts involved in the treatment of Gaucher patients to assess their response to enzyme replacement therapy (ERT). Most patients dosed with eliglustat tartrate met established therapeutic goals for hemoglobin, platelets, spleen volume and liver volume, demonstrating progressive and clinically meaningful responses in multiple organ systems. At three years, 100 percent of patients met at least 3 of the 4 therapeutic goals developed for hematologic and organ volume parameters.

The three-year data also included analyses that suggest eliglustat tartrate positively impacts indicators of bone disease through three years of follow up. These indicators include bone mineral density in the lumbar spine, as measured by dual energy x-ray absorptiometry (DXA), and dark marrow signal in the femur, as visualized by magnetic resonance imaging (MRI). Dark marrow reflects the infiltration of lipid-laden Gaucher cells into bone marrow. Specifically:

- In the 18 patients at baseline with dark marrow in the femur visible by MRI, five improved by one year, seven by two years and 10 by three years, with the other eight patients remaining stable.
- In the 15 patients with results available at all time points, bone mineral density in the lumbar spine showed clinically and statistically significant improvements after one year of treatment (T score = +0.4) which further improved after 2 years (T score = +0.6) and were sustained after three years of treatment.

The reviewed data suggest that eliglustat tartrate may have a meaningful clinical impact on bone disease in Gaucher disease type 1 patients."

Read more at:

http://www.businesswire.com/portal/site/genzyme/index.jsp?ndmViewId=news_view&ndmConfigId=1019673&newsId=20110218005932&newsLang=en

Raptor Pharmaceutical Completes Enrollment in Pivotal Phase 3 Clinical Trial of DR Cysteamine for the Potential Treatment of Nephropathic Cystinosis



Raptor Pharmaceutical Corp. has announced it has completed enrollment in its Phase 3 clinical trial of its proprietary delayed-release oral formulation of cysteamine bitartrate ("DR Cysteamine") in patients with nephropathic cystinosis

The pivotal Phase 3 clinical trial is designed as an outpatient study of the safety, tolerability, pharmacokinetics ("PK") and pharmacodynamics ("PD") of DR Cysteamine dosed every twelve hours in patients with cystinosis, compared to the current standard of care, immediate-release cysteamine bitartrate, which requires dosing every six hours. Raptor expects over 30 patients to complete the eight-week study protocol. All patients completing the Phase 3 clinical trial have the option of enrolling in a long-term follow-on study where they continue to receive DR Cysteamine twice daily for the extent of the study.

Read more at:

<http://ir.raptorpharma.com/phoenix.zhtml?c=198466&p=iroNewsArticle&ID=1518876&highlight>



Amicus Therapeutics Provides Positive Data Update from Phase 2 Long-Term Extension Study of Amigal™ for Fabry Disease

Company also announces encouraging new preclinical data from Chaperone-ERT Co-Administration and Neurodegenerative Disease Studies

CRANBURY, N.J., Feb. 16, 2011 /PRNewswire/ -- Amicus Therapeutics (Nasdaq: FOLD) today announced that additional positive data from the ongoing Phase 2 extension study of its investigational drug Amigal™ (migalastat HCl) for Fabry disease will be presented at the Lysosomal Disease Network WORLD Symposium in Las Vegas, Nevada, February 16-18th, 2011. In addition, the Company announced that it will present encouraging data from its preclinical studies evaluating the co-administration of pharmacological chaperones with enzyme replacement therapy (ERT) in Fabry disease, as well as from preclinical studies examining the use of pharmacological chaperones for the treatment of genetically defined subpopulations of Parkinson's disease and Alzheimer's disease.

Preliminary Data Update from Phase 2 Long-Term Extension Study of Migalastat HCl for Fabry Disease

Twenty-six subjects completed either 12 or 24 weeks of treatment with migalastat HCl during the initial Phase 2 studies and 23 subjects enrolled in a separate, long-term extension study designed to evaluate the long-term safety and efficacy of migalastat HCl. Over the course of the initial Phase 2 and extension studies, 15 subjects have been treated with migalastat HCl for more than 3 years and 7 subjects have been treated with migalastat HCl for more than 4 years. Seventeen subjects continue to receive treatment in the ongoing extension study.

During the course of the extension study, treatment with migalastat HCl has continued to be generally well tolerated, with no drug-related serious adverse events. The most common adverse events have been headache, arthralgia, diarrhea and fatigue.

