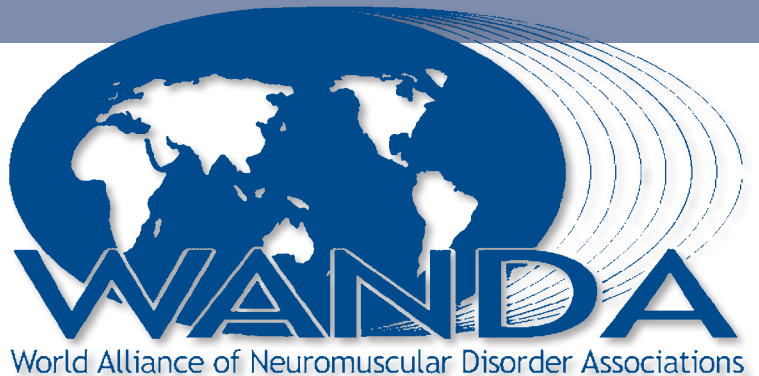


WANDA

prospectus 2009 - 2012

Including the proceedings of
Fifth International Assembly of the World Alliance
of Neuromuscular Disorder Associations



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President's Statement

It is a great pleasure to be able to present this Report and Prospectus for the World Alliance of Neuromuscular Disorder Associations (WANDA) and to thank Immediate Past President, Piraye Serdaroglu, for guiding WANDA until mid 2007.

Although the Report from the fifth International Assembly of WANDA in Istanbul in 2006 is very late (for which I apologise), I hope that you will enjoy reading it and, perhaps, if you were lucky enough to be there, you will be able to relive the atmosphere of optimism that was expressed at the meeting.

As a second component to this publication, our Prospectus provides some history, background and the recent progress of WANDA and then looks to the future. It suggests methodologies and models for realising treatments for neuromuscular diseases and summarises what WANDA expects to achieve in the coming years.

Finally, WANDA will hold its sixth International Assembly in conjunction with the exciting program of the twelfth International Congress on Neuromuscular Diseases (ICNMD XII) in Naples, Italy, between 17th and 22nd July 2010,

In line with previous assemblies in Munich, Kyoto, Adelaide, Vancouver and Istanbul we would very much like to welcome you in Naples, listen to your progress, consider obstacles and opportunities, discuss common policy issues and decide on WANDA's program and projects for 2010 - 2014. During the Assembly, the latest news will be reported to you from the sessions of the ICNMD XII. The WANDA program promises to be very informative and to allow positive interaction between people with neuromuscular disorders, their families and carers, allied health professionals, delegates from relevant national and international neuromuscular disorder associations, as well as, specialist clinicians and scientists.

For a small, but increasing, number of neuromuscular diseases there are approved and effective drugs on the market. And there are more to come. In Naples we want to talk to you about what the associations for neuromuscular diseases can do to increase research effort, support drug development and facilitate general advances in the field of treatment and management. To achieve our goals a united effort is necessary from all stakeholders but most prominently from the area of science. It is for this reason that we would be most pleased with your participation.

We especially ask all of you to advertise the twelfth ICNMD among your medical and scientific network and to encourage doctors and researchers to attend. Please block the date into your diaries for 2010 and include these events in your agenda for promotion.

With best wishes and sincerely yours,

Allan Bretag Adelaide, March, 2009

Reports from the
**5th International
Assembly of WANDA,
Istanbul, Turkey,
5th July 2006**

Introduction



*Prof. Piraye Serdaroglu and
Prof. Coşkun Özdemir
in Istanbul 2006*

On the 6th of July 2007, the quadrennial meeting of the World Alliance of Neuromuscular Diseases Associations (WANDA) was held alongside the XIth International Congress on Neuromuscular Diseases (ICNMD-XI) in Istanbul. The meeting was hosted by the Turkish Association of Neuromuscular Diseases. All NMDA associations were invited and the meeting was open to patients, carers and families.

Neuromuscular patient/parent organizations (NMDAs) draw inspiration from sharing experience with each other in a supportive and cooperative environment. In the same way, each national organization can seek guidance from the organizations of other countries, leading to greater integrity and team-work. The shared goals can only be accomplished by a strong network of National and International organizations sharing information with patients and carers. European Alliance of Neuromuscular Diseases Associations (EAMDA) sets a great example as an umbrella organization for the structured solidarity of NMDAs in Europe. The targeted collaboration between the EAMDA and the scientific community, European Neuromuscular Center (ENMC), has facilitated very important studies on NMDs.

At the world level WANDA is the non-profit umbrella organization and its strength depends on the collaboration of continental and national structures. This network is extremely important as the advances in scientific research have come to a level where they can meet with their two vital allies: the patient / parent organizations and the industry.

It is gratifying to see the beginning of an era of success. The role and input of WANDA is largely responsible for developing strategies to join forces i.e.: to bring the patients, carers, the scientific community and industry together. In this way we can reach our goals of finding cures for neuromuscular diseases, draw lessons from past experiences and successes in order to design future paths. This is why the main theme of the WANDA - ICNMD-XI plenary meeting is "Roadmap to Treatment".

The plenary meeting was followed by reports and discussions about different experiences of NMDAs all over the world. These final and very valuable contributions maintained the main spirit of WANDA, forming the basis for drawing the "Roadmap to Treatment".

Piraye Serdaroglu, President, WANDA, July 2006

Welcome Speech from Turkish NMDA President



*Poster Display at the WANDA
conference in Istanbul*

We are very happy that the WANDA meeting is being held in Turkey along with the International Neuromuscular meeting and it gives us great pleasure to welcome the representatives of WANDA. We look forward to getting together with the members of international associations. As you all know, neuromuscular disorders are prevalent all over the world. The majority of these conditions are still untreatable. Considerable number of these disorders are hereditary or genetic diseases. Until a cure is found, non pharmacological management and other ways of improving the quality of life of patients remain very important issues. Obviously methods for patient management are rapidly improving but the application of these methods vary across the countries due to availability and socio-economic/cultural factors in society. While there are numerous centres of excellence, satisfactory genetic and rehabilitation centres, nursing care at home and availability of technical devices for the patients in highly developed countries, there are only a few specialized centres dealing with neuromuscular disorders in developing ones. Unfortunately Turkey is an example of the latter. As you all know opportunities to find physical therapy, educational assistance and vocational rehabilitation are limited for neuromuscular disease sufferers. The concept of rehabilitation is still not appreciated or understood by many members of our society and our social services are far from satisfactory. Also we need to improve patient education in order to protect them from expensive and useless alternative therapies which are offered around the world. There have been some pleasing new developments in the treatment of neuromuscular disorders providing scientists, patients and families with hope. New drug trials have also been commenced in the last few years. However we should realize that cures for these diseases may still be some years away. We should emphasise on the importance of satisfactory management and try to improve social and rehabilitation services in our country.

Professor Coşkun Özdemir, President of the Turkish Muscular Dystrophy Association



The International Pompe Society

The International Pompe Association was established in July 1999 in the Netherlands in response to encouraging developments towards an effective medicine for Pompe disease, a rare and progressive neuromuscular disorder. It was felt that Pompe patients required a strong global voice to represent them in discussions with industry, health authorities and governments.

The IPA board consists of representatives from patient organisations from three continents who provide extensive experience and expertise.

Its mission is to:

- Campaign for early diagnosis and effective, affordable and safe therapies
- Strive to provide information and support to all patients, their families and others with interests in Pompe disease

Successful patient organisations have several essential characteristics:

- Work closely as a team
- Understand each member's expertise, abilities and knowledge
- Communicate in a common language (English)
- Established contact with relevant parties
- Clear goals
- Well informed, constructive, critical and alert
- Organised for strength
- Maintain short communication lines for rapid and effective response.

