Lysosomal Diseases New Zealand
Annual Report to the Trustees
December 2015

This report covers the financial year 1st July 2014 – 30th June 2015 plus some commentary on other things that have taken place between the end of the financial year and our meeting taking place in December 2015.

Overview: This financial year has again been filled with many highs and lows while we waited for Pharmac to begin funding new medicines through their contestable fund. Expectations of progress keep slipping away as our hopes are regularly dashed.

Finances: Raising funds continues to be a worry for LDNZ. The numbers of grants that we can apply for are becoming less and less as funding via gaming trusts decreases. During the year, we approached the following organisations:

- Endeavour Foundation, Four Winds Foundation, Lion Foundation, Pelorus Trust, The Trust Community Foundation, Infinity Foundation**.** ***All these grants were declined***.

Lottery Community remains our major funder and although last year they increased our funding in this round we have seen a decrease in funding, giving us a grant of $17,000. Securing funds via the gaming trusts is extremely difficult but we continue to submit applications through this pathway and receive small grants for Admin Salary and operations. We continue to receive small donations from families or in memory of people who have passed away and have received donations around Rare Disease Day.

- During the financial year we raised $42,912.11
- Our cash balance at the time of our Audit is. $61,656.14 as at 30th June 2015

Since the end of year balance we have received the Lottery Community grant which gives us a balance of funds in the bank of $78,656 in December 2015. This gives us confidence to continue with some of our activities. Also since the end of our financial year we have approached BioMarin, Genzyme and Shire requesting grants from each of the companies. We are optimistic of receiving support from these companies to help us pursue our mission. We have received occasional grants from companies in the past, but the difficulty in accessing the sort of community funds and gaming trust income that we got in past years, means we now need to rely on them for a larger slice of income.

Access to Medicines: This issue has become a thorn in our sides. Pharmac did a really good job shutting down our advocacy for medicine access for rare diseases, by announcing the orphan drugs fund nearly

20 months ago, then causing significant delays to any decisions. It is very difficult to get any traction with publicity when a policy is announced and we are waiting for a decision.

Pharmac have announced funding for one treatment under this scheme and are consulting on another, but these were not new drugs, they were already being funded by the NPPA Scheme. We have let Pharmac know that we are not pleased to see them funding in this way. The Contestable fund was clearly intended for drugs that were not currently funded by Pharmac. We have become very suspicious that the $5 million contestable fund has largely "disappeared" by being extensively delayed. Under the official information act we have applied for the Pharmac funding budget to see if we can discover if the funding still exists or if it has been sucked up by the DHB's. We are very clear that this is not so much a Pharmac issue but now a Government and DHB issue in that they have restricted the budget Pharmac manages, so it is getting more difficult for them to make any progress with the contestable fund.
We also saw an NPPA application for one of our adult Pompe patients declined – despite the fact that Pharmac had been offered a new price by Genzyme.

We believe we are unlikely to see any significant gains from the orphan drugs fund in the near future and we are preparing for a renewed campaign in the new year to raise this issue again.

Disability Support Issues:

1. **Funded Family Care**

Our advocacy on this topic did lead to a review of the policy by the ministry but a soft report and a weak response lead to no significant change to this policy. It remains a significant concern that families and their seriously disabled kids are being so gratuitously exploited in the application of this policy process, as evidenced by the very low uptake of the package by those caring for their own adult family members. Unfortunately, other groups that have led the charge on this have allowed the issues to go "off the boil" for now. It is difficult to see how LDNZ could revive this on its own.

2. **Access to health services**

It is notable that the extension of the metabolic service staffing and their added outreach clinics across the country are having some very good impacts on specialist oversight for some of our families. But there are still pockets of significant problems. The issues pursued through the H&D commissioner's office in 2006 seemed to produce some gains in various ways but in the case of Bay of Plenty DHB the whole agreement for care management has fallen into a black hole. We are exploring the best way to revive this issue to ensure that DHBs do what they are supposed to do, and do what they say they will do. Our work on this is intended to gather more information from families across the country to see how widespread the problems are.

WORLD Lysosomal Conference Feb 2015

LDNZ sent Prof David Palmer to this meeting and ISMRD paid for John and Jenny to attend. John and Jenny were also there on behalf of ISMRD and did significant work for LDNZ in advocacy for all Lysosomal Diseases, and to learn about new developments in research and therapies.

Of particular interest were discussions with PTC Therapeutics. John and Jenny were invited to their Monday evening presentation on this treatment. PTC124 has been developed into a newer drug called Translana. This therapy is read through therapy and reads through stop codons or nonsense mutations.

