



December 2009

Hello to all our Lysosomal patients and their families. LDNZ has had a very busy and rewarding year and we are a little stunned that at the end of the year there's still so much to do. With the commitments on our time we have not managed to prepare a full newsletter, so here are some of the highlights for 2009 in this quickly prepared Christmas card insert.

Medicine funding and access

2009 commenced with the change in Government giving us the opportunity to push hard for therapies for our diseases. During January and February we set up meetings with all the political parties to highlight the need for specialised medicine for Lysosomal patients. We took 15 affected patients and family members to Wellington over a two week period where we had the opportunity to outline the issues, and those affected were able to tell their stories and ask for the much needed help in gaining access to the various ERT's. During this process Pharmac undertook its own review of the 6 ERT's that are now available. One small but very significant move on their part was to accept the recommendation of their expert committee that access to therapy would be on a case by case basis through the Exceptional Circumstances scheme that Pharmac operates. This was an important step as their previous approach had simply been to reject all applications.

In August Jack Peacock (Hunter syndrome), was granted access to Enzyme Replacement Therapy. We are thrilled to hear that Jack is doing really well. He has more energy and Kirsty reports that he doesn't seem to need his food supplements any more - they are coming home untouched. It is wonderful to hear the sparkle in Kirsty's voice when she talks about Jack. Getting to this end has been a very long and stressful process for the whole family. We look forward to following and reporting Jack's progress through 2010.

However this one success tends to dramatically highlight the situation of those who are still waiting for therapy, and we are very conscious of this. There is still much work to be done and we are keeping a close watch on developments and in close touch with the companies, the doctors and with Pharmac. Unfortunately there is a need for more data to be published about the effects of these treatments, and it is as frustrating for us waiting for this, as it must be for all the patients and their families. LDNZ continues to monitor the situation closely and will act as soon as any new information or opportunity arises.



Fundraising and Promotion

We held a very successful Charity Dinner in Tauranga in September which raised \$30,000. To all the families who attended and donated items for Auction thank you so much.

Along with hosting these dinners we do quite a lot of publicity and awareness for our group of diseases, which has paid off with a unique opportunity to promote our organisation. Jenny was approached during the charity dinner by Steve Bird from Bird wines. He was so taken with our organisation and those affected by these diseases. Just this month his interest led to advice that Cellar Select, Ohauti wines and Steve Bird have chosen to support LDNZ with a specially labelled wine depicting LDNZ and Cellar Select. For every dozen bottles sold LDNZ will receive \$50.00. We are very excited at this opportunity. We have Kirsty designing the label, but we still need a name, so here is your Christmas challenge. Email your suggested name to jenny.noble@xtra.co.nz or tell us your pick of:

1. **Covered Porch** – cause that's where you should drink a fine wine in the hot summer sun
2. **Farm Gate** – rustic and sentimental/traditional/Kiwi-ish
3. **Hautoa Range** – sounds good and means courage and the range means strength as in mountain range
4. **Hautoa** – means courage in Maori



International MPS and Lysosomal Biology meeting, Adelaide, June 2010

Planning for the 11th International MPS and Related diseases symposium is well underway. We are very proud to be one of three organisations hosting the symposium in Adelaide and look forward to being able to support some of our families to attend the meeting. From January 2010 we will be accepting indications from families wishing to attend. For more information on the symposium please visit www.mps2010.com.au

Natural History Study for Glycoprotein Storage Diseases



LDNZ, Greenwood Genetic Centre and ISMRD have joined in a partnership for research progress

The Journey began in 2005, when a family from Georgia traveled to Greenwood, South Carolina, so that their child affected with Mucopolysaccharidosis II could be evaluated by Dr. Jules Leroy. Dr. Leroy is a visiting Senior Scholar at the Greenwood Genetic Center and had reported the first patient with ML II in the medical literature 40 years prior. With that visit an important relationship was established between ISMRD (The International Advocate for Glycoprotein Storage Diseases) and the GGC. 2006 saw the commencement of the first ever Natural History Study for Mucopolysaccharidosis type II and III with 15 families attending the GGC. With the commencement of the study there have been more than 60 affected patients from around the world supplying samples and information so this group of 9 very rare lysosomal diseases can be brought closer to therapy.

Extension of the Natural History Study to New Zealand and Australia

LDNZ is thrilled to have had the opportunity to build on its partnership with ISMRD and the GGC and be a part of building the largest Natural History study in the world for the very rare Glycoprotein Storage Diseases. Funded by the scholarship that Jenny received from AMP in 2008, and with a grant from the US MPS Society we were able to bring Dr Sara Cathey and Dr Lucia Horowitz to Australia and New Zealand to see 15 affected patients.

There were two long weeks of gathering data from the families and from their medical files in both Sydney and in Auckland. It has been a real privilege to work with Sara and Lucia and the families on a project that includes New Zealand and Australian families and which in time will create research opportunities for these very rare diseases.



Managed Clinical Networks

Many of you will remember the complaints we submitted to the health and disability Commissioner and Ministry of Health a couple of years ago outlining problems with access to appropriate clinical care. Although we have had resolutions to some of these complaints, we have continued to advocate for a better system to deliver these services.

On 17th November NZORD convened a workshop to discuss the development of managed clinical networks for patients with rare diseases. This is a huge project that LDNZ is directly involved with and will continue into 2010. We are thrilled to finally see the issues of access across DHB boundaries and problems with appropriate care for many adult patients with Lysosomal diseases, finally getting on the policy agenda of the Ministry and the new National Health Board. We are very pleased to confirm that the head of the NHB invited us to submit two proposed models for managed clinical networks for their consideration, and Lysosomal diseases will be one of the first group of conditions considered.

The board of LDNZ would like to wish you all a very merry Christmas and a safe and happy new year. John and Jenny will be closing their offices on 24th December. John will be back in the office on 11th January and Jenny 18th January. Take care and have a wonderful break.