Over the years the IPA, with members in 34 countries, has evolved into an international patient network for information exchange. Through this they were able to make major contributions to Pompe related research:

- Identifying patients for clinical trials
- Organising international conferences
- Commissioning an international patient questionnaire. This led to scientific publications and encouraged patients to participate in Genzyme Therapeutics' Pompe Registry

Since we work for patient's interests we must act as an equal partner to professionals. There is a mutual acceptance of distributed responsibilities and cooperation between patient associations, physicians, hospitals and Genzyme.

Contact with all parties is very important: Individual patients in different countries provide information and knowledge of what is happening world-wide:

- Cooperation with physicians to support patients and to contribute to research and knowledge
- Finding patients for clinical trials and helping them access treatment
- Maintain contacts with industry to support and inform patients, to share experience and knowledge



Maryze Schoneveld van der Linde (centre)

Communication is the most important aspect of maintaining a network. The IPA website ([www.ipa-pompe.com](#)), together with an online forum where patients worldwide can meet, are good tools to inform patients and families to better understand patient needs.

A patient organisation must be aware that it will interface with a diverse network of parties, some of whom support its goals, but also others that will resist them. The IPA has dealt with animal welfare organisations that were opposed to the use of animals to produce medicines. We told our story on television and written media to show the general public and politicians how beneficial and lifesaving this medicine would be.

The IPA has learned much over the years, and we'd like to share our experience with you to help you develop and become a successful patient organisation. You should have a proactive and critical attitude towards developments, so don't wait for bureaucracy and competition. Establish close contacts with all parties. Empower patients to become leaders. Use your own knowledge and experience. The professional support of an experienced and strong patient organisation is very important. It can provide you with access to pre-existing networks, documentation materials, skilled staff members, expertise etc.

In April 2006 the enzyme replacement therapy, Myozyme, was approved by both EMEA and the FDA. It's on the market now and many Pompe patients worldwide have started treatment. We are now working for patients in countries whose governments have still to approve Myozyme or its funding. We will continue our work until our mission has been fulfilled.

The European Neuromuscular Centre (ENMC)



Participants at the Myotubular Myopathy Workshop

The European Neuromuscular Centre (ENMC) was first established in 1989 in Paris, France.

The secretariat is based in Baarn, The Netherlands and the countries involved are Austria, Denmark, France, Germany, Italy, The Netherlands, Switzerland and the United Kingdom.

The ENMC aim: to usefully and efficiently contribute to the eradication of neuromuscular diseases, to improve efficiency in European neuromuscular research and to facilitate and support research communication between European (and international) researchers and clinicians.

Between 1990 and 2006 the ENMC held 142 workshops with a range of topics such as Duchenne Muscular Dystrophy, Spinal Muscular Atrophy, Congenital Muscular Dystrophy, Charcot-Marie-Tooth disease, Emery-Dreifuss Muscular Dystrophy, Myotubular Myopathy and Fascioscapulohumeral Dystrophy. Over 2 000 people participated with 28 countries represented.

The ENMC trial network aim: to facilitate and support relevant European multi-centre clinical trials on neuromuscular diseases making use of the existing ENMC prestige, ENMC network and ENMC facilities.

One of our future challenges is to start including structured input by patients in developing trials. It is hoped patient representatives will help solve ethical issues, will aid in communication with other patients, parents and carers, will help recruitment of patients in trials and will assist in identifying funds for trials.

The ENMC trial network can help researchers with key steps in devising and designing clinical trials. This may involve identifying trials relevant to our neuromuscular disease patients, giving assistance drafting trial protocol, offering assistance with trial funding applications and finding or joining partners for multicentre trials. We hope to begin a disease register, to offer training courses for trial development and to identify potential treatments through our interactions with scientists.

So far the ENMC has achieved a successful collaboration between scientists and patient organisations with many high quality trials published. We believe the ENMC has continued to develop along with science and technology. We are ready for the trial decade.

Dr Peter Streng, Chairperson EAMDA

The Patient Perspective

Strategic Role of Patient Organisations in the Research and Development Process

The EAMDA or European Alliance of Muscular Disorder Associations was established in London in 1971. Since commencement our objectives have been to help people suffering from neuromuscular diseases by assisting with both physical and mental wellbeing, and supporting research into causes, treatment and cure for all neuromuscular diseases. Our organisation has twenty one members throughout Europe.

Lately there have been changes in the way healthcare is provided, from the 1970s when the focus was on diagnosis, the 1980s when quality of life became important, the 1990s when a more global, holistic approach to clinical management was the ideal, to the present when prevention, testing, genetic screening and individualised treatments are the goals.

In parallel, there have been changes in patient's roles and attitudes. In the 1970s, patients were not expected to have any input into their treatment. In the 1980s they were empowered to educate themselves about their disease and to begin to have a say in treatment choices. From 2000, patients have begun to be seen as joint managers of their condition, working with doctors and therapists in a position of equal power. In the future we expect patients will have even greater roles in the areas of scientific research, drug studies and collection of epidemiological data.

Already patients have an indispensable role in educating the public about neuromuscular diseases, advocating for research studies to take place, offering to be part of treatment studies and raising funds for research, treatments and equipment. As well as being active partners in Muscular Dystrophy associations around the world, patients have other groups in which they specifically work with science and industry, offering up their unique experience and history to assist scientists to solve problems in disease management, in monitoring the effectiveness of new therapies and finding candidates for research studies.

In future patients might be expected to play a larger role in recruitment for clinical trials. They might be expected to have a say in trial design, in counselling other patients about expectations, providing information to other patients before and after trials and in reporting on trial outcomes.

When a trial is commenced, there are many factors needing consideration such as comprehensibility of patient information, the risk vs benefit, the quality of the research centre proposing the trial and reimbursement/ insurance for participants. The patient perspective may assist researchers in detecting potential problems in these areas before they arise.

Although some of these patient organisations are already having input into trials of new treatments, there is still a need for greater funding for these groups if they hope to achieve their potential.

Dr Peter Streng Chairperson of EAMDA



Parent Project Muscular Dystrophy

The diagnosis of Duchenne muscular dystrophy (DMD) is devastating for patients and their families. There is no standardised treatment, no cure, and boys with this disorder typically die by age 25 years because of respiratory or heart failure. Until 1994, parents of boys with DMD had nowhere to turn for support and no concerted efforts in clinical research were being made. This changed when Parent Project Muscular Dystrophy (PPMD) was founded with the goal of creating awareness of DMD and generating interest and funding for research.

PPMD has grown from a support and advocacy group to an organization that provides various services for parents and children with DMD. Services include an annual meeting, education, a quarterly newsletter, and a comprehensive Web site (www.parentprojectmd.org). The group also has formed partnerships with industry to encourage drug development and research funding.

DMD is the most prevalent form of muscular dystrophy, affecting about 1 in 3500 male newborns. Pat Furlong, director and founder of PPMD, knows firsthand about the disease and its consequences. The genesis of the parent-centered organization was spurred by her frustration in finding support and resources for her 2 sons. Both had DMD and died at ages 15 and 17.

When her children were diagnosed in 1984, Furlong began traveling to clinics across the United States to learn more about standards of care and research for this disease. "There was no money and no federal investment in research," she said "Doctors told me there was nothing they could do for my sons and that they would ultimately die in adolescence of the disease."

Even though the gene for DMD was identified in 1986, no prospective treatments were discovered for the next several years. Furlong was amazed by the lack of communication about DMD. Scientists were not talking to clinicians and clinicians were not talking to patients and families. She believed that parents of patients should mobilize and lobby for research on DMD. Thus, the PPMD was formed.

French Anderson, a Nobel Prize winner for genetic discoveries, attended the inaugural meeting of PPMD at Furlong's request. He discussed the idea of viral gene therapy and the importance of centres of excellence in DMD care and research.

In 1995 the University of Pittsburgh Medical Center became the first centre of excellence, providing imaging and research on basic science and the feasibility of adult-derived stem cell treatment for DMD. The University of California soon became a second centre of excellence, focusing on the inflammatory component of DMD.