After the meeting, Jenny held further discussions with senior staff and the CEO of this company about the applicability of their technology to other lysosomal diseases, and in particular those that have little research occurring for them...

We also attended the round-table meeting with the company and 6 patient support group leaders, where the company explained their work to try and apply their product to the treatment of MPS 1 patients who have nonsense mutations.This discussion eventually lead to questions about the ultra-orphans, such as the Mucolipidoses. . They are very interested in this group as there is no therapy and no treatments being developed and they had also received the support of the UK MPS Society that this would be another good group for the company to consider.

One of the questions that came from this meeting was to consider how many other Glycoprotein Storage Disease patients might have a nonsense mutation, and as an example, figure were obtained as to how many patients with Alpha Mannosidosis patients have nonsense mutations. This lead to further discussions with the company who are now interested in these rare and complex disorders as we could quite possibly provide a good number of patients for them to work with.

During one of the break times Jenny met up with David Bedwell who is a research scientist and got to talking about Mucolipidosis and nonsense mutations. It's funny how from one tiny discussion things can lead onto to bigger things. David works for PTC Therapeutics who in turn went and spoke to the company about our discussion. Essentially both John and I spent some time talking to the company and to David who in turn both agreed that firstly they would like to work with the Mucolipidosis cell lines that have nonsense mutations, to see if Translana would work, and also explore combination therapy with other approaches. Jenny spoke to him at a meeting in July this year and progress was being made in getting cell lines from Brazil and South Carolina.

This is a good example of the great opportunities to promote research, that can arise out of such meetings, and highlights the value of attending such meetings. There were a number of discussions and links made with other companies and researchers at the same meeting, but this specific example highlights some possibly exciting steps forward for one very rare disorder.

Also of note at the WORLD meeting was the presence of NZ's Callaghan Innovation who are closely involved with a number of companies developing therapies for lysosomal diseases. It shows how small the world is and what an important role NZ science has in understanding and treating these diseases.

Cure Kids, the All Blacks, and the BARN meeting in Christchurch:

Batten disease research got a great boost when Cure Kids and the All Blacks did a major fundraiser prior to the rugby world cup. This resulted in a high profile for Batten families too, and we expect the fundraising campaign will lead to funds that can continue this important research here in NZ.
The collaboration of Lincoln and Otago Universities on developing gene therapy for the sheep with type 5 and type 6 Batten disease, is making good progress, and a boost to funding will allow them to take the research to the next level.
The year's exciting activities around Batten culminated in a Batten disease research network meeting at Lincoln in September where researchers, families, advocates and funders spent the day discussing progress and future plans.

Contact with families: LDNZ continues to be in touch with our families via the ¼ newsletters and phone contact as needed.

Family Support: Family support continues to be on an as needed basis. Some of the issues this financial year were:

- Advice on fundraising for Batten Disease and how LDNZ can hold funds on their behalf.
- Seeking information for a briefing on Disabled Children out of home care policy
- Palliative care issues
- Support given over the use of NASC hours and how the support agency should be using them to support the family.
- Enlisting the help of Morquio families to supply urine samples for screening
- Support given during the launch of CureKids funding project for Batten Disease
- Hospital access issues for patient with Tay Sachs in Tauranga.
- Care package sent to Lysosomal family who have been in hospital for an extended stay
- LDNZ in contact with Pompe Patients to discuss Pharmac and the delays in funding new orphan drugs
- Support for patient who is experience Fabry pain

Trust Accountability:
The trustees meet in December each year for their Annual Meeting and to plan activities for the year ahead. Activities based on that plan are 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.

Resignation of Dr Dianne Webster for the board of LDNZ:
During 2015 we received notice from Dianne that she would step down for her trustee role this year. LDNZ has been incredibly well supported by Dianne since the very beginning, even before we actually incorporated in 2000. She freely gave an enormous amount of helpful advice about things we could consider and challenges we would face, and how medical care and services, in particular, could be improved for our families. We are incredibly grateful for all she did for us, and we thank her most sincereley for her contribution to our organisation.

Keeping our books in order:
LDNZ has been ably assisted by Tim Hannagan who oversees the books and assists with GST returns and other matters. We are very grateful for his assistance with this.

Future priorities:
The 2014 trustees meeting developed a strategic plan to guide our future activities. This plan is attached for information. Circumstances meant that organising the planned conference did not proceed, but other possible events are under active consideration.

John Forman

Chairperson, LDNZ