Renal function continues to be evaluated by two measures in the extension study, estimated glomerular filtration rate (eGFR) and 24-hour urine protein. Preliminary data indicate that eGFR has remained stable out to 3-4 years for all subjects continuing in the extension study and the average annual rate of change in eGFR in subjects identified as responders to migalastat HCl, excluding hyperfiltrators, was +1.6 mL/min/1.73m². Additionally, reduced 24-hour urine protein continues to be observed in multiple subjects identified as responders to migalastat HCl, with a mean 21% and median 34% reduction from baseline in this group of subjects.

Co-administration with ERT in Fabry Disease and Pompe Disease

Amicus previously reported promising preclinical data demonstrating that the co-administration of a pharmacological chaperone with ERT has the potential to address key limitations of ERT. The addition of a pharmacological chaperone has been shown to prevent the loss of activity of ERT in the circulation, increase tissue uptake, and increase substrate reduction in multiple disease-relevant tissues. Preclinical proof of concept has been established for Fabry disease and Pompe disease.

The Company will present a review of new and historical data from preclinical studies of migalastat HCl co-administered with ERT in an animal model of Fabry disease. Amicus and its partner GlaxoSmithKline PLC (GSK) are sponsoring an ongoing Phase 2 study evaluating the co-administration of migalastat HCl with ERT for Fabry disease. Results from this study are expected in the second half of 2011.

In addition, Amicus will present pharmacokinetics and muscle distribution data from a Phase 1 study of AT2220 that support the planned Phase 2 clinical study of AT2220 co-administered with ERT in Pompe patients. The Company expects to initiate this study in the first half of 2011 and to report preliminary results in the second half of 2011. The Company intends to seek U.S. FDA approval to lift the current hold on the AT2220 program as part of its development plan.

About Amigal

On October 29, 2010, Amicus announced a definitive agreement with GlaxoSmithKline PLC (GSK) to develop and commercialize Amigal (migalastat HCl), currently in Phase 3 for the treatment of Fabry disease as a monotherapy. Under the terms of the agreement, GSK received an exclusive worldwide license to develop, manufacture and commercialize migalastat HCl. GSK and Amicus are also investigating Amigal as a treatment for Fabry disease when co-administered with ERT and have commenced a Phase 2 study as noted above.

The Phase 3 study (Study 011) of migalastat HCl is ongoing and patients are being enrolled at 36 investigational sites worldwide. A majority of the planned 60 subjects have been enrolled in the study. The Company expects to complete enrollment in the first half of 2011 and to report top line results from this study in the second half of 2011.

Amicus and GSK intend to commence an additional Phase 3 study (Study 012) in the first quarter of 2011. Study 012 will be an 18-month, randomized, open-label study comparing migalastat HCl to enzyme replacement therapy (ERT) in approximately 60 subjects. The primary outcome of efficacy will be renal function as measured by glomerular filtration rate (GFR).

To read more go to: <http://ir.amicustherapeutics.com/releasedetail.cfm?ReleaseID=550504>

Reports from Batten Conference Australia and Metachromatic Leukodystrophy meeting New Zealand

5th Battens Conference Seaworld Nara Resort Queensland, October 2010 By Lynda and Torben Dougan

Well what a wet weekend! As per normal the conference was extraordinary. A lot of useful information was obtained. Particularly the manual handling speaker. She was extremely helpful following the seminar going over several techniques on a one on one basis. The researchers and scientists are amazingly dedicated to our cause.

It is pleasing to see the clinical trials taking place for the Infantile and Late infantile strains. We Juveniles live in hope!

It was fantastic to catch up with old friends also. To see the change in the children, to laugh and share stories.

It is very sad to think this was probably the last conference in this form. I feel that perhaps the network opportunities for Juvenile families will be lost as we are so few to the other strains, when the conference goes regional. I feel it also highlighted the need for us Kiwi's to band together. Our facilities and services are very different to those in Australia. We must pool our knowledge and support. Perhaps our own identity will help us do this. This was spoken about briefly and I believe we need to research the pros and cons of this very carefully so as not to reinvent the wheel.

Jon Cooper of Kings College London told us all of a fabulous opportunity to go to London 2010, To join with many more specialists and families. I know we are all very excited at the chance of going. We wish to work together to make it happen.

Torben and I had a great time despite a few hiccups along the way. We must say Air New Zealand made the experience easy for us. This was the first time I had travelled with Torben immobile on my own.