The community of PPMD members began advocacy efforts in earnest, writing letters to Congressmen and hiring lobbyists in Washington. As a result, the federal government passed the Muscular Dystrophy Community Assistance, Research, and Education Amendments (MD CARE) Act in 2001. This led to new funding, the establishment of 6 research centres of excellence, and a surveillance program through the CDC, with the goal of creating a registry of boys with DMD. A Muscular Dystrophy Coordinating Committee was formed with a mandate to design and implement a research and care plan.

PPMD provides a virtual community for parents of boys with DMD. "This is a devastating and rare disorder, and for at least 40% of boys, there is no family history of the disorder," Furlong said. "Parents of these children need support."

PPMD is working with Cincinnati Children's Hospital Medical Center to develop a standard of care that will help boys live longer, walk better, and have an improved quality of life. The collaboration has resulted in an evidence-based DVD on the care and management of DMD, which has been approved by the CDC.

*Extracted from the Applied Neurology article March 2007 by
Alice Goodman*

United Parent Projects Muscular Dystrophy (UPPMD)

UPPMD is a project that is owned and managed by organisations that have been set up in many different countries since 1994. These organisations are run by parents for parents of children diagnosed with Duchenne or Becker muscular dystrophy. UPPMD is managed by parents who have been chosen to lead national projects. The organisation came together as it was realised that there was no sense in working in isolation from each other. We all share the same aims and dreams and those will be realised more efficiently if we share our collective experiences and resources. There are many challenges that face our community, in particular there is a need to advance scientific research, by a collective funding such research and promoting the sharing of information amongst researchers. It is also important to ensuring that there is no duplication of effort in that research. There is also a compelling need to ensure that wherever we are in the world, our children can benefit from a standard of care that informed by the best practice of the best clinicians from all over the world. It is equally important that information available to parents is current and is based upon the latest research. In joining together, we have not set up a bureaucracy. UPPMD is a responsive Parent led organisation. Only when organisations listen to parents concerns, will parents have the faith to support their work. This is why we set up our national organisations, and now we have turned our ears to all the parents of the world. DMD is a treatable condition and as such, you will find information about quality of care, the latest test research and events that are taking place near you. We are not alone in our search. All of the projects in our community are run by parents for parents. If your country is not represented in our community please contact us. We can advise you on how to set up a project, share with you the latest research information and provide the support that stems from our world that is 'United In Hope'. The mission is to improve the treatment, quality of life and long term outlook for all individuals affected by Duchenne muscular dystrophy through research, education, advocacy and compassion.

Streamlining the pathways towards treatment by parent/patient groups and NMDA's together with science and industry:

- Publicity to raise awareness
- Funding research towards a cure
- Lobbying
- Connecting different worlds
- Clinical trials
- Collaboration with Industry

Parents:

- LIVE with the Disorder
- Often have novel ideas about treating the disorder
- Care intimately about progress
- Provide input for clinical trials
- Access to the patient population and key opinion leaders
- Partnering provides realistic hope
- Inspirational

Industry:

- Business mentality: driven, accountable, responsible
- Commitment to commercializing innovative science
- Care intimately about progress
- Experienced in advancing preclinical studies and conducting clinical trials



Elisabeth Vroom, founder of UPPMD

Report from Turkish NMDA

We had a big meeting in ISTANBUL with the WANDA family on the 5th of July 2006. The timing was perfect because the International Congress on Neuromuscular diseases was held concurrently in Istanbul. Turkish families affected by neuromuscular disorders had a chance to speak with researchers, scientists, professional doctors and affected families from foreign countries. In this way, we have improved our knowledge of neuromuscular diseases. We have also seen how the system is working abroad as 200 presenters shared their stories.

After the Pompe disease research developments were presented, we had lots of optimism for other neuromuscular diseases. Neuromuscular diseases are rare so after seeing Pompe's success, we came to the belief that if other associations had been given more support in finding new treatments, the researchers may meet with the same success. In my opinion, researchers are actively studying genes and mutations. However, there are many important questions researchers still have not answered. Large corporations, scientists, drug companies, medical associations, politicians, social organisations and the media should be approached to give their support for research developments. If they all work together, it would be greatly encouraging to those affected by neuromuscular diseases.

We had a chance to talk with patients and families at the end of the annual meeting. From our conversations, we came to understand that many of the patients have been waiting in the hope that new treatments will become available and many are also waiting for answers about new research findings. They would like to know about the developments and treatments but unfortunately they have only been given very basic answers when they would like to be given all of the details. Speakers were only given a short time to answer the questions of patients and their families so some were unable to have their concerns answered. We also felt that there wasn't enough interest by scientists in the WANDA meeting and it would be pleasing to see more presenters in future..

" We are a huge family all over the world. We want improved quality of life, recognition of our rights and access to new opportunities. Our starting point is human rights. If we are prepared to lead in new directions, we can succeed together ". We have to keep in touch more. Also we have to find ways for our lobby and finance facilities to raise the necessary funds to support research, industry, patients and their families.

Once again, I want to say thank you very much to all who attended the conference, to presenters, supporting businesses and organisations.

Mr Hakan Ozgul, Accountant for the NMDA Turkey



Mr Hakan Ozgul

Developments in Research at the MDC

The Muscular Dystrophy Campaign is the major funding resource in the UK for scientific and clinical research projects focussing on understanding the symptoms and potential causes of muscular dystrophy and related diseases. Since it was founded 47 years ago the charity has pioneered research into muscle disease and as a result the understanding of the underlying genetic causes and their effects has advanced. This has not only had an immediate impact on the diagnosis and prognosis of the different types of muscle disease, but it has also led to the identification of treatment targets and therapeutic approaches.

The charity invests £2 million every year into research projects most of which are based at Universities and Research Centres around the country. An efficient application process including a rigorous peer review system ensures that only science of the highest quality is funded. Our research strategy determines our priorities as a patient organisation and these priorities will have a strong impact on our communication and campaign directions.

The three major aims are:

- To reach a greater understanding of the basis of normal and abnormal muscle function with the long-term aim of identifying candidate treatments or management strategies for muscular dystrophies
- To fast-track promising treatments when they are close to clinical trials to ensure rapid transition from the laboratory to the clinic
- To progress safe and efficacious treatments through clinical development to the marketplace in the minimum timeframe
- The charity currently supports 22 live research projects into 14 different diseases.

Major achievements:

- Prof Kay Davies and her group have discovered a protein called utrophin, which can replace dystrophin, (the protein missing in boys with Duchenne) if it is present in sufficient quantities in the muscle. Large scale drug screens were set up to find a compound which increases the production of utrophin. VASTox, a leading biotech company (co-founded by Prof Davies) announced last year that they had identified a number of compounds which increase the level of utrophin in a living organism. The company envisages the start of clinical trials in the coming years.
- Prof David Rubinsztein in Cambridge is developing a treatment for oculopharyngeal muscular dystrophy (OPMD). OPMD is characterised by a potentially toxic accumulation of proteins forming in the nucleus of muscle cells. Prof Rubinsztein has identified drugs that might be able to reduce this effect and has recently published his work in the Journal of Human Molecular Genetics.
- Prof Doug Turnbull and his team in Newcastle obtained approval from the Human Fertilisation and Embryology Authority (HFEA) for their research, investigating a pioneering and challenging approach to prevent the inheritance of mitochondrial myopathies.
- Dr Mary Reilly in London is conducting a clinical trial to investigate the benefits of Vitamin C for Charcot-Marie-tooth disease type 1A (CMT1A). Vitamin C has been shown to improve the symptoms in an animal model of the disease, but it is now crucial to test whether it has the same effect in humans. This study will be combined with an ongoing trial in Italy and if this treatment works, a low cost effective therapy will be available for this common disease.

MDC: United Kingdom

The Muscular Dystrophy Campaign assisted a UK scientific consortium in applying for government funding for a clinical trial into the safety and efficacy of using antisense nucleotides to restore dystrophin production in boys with Duchenne. The study is led by Professor Francesco Muntoni in London and the objective is to turn a severe Duchenne type into the milder Becker type. The work began in 2005 and considerable progress has been made since then in optimising the technique, preparing for the clinical trial and in obtaining some additional funding to start addressing some of the long term issues of this treatment approach. In addition to having secured £1.6 million from the Department of Health, the charity has committed an additional £500,000 over the next three years for research projects that are directly linked to the "Exon Skipping-Molecular Patches" clinical trial.

