



Chairman's Report continued

Mission Statement

- Increase community awareness and understanding of lysosomal storage disorders, their effects and potential solutions, through education and publicity;
- Increase political recognition and understanding of lysosomal storage disorders, their effects and potential solutions, particularly the cost-benefit advantages offered by therapies;
- Encourage research involved with the diagnosis, management and treatment of patients affected by lysosomal storage disorders;
- Maintain close links with family support groups;
- Promote and establish links with support associations for other, non-lysosomal, organelle diseases
- Promote and establish links with similar organisations internationally.

LDA has been established as a national umbrella group, and its role is to establish and represent the needs of all patients affected by lysosomal storage disorders, their families and carers. LDA aims to emphasise the common aspects of these disorders to maximise resources for support services, treatment and research. The aims are further outlined in the Mission Statement, which is reproduced in this issue.

LDA will work together with parent support groups built around specific disorders. It is recognised that these groups perform a vital function in helping patients and families come to terms with a new diagnosis, and with ongoing day-to-day management and support issues. LDA does not, in any way, wish to minimise or supersede the role and functions of these groups. Because of the individual rarity of many of these disorders, LDA intends to pursue issues generally beyond the scope of the family-based groups. The opinions and contributions of the support societies are central to LDA. The Board is very keen to ensure that LDA is relevant to the needs of the support groups for lysosomal storage disorders.

Every year, 50 new patients are diagnosed in Australia: 40 of these patients will be affected by Gaucher, Fabry, Pompe and Batten diseases, Hurler, Sanfilippo, Hunter, Morquio, Maroteaux-Lamy syndromes or metachromatic leucodystrophy. The other 10 patients will be affected by one of more than 30 other lysosomal storage disorders. A brief overview about lysosomal storage disorders, the specific enzyme deficiencies, gene location and the status of animal and human clinical trials is presented in this newsletter (see page 5). This information will be updated in each issue of our newsletter to reflect the scientific and medical advances. In each issue, we will also profile a specific lysosomal storage disorder, beginning with Fabry disease in the next issue.

Over the past 20 years, research has led to the development of techniques to enable the early diagnosis and treatment of

patients suffering with lysosomal storage disorders. Two therapies are currently being intensively researched – enzyme replacement therapy and gene replacement therapy. Both forms of therapy will enable the stalled recycling process to start and work normally, thereby reversing the pathology caused by the accumulated storage material. Whereas further research is required to enable the successful application of gene replacement, enzyme replacement therapy is currently in clinical practice, in trial or planned for seven different lysosomal storage disorders: Gaucher, Pompe and Fabry diseases, Hunter, Hurler-Scheie, Maroteaux-Lamy syndromes and Niemann-Pick disease type B. Enzyme replacement involves the regular infusion of manufactured enzyme into the patient to compensate for the missing specific enzyme. This form of therapy has been most successful for patients affected by Gaucher disease. More than 3,000 Gaucher patients are now treated by this method world-wide.

The success of enzyme replacement therapy for Gaucher disease has offered an improved quality of life for patients, families and carers, and has given hope of a solution to patients affected by other lysosomal storage disorders. An overview of enzyme replacement therapy, its benefits and limitations, is presented in this issue.

This, our first issue of the newsletter, is primarily one of introduction to LDA, its aims and objectives, and its Board. Newsletters will be published quarterly, with information that we hope you will find informative, accurate and up-to-date. As well as being mailed, each issue will also appear on the LDA website (<http://www.lda.org.au>). Feedback on the content of our newsletters, and suggestions for future issues are encouraged. These can be emailed, faxed or posted to the Public Officer.

John Hopwood
Chairman