

1ST ASIA-PACIFIC LYSOSOMAL CONFERENCE CHRISTCHURCH 20TH-23RD NOVEMBER 2008

FSG was very fortunate to have two representatives, Megan (FSG President) and Mardi (Vice-President), attend the Lysosomal Conference in New Zealand. Mardi and Megan have compiled the following report about the conference to share with FSG members.



What a wonderful event this was, after 2 years of planning, fundraising and planning, MPS (Mucopolysaccharidosis) Australia and Lysosomal Diseases of New Zealand combined forces to host an amazing, organised Conference. Both families and professional world leading experts in Lysosomal diseases came together to meet and share information and provide a knowledge base that is difficult to gain in New Zealand. The conference started out as an Asia-Pacific conference and became an International meeting with Professionals and other support group leaders coming from: Germany, Norway, USA, Japan, Hong Kong, United Kingdom, Israel, Canada, Australia and New Zealand.

The opening of the conference was held on Thursday 20th Nov, at 6pm on the Village green at Chateau on the Park, as the sun was setting. Delegates were welcomed by KoTane the Cultural experience group from Christchurch. Professor John Hopwood from Adelaide was chosen as the conference chief and Bowen Oliver MPS6 represented all those affected by Lysosomal Disorders. Peter Joyce from the Christchurch school of medicine opened the conference with John Forman LDNZ and David Oliver from MPS Australia responding. It was a wonderful time of fellowship, meeting old and new friends. Mardi and I were the only 2 represents of Fabry's Support Group Inc Australia attending and we were very excited to meet some new Fabry's patients from various parts of New Zealand.

Friday began the first full day of the conference program covering a very wide range of topics. The plenary addresses were presented by Prof. John Hopwood Australia, Steve Walkley USA and Brian Winchester UK. Their presentations gave an overview of Lysosomal Diseases, The challenges of LSD's in particular the problems faced in trying to cross the blood brain barrier and the central nervous system and Biochemical and Molecular diagnosis of Lysosomal Diseases.

Over 250 babies born in Australia are born with a "Lysosomal" disease. Most babies present 'healthy' at birth and symptoms are progressive. Over one quarter of the world's population is directly/indirectly affected by a rare disease. Lysosomal Storage Diseases (LSD) affects 3-5% of the general population.

LSD's in Australia - 1: 1,000 births

Some of the hurdles are:

- Treatment of CNS pathology
- Screening detection – newborns
- Prognostics in asymptomatic patients
- Definition of irreversible pathology
- Maintenance of long term treatment

There were presentations on Batten disease and research into Sanfilippo Disease.

Ed Wraith from the UK discussed existing therapies their benefits and limitations. Enzyme replacement therapy is a safer approach for treating LSD's but is ineffective in disorders affecting the brain as the enzyme is unable to cross the blood brain barrier. ERT is very expensive and is not available to patients from countries that have other competing health care needs.



Photo from left Maureen Murray (NZ), Aubrey Walters (NZ), Mardi Versteegen, Beverly Murray (NZ) – back, Megan Fookes, Kathleen Walters (NZ), Anna Percy (NZ), Neville & Carolyn O' Kane (NZ).

The day closed with a Poster viewing session, where each of the Lysosomal Disorders had informative displays and posters discussing their particular Lysosomal Disease and its potential treatments. That evening Genzyme representatives Dan Brown and his wife Amanda and Anthony Earp invited the New Zealand Fabry patients along with Mardi and I to dinner. The next day the presentations looked at Clinical Management of the various disorders.

Highlights for us were Dr Jim McGill of Queensland, looked at the benefits of commencing Enzyme Replacement Therapy early:

With accumulating experience in the use of ert, especially in the MPS's and Fabry disease, it is becoming increasingly clear that many of the features of these disorders are irreversible if therapy is started late. The benefits of commencing ERT early in life had been clearly demonstrated in animal studies 1, 2. For MPS vi there are now several studies of sibling pairs in which the second affected sibling has started therapy at a young age with much improved response compared to the older sibling. Dr McGill presented his findings with clinical information collected from patient data.

Dr Kathy Nicholls of The Royal Melbourne Hospital Australia presented on Chaperone Therapy for Fabry Disease:

The mutations causing Fabry disease are many and varied. While some result in an absolute enzyme deficiency in the cells containing that mutation, the problem resulting from many other mutations is that defective enzyme is produced. This situation is not unique to Fabry disease, but is common to many diseases. An enzyme may be defective because its 3-dimensional structure is altered so that it does not effectively transit its pathway within the cell to its site of action- for Fabry enzyme, the lysosome. Resulting, small molecules have been specifically designed to bind to defective enzymes, with the aim of overcoming this problem and boosting enzyme activity. The drug AT1001 has been developed by Amicus Therapeutics (USA), and has been tested in Phase 1 and Phase 2 multicentre trials. In vitro testing enables the degree of boost to enzyme activity induced by AT1001 to be measured, with variable drug efficacy depending on the patient's specific mutation. Data to date indicate that AT1001 is safe and well tolerated. Measures of invitro enhancement of enzyme activity are consistent with in vitro testing. Results of Phase 2 studies will be presented 1, 2.

After lunch the conference was broken down into workshops for families and professionals. Each workshop had three short presentations and then open discussion, where families could ask questions and share information on how they manage their particular disorders. Professor David Sillence of Westmead Hospital Sydney Australia presented his information about treatments for Fabry Disease. Kerrie Devine, ERT treatment nurse from Westmead Hospital Sydney presented data based on treating patients with Pompe and Fabry Disease. We also discussed home infusions, many patients are now receiving ERT in their homes and some prefer to be treated at their local hospital under the care of the hospital staff.



Professor David Sillence presented "Fabry Management 2008" and this has been included as a separate attachment from this newsletter for FSG members to read.

Photo from left Prof. Bob Jolly, Prof. John Hopwood and Prof David Sillence

That evening was the Gala dinner where everyone dressed up and enjoyed some good old Kiwi hospitality. KoTane came back and entertained with some traditional dances and games and interacting with the audience.

Later in the evening LDNZ honoured Prof David Sillence (from Sydney Australia) and Prof. Bob Jolly (Palmerston North New Zealand) with Life time awards. Prof Sillence (Australia) has played a very special role in the lives of many Australians and Kiwis affected by LSD's.



Photo from left Judith Forman (parent of twins with Alpha-Mannosidosis), Mardi Versteegen (FSG Vice-President), Megan Fookes (FSG President) and John Forman (parent of twins with Alpha-Mannosidosis and Chairman of Lysosomal diseases NZ).

LDNZ also presented the Australian MPS Society with their 25th Anniversary cake and David Oliver (President of MPS Aust) gave a presentation about the history of MPS Society. A photo and cake cutting ceremony took place for the past and present MPS Society presidents.

Sunday the last day of the conference brought together all LSD's to discuss the issues surrounding New Born screening and Access to Medicines. In particular in the case of New Zealand where treatments exist for LSD's and are approved but there is no funding. In Australia Fabry patients are very fortunate to have access to treatment via the Life Saving Drugs Program. In New Zealand they have no such program and they are the only developed country to not have such a program to fund an already approved treatment!!! Hence, many Fabry patients are not receiving any treatment or are commuting to Melbourne Australia and are participating in the Amicus AT1001 trial (Chaperone Therapy) under Dr Cathy Nicholls.

After the closing of the LDNZ Conference, we came together over a buffet lunch in preparation to fly out that afternoon back to Australia.

Finally FSG would like to thank Genzyme for their sponsorship which enabled the two FSG representatives to attend this conference. Without this generous support it would not have been possible.