So what are the future challenges?

Ensuring that promising technology is brought from the laboratory into a clinical context will come more and more into the focus of the charity's attention and concern. But supporting "Translational Research" is costly and it is necessary to find alternative avenues to facilitate or even fund them. Most of the genetic diseases the Muscular Dystrophy Campaign covers are very rare, a fact that presents a number of problems. A huge breakthrough was achieved by Prof. Kate Bushby and Prof. Volker Straub who were awarded £6.7million to set up TREAT-NMD (Translational Research in Europe - Assessment and Treatment of Neuromuscular Diseases) which brings together 21 partner organisations from 11 European countries to develop a network of excellence including clinicians, scientists, charities and companies. The Muscular Dystrophy Campaign is actively supporting this initiative which has been designed to address the fragmentation that is currently hindering the efficient development of therapies for neuromuscular disorders.

Dr Marita Pohlschmidt Director of Research, Muscular Dystrophy Campaign.



MDA: China

Patients and family members affected by neurological and muscular diseases in China have for too long been a silent group, left alone and unsupported by the rest of society. They have been unable to speak for themselves. At the same time, carers and family members may come to the verge of physical and mental collapse, as they are worn out by the countless hospital visits, frustrating experiences and the stress of looking after the patient all year around. They have no time, no energy, and no financial supports left to call for the assistance and attention they need. The people around them, including government officials, have little awareness about this group of people suffering from neuromuscular disorders, much less the ability to give them the necessary concern or fair treatments. As a result, this tragedy always repeats itself. It is estimated that there are 4.5million neuro-muscular disease patients in China and the population directly involved (patients, families and carers) are more than 13.5 million, not to mention the size of the population who are indirectly involved.

In April 2002, MDA China organized a symposium on neuromuscular diseases in Shanghai. Among the attendees were the prominent medical specialists and scholars, patients and their guardians, and local government officials. The symposium received overwhelming recognition among peers and the attending group came to a consensus that MDA-China, which represents the group of patients and families suffering neurological and muscular disorders along with the national and international healthcare professionals, should be established. It was in the very same month that the MDA China web site was launched. So far our web site has received more than 235,000 hits, over 3000 members, and 20,000 posts in the forum. With copyright authorization from MDA associations in America, Japan, France and Canada, the medical specialists have helped with the translation of many articles, which keeps us up-to-date on the latest research developments and information on neurological and muscular diseases around the world. The research and medical innovations bring hope and confidence to our patients. Forum discussions have involved the day to day needs of patient and carers, the difficulties in education, employment, medi-care medi-aid and welfare they have already faced or may face in the future. They have been active in posting their stories and sharing their ideas in the forum. In this way, more and more valuable experience becomes available to others. Some of the patients have participated in the management and maintenance of the web site.

Ms. ZHU Changqing (pictured), the head of MDA China, has made speeches in universities, propagating knowledge of neuromuscular disease and helping those young patients from indigent families to pursue their studies. For those who are unable to go to school, volunteers offer home tutoring. The College of Acupuncture and Massage of Shanghai University of Traditional Chinese Medicine organized volunteers who, after proper training, provide treatments for patients in their homes. Ms. Zhu has phoned and visited patients personally to hear their stories and offer support and assistance. Her efforts this year have attracted Newspaper, Radio, TV reporters and media reports for MDA China . We are beginning to receive assistance from companies and businesses for people at all levels. Working around their tight schedules, our medical specialists helped to revise the application for establishing MDA China, which has been submitted to Shanghai Disabled Persons' Federation, China Disabled Persons' Federation and some National People's Congress members. Now we are facing the problem of registration and fund-raising. We need our government's support and action. Let's join in efforts to make a better tomorrow!

Ms Changqing Zhu, MDA China



MDA: Japan

The Japan Muscular Dystrophy Association was established in 1964 by Shizuo Kawabata. His wife Shizuko Kawabata, has been the president ever since. She was later succeeded last spring by Mr Toshio Fukuzawa.

In our Association, there are more than 3,000 members including patients, their families, friends, and supporters. In establishing this Association, we have kept foremost in our minds and hearts the major objectives which included exchanging scientific information and promoting research aimed at discovering effective therapy for muscular dystrophy. At the present time, exchange of information has been done by members through personal communication, the internet, and the association. This exchange of information among members has encouraged patients to live their lives to the fullest and to stay updated on the progress of basic and clinical research in muscle diseases. In addition, members have the opportunity, at regional and national meetings, conferences and seminars organized by our Association, to meet and discuss common problems encountered daily and together find solutions on how to deal with these challenges. The biggest conference is held on the third Friday and Saturday of May every year, when many patients, parents and supporters including Government officers, medical doctors and teachers get together for information exchange and to discuss further progress in the Association.

The Association is managed by annual membership fee and a small amount by donation; therefore we cannot financially support the patients' medical care and

research projects but with our strong urging and support, the National Institute of Neuroscience, National Center of Neurology and Psychiatry (NCNP) was founded in 1976. Here, world class research is carried out to elucidate the pathomechanism of the muscular dystrophies and develop the most effective treatments for muscle diseases. The NCNP has organized 4 nationwide research groups for muscular dystrophies, including two clinically oriented groups and two basic science groups, focusing on the care of the patients in the former groups, and exploring pathogenetic mechanism of the disease and developing new therapeutic methods including gene therapies in the latter. All groups are supported by the Ministry of Health, Labor and Welfare, Japan. Our Association coordinates these research endeavors and works constantly to obtain further financial support for these research groups,

We maintain an active interest in WANDA. We organized the very successful WANDA meeting in Kyoto in 1994 just after the 8th International Congress of Neuromuscular Diseases. At the meeting more than 100 participants got together from various countries and enjoyed Professor Dubowitz' lecture. We had a dinner party and, of course, sightseeing. These activities set the stage for exchanging knowledge about muscular dystrophy and its associations in different countries and establishing great friendships.

Mr Toshio Fukuzawa President JMDA



ABDIM: Muscular Dystrophy Association of Brazil

Founded in 1981 by geneticist Mayana Zatz, one of Latin America's leading scientists, the Brazilian Muscular Dystrophy Association (ABDIM) is a nonprofit organisation that offers treatment to people with muscular dystrophy, helping to enhance their quality of life and prolong their life expectancy.

In more than 20 years of activity ABDIM has cared for some 15,000 people and helped to achieve significant progress in the treatment of muscular dystrophy. In late 2003 an agreement with the São Paulo State Department of Health made restructuring possible and provided sufficient funding to cover most of the association's costs.

The partnership has leveraged such significant improvements as an increase in the range of treatment offered, longer working hours, guaranteed supplies of BiPAP machines and free DNA tests for poor families with a history of neuromuscular disease.

Despite all these efforts, however, ABDIM is still unable to cover more than metropolitan São Paulo or only about 5% of the people in Brazil affected by muscular dystrophy. We try therefore to prioritise the provision of care to the poor.

ABDIM's Mission

ABDIM's mission is to be a national referral centre for the care of muscular dystrophy patients, providing means for rehabilitation, therapy, clinical case management, recreation and socialization. We also carries out clinical research and disseminates information about the disease and its treatment. Our overriding aim is to extend patients' life expectancy, enhance their quality of life and help them lead productive lives in society. To this end we promote training courses to enable physicians and health workers to identify and treat the disease.

Goals

ABDIM's top priority at present is to consolidate its position as a national referral center in Brazil for the provision of treatment and support to people with progressive muscular dystrophy. This entails combating one of the greatest obstacles to care and treatment of the disease, namely ignorance and the resulting rarity of early diagnosis. When therapy begins late, patients suffer unnecessarily. The following are fundamental to this process:

- training professionals throughout Brazil to identify and treat the disease, transfer knowledge, and exchange experience and information
- developing a program of mass information on the disease to educate doctors, teachers and the broader public
- creating a database with details of people with progressive muscular dystrophies to enable the public health system to optimise outpatient and home care.