A very, very large thank you to LDNZ, for making this trip possible. I know how hard you all work to make this possible. Also Battens Au for paying our conference registration fees.

Again Thank you

There are two other reports submitted by Ra Timms and Sharon Noble on our LDNZ website, to read more about this wonderful conference go to our website address below.

http://www.ldnz.org.nz/newsletters/conference_reports/5th_batten_conference_2010_australia

Metachromatic Leukodystrophy family meeting Wellington

On 26TH of February we were able to attend the MLD foundation family conference here in wellington. It was wonderful to finally meet Dean, Teryn (and Lindy) who run the MLD foundation which has been very supportive of all MLD families here in NZ and around the world.

Dean and Teryn gave us a presentation covering many of the different aspects about MLD, from diagnosis, future treatments and pain management. John Forman was also able to talk about some of the issues that we here in New Zealanders with the limitations of our health system. Afterwards we were able to have a nice meal together spend some time socializing. It was great meeting everyone and we thoroughly enjoyed our day.

I'd like to thank Dean & Teryn for hosting the conference and acknowledge that without the support of LDNZ it would not have been possible and to personally thank Jenny & John for everything they do for Lysosomal families here.

Rob and Mandy Wilson



On February 26th nearly thirty five people from all over the country gathered in Wellington to share, compare, and learn about MLD at New Zealand's first ever focused gathering of families affected with MLD. Many parents brought their affected children and loved ones while some who had previously lost loved ones brought memories and their experiences to share. Dean and Teryn Suhr from the MLD Foundation led discussions about the disease, research, clinical trials, and practical care. Dr. Esko Wiltshire from the University of Otago in Wellington and Dr. Julie Hauer from Boston (via teleconference) were the primary medical experts, while information from Dr. Callum Wilson (Auckland – Starship Hospital) from New Zealand's national metabolic center was also shared. We discussed the Gene Therapy clinical trial currently under way in Milano Italy and Shire HGT's re-initiation of an Enzyme Replacement Therapy with their HGT-1110 enzyme later in 2011.

We were very pleased by both the turnout of families from across the country as well as the open sharing of medical, financial, and social service concerns that occurred throughout the day. As a first ever focused gathering of MLD affected families, we were able to discuss the subtle aspects of caring for loved ones with MLD. After a busy and long day of meetings we all sat down for a casual dinner as we continued to get to know each other.

We encourage the families to stay connected, to reach out to other families with or to be diagnosed with MLD, and to utilize the resources of LDNZ for local support. You have a gem in both John Forman – he is internationally recognized and active on behalf of rare diseases, and Jenny Noble, your national LDNZ coordinator. We are always available to help and hope to return to your beautiful country again soon.

Dean Suhr, MLD Foundation, <<http://MLDfoundation.org>>

Thank you for your Support!

We wish to acknowledge all these wonderful people who donated funds to LDNZ since our last newsletter.

*Grantea Downs School, L McKinnon on behalf of Tasman, J Labedzki,
K. Du Frense, Grantea Primary School, Ballantynes Timaru,
Scenic Hotel Timaru, Lions Club Timaru, DB Brewery's*



13th International Conference on Neuronal Ceroid
Lipofuscinoses (Batten Disease)
and
1st Worldwide Meeting for Batten Parents Organisations
28th -30th March 2012 - London

More information will follow as the website gets updated please go to :

www.ncl2012.org

**NCL2012 Scientific Program
includes sessions on:**

Genetics & Biology of the NCLs
Disease Mechanisms
Links to Other Diseases
New Clinical Perspectives
Experimental Therapies

Please help the work of Lysosomal Diseases New Zealand

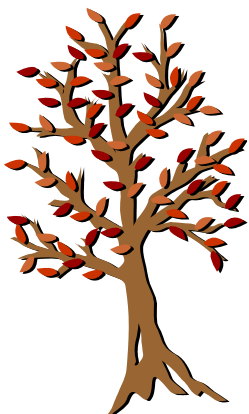
Donations over \$5.00 made to Lysosomal Diseases New Zealand are Tax deductible.

Funds raised by LDNZ cover the following areas

- ☞ Funding of all administration expenses for our group.
- ☞ Supporting families wishing to attend Conferences.
- ☞ Working with 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.
- ☞ Organising information and support to families.

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
E-mail: john.forman@xtra.co.nz
Phone 04 566-7707

Field Officer | Administrator: Jenny Noble
Email: jenny.noble@xtra.co.nz
Phone 07 544-8886

Trustees:

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