Professor Mayana Zatz President ABDIM

Movitae: In Favour of Life

Founded in 2003 Movitae is a non government, non profit organisation (NPO), intended to support and promote society's discussion about stem cell research and technology.

Movitae is intended to inform, orient and mobilize Brazil's civilian, government and scientific communities, contributing in this way to stem cell research advances.

Furthermore, the organisation's work helps sufferers from many serious diseases to enhance their quality of life and feel better emotionally, whether or not stem cell technology will be potentially helpful to them.

Our goals are also: to promote access to information regarding various diseases and available treatments, alerting patients and their families to the risks associated with specific treatments without constraints due to potential ethical or philosophical biases.

Our organisation exchanges resources and ideas with associations, foundations, and research centers that share our principles.

We also created the Stem Cells Virtual Institute (<http://www.ivct.org>), where scientists will be able to share knowledge and theories about stem cells, and the layperson will find information about the technology.

Andréa Bezerra du Albuquerque, President, Movitae



Andréa de Albuquerque (top right) with patients and families in Brazil

Muscular Dystrophy Foundation of South Africa



In South Africa, the incidence of MD is approx. 1 in 1200 people mirroring our very diverse population. In a country of approx. 47 million, about 39 thousand have MD. As a country we are not deemed either highly indebted or poor, yet in reality the majority of South Africans are indeed deeply poor. It is a battle to reach these people. Most of them are in rural areas, villages. Some do not have access to doctors. Many remain undiagnosed.

We have one counselor in the Cape who goes into the countryside to meet families affected by MD. He takes students with. The benefits are astonishing. We are encouraging the other branches to follow this example. We have approached the Department of Social Development to assist in paying the salaries of social workers that we would like to employ. Even though the Government in general seems disinterested, we have received great support from the Department for Social Development and the regional Departments of Health.

Another challenge is crossing the language barrier. We have 11 official languages. Currently our literature, television advertisements, radio advertisements and posters are only in English. Translating information is difficult as many words and phrases that we take for granted are not found in other languages. Our magazine is in English. We have invited people to submit stories in other languages too, but we will need proof-readers who understand not only both languages involved, but also MD terminology.

We want to offer support to everyone who needs it from understanding the diagnosis to adapting lifestyles through information sharing. We have a number of nuclear groups who focus on a specific condition and general support groups who help with day-to-day living. Our branches have equipment libraries and are able to source specialized equipment if required.

Maxine Strydom works with Parent Project USA and shares information with South African families dealing with MD. She works with families with children with any MD.

We have relationships with medical professionals who provide updated fact sheets that we publish in our magazine and on our web-site. We encourage students, e.g. physiotherapists, to do projects on MD. Our hope is to develop a culture of interest at student level. Until recently much of our funding was aimed at research itself. In addition, it now covers projects such as diagnostics and social impact studies.

We must educate communities as many people believe that a child's disorder is the mother's fault. There are active campaigns by SAIDA (South African Inherited Disorders Association) aimed at changing this perception. Counselors / social workers in each region will be able to communicate in a way that changes opinions but does not disrespect culture. We want to gain people's trust and show them how we are able to assist.

To educate the population and to get the services out in the fields, we need funding and awareness. They go hand-in-hand. It is easy for us to reach those of higher economic standing. They have televisions, use the internet, belong to medical assistance programs, know how to find neurologists and how to find us.

Until recently our awareness has been on a regional level with only media projects like radio and television advertisements being at national level. Regionally, fundraising is done for operational costs and on a case-by-case basis to assist in purchasing equipment. This is done through approaching companies, trusts and individuals for donations, and organising activities such as golf-days. Nationally we have a marketing company that sells products on our behalf. These funds are for national operating costs, research, awareness and patient assistance projects.

The list of challenges is endless: diagnosis, schooling, transport, employment. We are doing our best to take on each of these. This is our reason for existence.

Sherry-Lynn McDougall, National Administrator MDF South Africa

Neuromuscular Disease Organisations in Australia

Developments in Neuromuscular Disease Organisations in South Australia and Elsewhere in Australia.

In Australia, MDAs are state-based organisations rather than there being a single national association. Despite many attempts over more than 30 years, individual state MDAs have resisted amalgamation although one of these, the MDA in the Australian state, Victoria, calls itself "Muscular Dystrophy Australia". Muscular Dystrophy Australia is a strong and well publicised organisation with an extensive web-site. It raises substantial funds annually and provides a wide range of services to its clients and considerable funding to medical and scientific research. However, its image is misleading in that it over-emphasises its own importance as being the association that represents all of Australia and fails to provide essential information about other Australian MDAs and similar organisations in Australia and internationally. Many of us, both in Australia and in our international NMD community, are attempting to resolve these anomalies but have not yet managed to do so.

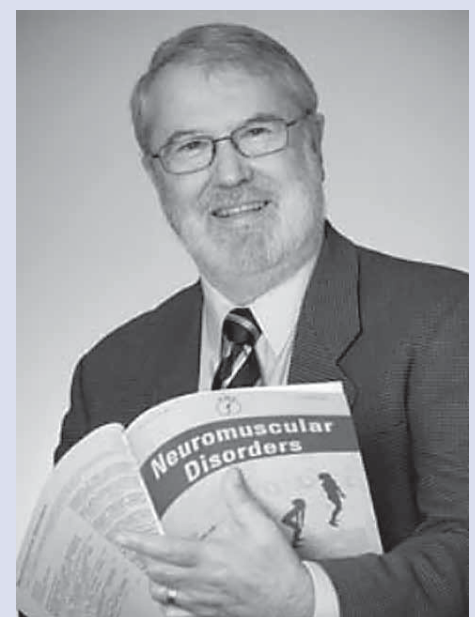
It is therefore necessary for me to report on the activities of each of the other state MDAs individually.

Firstly, MDA South Australia caters for members in both the state of South Australia and the Northern Territory. Together these cover an area equivalent to that of Western Europe but with a population of only 2 million (compared to Australia's total population of 20 million). Annually, this association raises about 1.5 million Australian Dollars (1 million Euros) via special fundraising events, donations, subscriptions and lotteries. Its patient/client-base is about 1,500 and its mission is to enable individuals, families and carers affected by neuromuscular diseases to live as full and active a life as possible. Activities of the association include services for clients such as provision of hydrotherapy, speech therapy, physiotherapy, counselling, social work, respite care and an annual camp. As well, the association supports participation of a team of children and young adults in the national electric wheelchair sports. These sports are held alternately in each of Australia's major capital cities. When possible, the association also supports scientific and medical research as well as personal development programs for its staff and volunteers.

The MDA of Queensland has substantially the same goals and activities but caters to a population of some 4 million and raises an average of about 2.5 million Australian Dollars annually. The state of New South Wales has a population approaching 7 million but its MDA is much smaller than those of South Australia and Queensland. Hence, it is able to provide few services and little or no research funding. Small Muscular Dystrophy Associations also exist in the state of Tasmania and in the Australian Capital Territory, again with similar goals and activities to those mentioned above. The Muscular Dystrophy Association of Western Australia, this state having a population of some 2 million, has had different goals and activities. It has been mainly a fundraising organisation for neuromuscular research and, over the years, has made huge contributions to the research of Prof Byron Kakulas and his colleagues. It is, more recently, also beginning to provide services.

In addition to these organisations, Australia is also served by a group of other neuromuscular disease organisations that are not affiliated with the Muscular Dystrophy Associations and, these too, are often state-based rather than national organisations. They include, Parent Project Australia, a national Spinal Muscular Atrophy Association, Friedreich's Ataxia Associations, a national Motor Neurone Disease Association and a number of state Motor Neurone Disease Associations.

*Professor Allan Bretag, Director of Research,
MDA South Australia*



Association Française contre les Myopathies (AFM) Report

The Association Française contre les Myopathies (AFM, the French Muscular Dystrophy Association) was founded in 1958 and since 1987 has run its Telethon, recently raising more than €100million per year. From a total budget of more than €120million it spends in excess of 50% on research and development, around 30% on services, with well under 10% being contributed to administrative costs and the remainder held in reserve for future fundraising. Many hundreds of research grants and research fellowships are provided annually to basic science and clinical researchers worldwide as well as to associations, institutions and industry. AFM's strategy is to understand diseases from the genetic level on up to the pathology, to identify treatment opportunities and then to proceed to clinical development of potential therapies. In this regard, AFM was able to establish its giant research institution, "Généthon", at Evry, on the outskirts of Paris in 1990. Its original mission was to sequence components of the human genome irrespective of whether those genomic regions were likely to contain genes associated with neuromuscular diseases. In this way, it made a major general contribution to the Human Genome Project while, nevertheless, implicating a number of genes as having an association with neuromuscular and other diseases. Généthon proved to be the catalyst for the establishment of a surrounding "city" of private biotechnology companies, "Genopole". While continuing the funding of research into basic genetics, biochemistry, physiology and pathology of nerve and muscle function and disease, more recently, AFM has been involved in the development of potential therapies. These include gene therapy utilising adeno-associated viral vectors, exon skipping, read through of stop codons and stem cell therapy (with both adult and embryonic stem cells). In this regard, AFM supports veterinary medicine and the testing of a number of different animal models of human diseases. As well, AFM is engaged in a unique partnership with the Institute of Myology and other organisations that contribute, together, to research, clinical consultations, clinical trials, collections for DNA and tissue banks, dissemination of information and networking. Therapeutic strategies under investigation range from gene replacement and modification, through induction of compensatory proteins and enzyme therapy, to adjustment of the tissue degradation/regeneration balance, orthopaedic surgery, ventilatory assistance, nutrition and genetic counselling. A considerable number of clinical trials in both animals and humans have been funded by AFM and new trials continue to be funded, e.g., trials related to the therapy of Duchenne muscular dystrophy, limb girdle muscular dystrophy type 2C and myotonic dystrophy. It is of importance that strategies developed for the therapy of myotonic dystrophy may be applicable to other severe diseases that are caused by similar genetic defects, including Friedreich's and various other ataxias and Huntington's disease. Ongoing clinical trials in immune deficiency diseases, haematological and skin diseases are also being supported by AFM. Pharmacological interventions are also being pursued, such as, vitamin C for Charcot-Marie-Tooth disease and idebenone for Friedreich's ataxia. Finally, AFM advocates for government reimbursement of costs for expensive protein therapies such as Myozyme for Pompe disease and monoclonal antibodies for a variety of diseases. Future challenges to success include safety and ethical issues in relation to gene and stem cell therapies, adequate patient numbers for clinical trials, sufficient time and enough funds for both the research and the eventual treatments. Bottlenecks will only be overcome with co-operation between patients, advocacy groups, researcher collaborative networks, government regulatory bodies and industry.

Dr Serge Braun, Scientific Director, AFM



Press Release for Telethon in Italy

Italy's annual telethon raised over EUR 30 million for research into finding a cure for Muscular Dystrophy and other genetic diseases.

The total amount raised during the TV marathon of over 60 hours, which took place from 15 to 17 December, is slightly higher than in 2005 and is expected to grow when all donations are counted.

Since its inception in 1991, the telethon has financed hundreds of research projects into genetic diseases, as well as specialised institutes and tissue and DNA banks. The money raised is also used to help the career development of scientists in the field.

Thanks to generous support from the Italian public over the last 17 years, researchers have successfully identified and characterised the genes and the mechanisms involved in several hereditary diseases and have made important clinical discoveries.

One major result was the finding of two genes responsible for Timothy syndrome and short QT syndrome type 3. These are two forms of cardiovascular diseases characterised by sudden death and ECG (electrocardiogram) abnormalities.

Other genes identified with Telethon funds include PINK1, which is responsible for a familial form of recessive Parkinson's disease; ETHE1 which, if defective, causes a devastating infantile metabolic disorder; and MYH1, a gene responsible for hereditary deafness.

Results have also led to the elucidation of the molecular mechanisms underlying two genetic diseases: the neuropathy Charcot-Marie-Tooth type 4B and a form of hereditary deafness depending upon deficit of connexin 26.

December is clearly the season for such initiatives. During its annual telethon on the 7th and 8th of December, France raised over €101.5 million for medical research into muscle-wasting diseases, while on 14 December, Germany raised over €5.4 million for research into leukaemia. The campaign was led for the 12th year running by José Carreras, a Spanish singer who won the battle with leukaemia at the height of his career in 1987. Since 1995, he has presented a yearly live televised benefit gala concert in Leipzig to raise funds for his foundation, the José Carreras International Leukaemia Foundation. Over €62 million have been raised by this event.

For more information, please visit:
www.telethon.it

5th International Assembly of WANDA Summary



This exciting afternoon-evening WANDA symposium was scheduled for the 5th July, 2006, during the 11th International Congress on Neuromuscular Diseases (ICNMD-XI) in Istanbul. It was held in the main Congress Hall allowing ICNMD-XI participants to attend as well as patients and families of affected persons. Entitled, "Roadmap to Treatment", the symposium logo showed three roads leading towards a common intersection, successful treatment of neuromuscular disorders (NMDs). Beside each road were groups of people, representing patients, scientific and medical researchers and the pharmaceutical industry, all intent on reaching this destination.

Prof Coşkun ÖZDEMİR, founder of the Turkish muscular dystrophy association, opened the symposium. He was followed with welcoming words from Prof Piraye SERDAROĞLU, President of WANDA and one of the two Presidents of ICNMD-XI. The opening was concluded by Ysbrand POORTMAN, who has a grown-up daughter with spinal muscular atrophy and is one of the most active promoters of National, European and International patient/parent self-help groups. As one of the driving forces behind the foundation of WANDA, he was well qualified to reiterate its main objectives:

- to organise meetings every four years,
- to encourage the formation of new NMD associations,
- to promote increased collaboration between NMD associations, and
- to facilitate planning for better care and services for people with NMDs.

He explained how hard it was to obtain the correct diagnosis for his young daughter in 1970 and reported on advances since then due to increasingly collaborative research programmes, greater numbers of known patients, increased governmental support, improved public awareness and an enormous boost in funding. Despite this last, he emphasised that successful NMD research requires still more money, a considerable amount of luck, and again more money.

First speaker of the session was Dr Serge BRAUN, research director of the French NMD organisation, Association Française contre les Myopathies (AFM), who told of the enormous sums raised by the AFM Telethon (averaging at least 100 million Euros each year). For 2006 their budget is 123 million Euros, over 50% going into research and development, some 30% into services and 10% towards future fundraising, with only 8% required to administer it. From this income, 450 new research projects can be funded each year. In addition 100 fellowships are granted, both for basic and clinical researchers at universities and in industry, in France, elsewhere in Europe and Worldwide. Furthermore, AFM has, for many years, operated its own research centre, Généthon, at Evry on the outskirts of Paris. Genetic research has been the major strategy of AFM since most neuromuscular disorders are caused by gene defects. This approach has brought many breakthroughs in diagnostics and is now expected to do the same for gene therapy. Dr BRAUN also modestly mentioned the huge contribution AFM made towards deciphering the human genome. During this undertaking, more than 150 genes were found that are associated with neuromuscular disorders. Most impressive in this respect was the discovery of a link between the SMN-1 gene and spinal muscular atrophy. Dr. BRAUN emphasised the many tasks remaining for the future and, similar to WANDA's Roadmap to Treatment, he spoke of AFM's slogan, "The Pathways to the Drug". He noted that AFM remains a patient/parent organisation and encouraged patients and parents of other NMD associations to:

- serve on the boards of research institutions,
- participate in clinical trials, and then,
- help to publicise the results as soon as possible.

AFM is an outstanding example of a patient/parent NMD organisation and Dr BRAUN certainly impressed on the audience just how strong "the power of patients/parents" can be.

5th International Assembly of WANDA Summary

The following speaker was Peter STRENG, a member of the board of the European Neuromuscular Centre (ENMC), a research and information organisation founded and financed by several of the largest European Muscular Dystrophy Associations. He announced that between 1990 and 2005, ENMC had organised (and financed) 138 international NMD research workshops. One of the results was the establishment of 18 groups of experts on different NMDs who established definitive diagnostic criteria for the different conditions. The work of ENMC has resulted in some 225 publications, including scientific and lay reports, pamphlets for patient groups and booklets for the scientific and medical professions.

Next, Prof Katie BUSHBY revealed an extraordinary success. The European Union had just decided to provide 10 million Euros over five years to create a "Network of Excellence" among European NMD Centres. The project application, known as "TREAT-NMD", had been submitted by Profs Volker STRAUB and Katie BUSHBY of Newcastle-upon-Tyne University with professional support from ACIES (a French research and development consultancy company), AFM and ENMC. In essence, it provides another excellent illustration of WANDA's "Roadmap to Treatment" involving 21 partners that include national networks, research and clinical organisations, charities and industry. It will include eight study sections, each directed by a leader:

- The Swiss pharmaceutical company Santhera will lead the section Animal and Cell Studies, defining appropriate model systems, testing novel treatments and setting standard parameters for assessing "therapeutic effects".
- AFM will lead the section Production and Toxicology, identifying factors that hold up production and toxicity testing of potential therapeutic agents so that time to clinical use can be minimised.
- The University of Leiden will lead the section Targeting to Muscle, determining ways to ensure that therapeutic agents actually reach muscle.
- The German Muscular Dystrophy Network (MD-NET) will lead the section Databases, Registries and Bio-banks, identifying existing resources, allowing access to relevant material for translational (laboratory to clinic) studies and identifying the patient groups available for trials.
- The University of Stockholm will lead the section Standardisation of Diagnosis and Care, developing "TREAT-NMD" guidelines on diagnosis of NMD and on baseline standards of care to which participating centres will contribute and adhere so as to allow common starting points from which to assess any benefits in future clinical trials.
- Telethon will lead the section Outcome Measures, assembling and evaluating all the available tests for positive effects in the treatment of NMDs thereby making for ease of assessment and ready comparability between different studies.
- MD-NET will also lead the section Co-ordination Clinical Trials, assembling a group of experts on the application of trials in NMD, establishing European Union clinical trials regulations and providing support for trial development.
- ENMC will lead the section The Ethical and Patient Environment, aligning information and approaches within national and disease-specific patient groups, communicating uniform information and addressing ethical concerns of patients/parents in relation to trial development.

While not comparable with the annual budget of AFM (above), this new recognition of NMDs by the European Government should not be underestimated and might be able to be used as leverage to encourage other governments to support similar NMD networking.

In addition to such major achievements by large and multinational MND organisations, individuals and families frequently campaign for their own specific diseases. A Duchenne mother, Elizabeth VROOM, from the Dutch Parent Project, presented her experiences. She also discussed the role of the umbrella organisation, United Parent Project Muscular Dystrophy (UPPMD), in providing publicity, funding research, lobbying governments, forming networks and encouraging participation in clinical trials (while at the same time discouraging people from entering unsanctioned and uncontrolled trials). She noted that an initial charitable investment was a worthwhile way to encourage venture capital into a pharmaceutical company that had a promising product.

5th International Assembly of WANDA Summary



With few exceptions, only small pharmaceutical companies take the challenging road towards therapies for rare NMDs. Dr Thomas MEIER, from one of these, Santhera, reported on current studies with Idebenone, a drug that might prove useful in treating both Friedreich's ataxia and Duchenne MD. His presentation was subtitled "collaboration between industry and patient organisations" and he acknowledged the support of both UPPMD and the German patient/parent organisation, "aktion benni & co e.V." Genzyme, unusual among the bigger pharmaceutical companies, has already achieved success with one rare NMD, in this case Pompe's disease. On behalf of Genzyme, a young Pompe patient, Maryze SCHOENVELD VAN DER LINDE, gave a dramatic account of the development of this therapy, leading to her eventual treatment, from her point of view as an affected person. By 1996 the small company Pharming was able to produce the enzyme missing in Pompe's disease using gene-manipulated rabbits. Pharming gave up the project, however, and it was taken over for further development by Genzyme, using transgenic cell cultures. Hans SCHIKAN, Vice-CEO of Genzyme, then explained how the final product Myozyme was brought to readiness for marketing, with industry, patient organisations, the University of Rotterdam and the Government of the Netherlands all playing essential parts. Finding enough affected children from the farthest corners of the globe, necessary for the successful completion of a clinical trial, induced that mixture of anxiety and excitement we all associate with an action-adventure story. Pompe's disease has, finally, become the ray of hope for all researchers, clinicians, parents, patients and representatives of industry who are still battling their way along the road towards therapy for other NMDs.

After a break for refreshments, the evening session proceeded with impressive presentations by representatives of a number of patient/parent organisations from around the world. Some of them already look back on a long and successful history, like the Muscular Dystrophy Association of South Australia that organised ICNMD-IX and the 3rd International Assembly of WANDA in Adelaide, Australia, in 1998. Others, like South Africa, are gathering strength, while some, like China, are just beginning. One of WANDA's major roles is to assist these organisations in their endeavours.

In Istanbul, it was decided that the 6th International Assembly of WANDA will be held in Naples/Italy, in 2010, in association with ICNMD-XII. It is to be organised by Prof Giovanni Nigro, one of those muscle disease experts who sparked the foundation of WANDA in 1990.

*Reinhardt Rüdell, Ulm
Allan Bretag, Adelaide*

WANDA

prospectus
2009 - 2012

WANDA: A Brief History

Additional achievements of WANDA include:

- Production and dissemination of an information kit designed to assist people to form MDAs in countries where none exists.
- Distribution of a regular News Bulletin.
- Forming cooperative alliances with such organisations as the World Health Organisation, the World Federation of Neurology, the World Muscle Society, European Neuromuscular Centre, European Alliance of Muscular Dystrophy Associations, Mediterranean Society of Myology etc.
- Attracting valuable and generous sponsorship from Cephalon, Pfizer Europe, GCA, Ministry of Health, Welfare and Labour – Japan, Muscular Dystrophy Association – Japan, Genzyme Corporation, Muscular Dystrophy Association South Australia.
- Uniting over 70 national Muscular Dystrophy Associations in the fight against neuromuscular disorders.
- Bringing the human face of neuromuscular disorders to the scientific world.

- To make effective representation, when necessary, to individual governments to ensure they are aware of the needs of their citizens with neuromuscular disorders and their families.
- To encourage the formation of neuromuscular disorder associations in countries without existing bodies, by providing information and advice.
- To develop strategies for better care and services for people with neuromuscular disorders and advice and support for their families.

WANDA Presidents:

1990 – 1994	Prof R Rüdél – Germany
1994 – 2002	Mr A Esworthy – Australia
2002 – 2007	Prof P Serdaroglu – Turkey
2007 –	Prof A Bretag – Australia

*Andy Esworthy,
Past President WANDA*



An early meeting of WANDA representatives in Naples

WANDA's Aims:

- To encourage and facilitate cooperation between national associations, in order to help people with neuromuscular disorders and work towards a cure and eradication of the disorders.
- To represent the interests of people who have neuromuscular disorders and their families in every nation without consideration of race, religion or political belief, so that people with the disorders are accorded care and dignity.
- To facilitate swift dissemination of information on treatment and research findings which may be of use to neuromuscular disorder associations around the world.
- To be accepted by the World Health Organisation as the appropriate authority to be consulted, and to make suggestions, on issues affecting people who have neuromuscular disorders and their families, researchers and service providers.

WANDA: Progress Report

WANDA seeks a world in which neuromuscular disorders are well understood, eradicated and the people affected are well supported.

The WANDA board consists of Piraye Serdaroglu, President and Vice Presidents Hisanobu Kaiya, Allan Bretag, Alex MacKenzie, Lee Leith, Ysbrand Poortman, Mayana Zatz and Ekrim Abd el Salam. Our websites can be found at: www.worldmuscleforum.org and www.wandaweb.org

Objective: To facilitate cooperation between national organisations.

Between 2002 and 2006, meetings were held in Vancouver, Kusadasi, Rio with the most recent in Istanbul. The next quadrennial meeting is in 2010 in Naples. At present our database contains over 130 neuromuscular disorder associations with new associations added periodically. We also have connections with international organisations such as the ENMC, the IGA (International Genetic Alliance patient/parent associations), the IAPO (International Alliance of patient organisations) and the WAO (World Alliance prevention and treatment). Our members have attended conferences such as the International Congress of Neuromuscular Diseases and the BioVision Global life sciences forum (Lyon). We played a part in founding the International Rare Disorders Forum and the International Forum of Good Clinical Practice. In addition we have a role in supporting disease specific groups such as the International Pompe Association.

Objective: to develop strategies for better care and services for patients and families.

The WANDA project was adopted in Vancouver in 2002. The "Roadmap to Treatment" refers to our philosophy of streamlining the pathways to treatment so that patients are identified, their current needs are understood, that they have access to treatments to improve their quality of life and that research continues so that they may have access to curative therapy when it becomes available. In addition the "Roadmap to Treatment" involves the sharing of information between patients, scientists and industry on the one hand so that scientists can collect data about the patient's experiences and also patients can find out more about the latest research findings.

Objective: encourage the formation of new NMD associations via websites and internet information.

Recently new NMD associations have started in Algeria, China, Nepal, Lebanon, various African and South American countries, former USSR countries, gulf states, Morocco.

Objective: to facilitate swift dissemination of information on treatment and research findings.

Neuromuscular disorder associations contained in the WANDA database can receive information about new research developments. We can also refer patients with questions to other websites with news sections such as the ENMC website. In this way patients can find information whether it be for effective treatments or to find out about treatments proven ineffective such as myoblast transfer.

The Future of WANDA

The plan for 2006-2010 is to focus on information sharing and communication within and between associations. We will achieve this by improving our website and database to include more neuromuscular disease associations and establish two-way communication with other associations with related goals such as the ENMC, the International Genetic Alliance patient/parent organisation, the World Health Organisation and UNICEF. In future we need to maintain our goal of raising public awareness about neuromuscular diseases so that our recent successes in fundraising continue.

Ysbrand Poortman Vice President WANDA



Roadmap to Treatment of Neuromuscular Diseases

(Based on the model for the successful treatment of Pompe disease)

Mission Statement

To ensure that patients with neuromuscular diseases are diagnosed early and to promote the development of effective, affordable and safe treatments for them

To ensure that reliable information and support are available for all patients, their families and other persons who are involved with neuromuscular diseases

Aim to build networks of cooperation between

Patient/Parent Organisations-Scientists/Research Institutions-Relevant Industries (Pharmaceutical, Biotechnology, Bioengineering, Nanotechnology)-Government Agencies-Philanthropists and Philanthropic Organisations-Human Genome Variation Societies-World Health Organisation, etc.

Process to build relationships between Patient/Parent organisations and their prospective industry and other partners

Develop databases of industries, research institutions and organisations with interests in neuromuscular diseases, open dialog and then establish formal communication and collaboration plans with them

Challenges for all parties concerned

Multiple companies and research institutions are involved in what may turn out to be relevant research and development

Patients come from many different countries with widely divergent degrees of development, cultures with unique ethical issues and very different governmental policies and perceptions of patient empowerment.

Realistic patient expectations must be established and maintained
There are frequently unfortunate delays between the discovery of promising therapies and the start of clinical trials which may be due to:

- Governmental regulations
- Low numbers of identified patients with orphan diseases
- Ethical issues and individual privacy considerations

Provision of accurate information about clinical trial policies and procedures is essential for trial participants, their families and carers and their assessors

Reimbursement is required for participation in trials so that patients, their families, their carers and their doctors are not disadvantaged by the clinical procedures, travel costs, time off work, etc.

Meeting the cost of new, expensive therapies is a major challenge for patients, families, health insurance providers and governments although some industry partners must be commended for providing therapeutic drugs at minimal or no cost in certain circumstances

Shared value of patient/parent organisations, industry, physicians and researchers

Accelerating the development of safe and effective treatments so that time to clinical employment of therapies can be minimised
Developing nationally and internationally acceptable clinical trial regulations through multi-centre collaboration and the assistance of the highest possible levels of governmental agencies and non-governmental organizations
Maintaining consistency of information relating to clinical trials and addressing ethical concerns of patients/parents as they relate to clinical trials
Establishing consistent diagnostic criteria and baseline standards of care so that multicentre clinical trials can be validly compared
Setting standard parameters for assessing therapeutic effects in both patients and laboratory model systems
Improving patient care by every means possible, including increasing access to treatments, services and information, as well as, fostering the development of sustainable healthcare infrastructure
Advocating for policies, benefits, funding and laws to best meet the needs of patients and their families
Raising disease awareness worldwide through appropriate levels of mass education, multi-media advertising, utilization of the internet via specific web pages and links from whatever related sites might be available; ensuring that relevant factual information and links to patient/parent organizations, sympathetic industry partners and relevant research institutions are available to be accessed within web-based resources, such as Wikipedia.

Roadmap to Treatment of Neuromuscular Diseases

Plan for exchange of information between the three groups and with the general public

Information gathering:

- From patients
 - surveys
 - establish a patient data bank
- From physicians/researchers
 - forum
 - links to websites
- From industry
 - need to establish the most effective method

Information dissemination:

- Through WANDA website
- Hard copy pamphlets
- Media releases

Information exchange: Aim is to establish links on the WANDA website with neuromuscular disease organisations, research centres and interested industries.

A coalition already exists between French muscular dystrophy association (AFM), the American muscular dystrophy association (MDA), Parent Project Muscular Dystrophy (PPMD) and United Parent Project Muscular Dystrophy (UPPMD). WANDA plans to seek inclusion in this group and will encourage inclusion of other neuromuscular disease organisations.

Examples of International Pompe Society successes using this model

Despite the disease being rare, with low patient numbers in any individual country, sufficient participants were recruited internationally for clinical trials and related insurance problems were solved leading to faster enrolment. Trial participants were encouraged and informed via the "Flying with Pompe" brochure; successful information exchange led to provision of up-to-date information to all concerned.

Prospective participants were surveyed, the results were collated and published providing useful information for clinical trial design. In these ways, trials were facilitated, quickly leading to the development of a new drug and the establishment of an optimum dose.

Basic Principles of Collaboration and Cooperation

Be pro active

- Espouse high ethical standards
- Identify possible obstacles
- Collaborate together to achieve outstanding mutually beneficial results
- Gain senior level support of collaboration
- Establish relationships early
- Respect, consider and respond to mutual perspectives and insight
- Coordinate and maintain consistency of contacts
- Communicate openly, honestly and transparently
- Follow through
- Promote evidence-based medicine
- Maintain high standards of research

Drug Development for Pompe Disease

A collaborative effort:

Challenges, achievements and future steps

Improving the lives of patients is Genzyme's top priority. Genzyme works collaboratively with multiple stakeholders including patient organisations, health care providers, regulatory bodies and reimbursement authorities, to achieve optimal care for patients with genetic disease and sustainable healthcare systems.

Pompe disease is a debilitating and often life-threatening neuromuscular disease, characterised by progressive degeneration of skeletal, respiratory, and cardiac muscle. Patients with this rare genetic disease have a deficiency of the enzyme acid alpha-glucosidase (GAA) that leads to progressive accumulation of lysosomal glycogen. It is a spectrum of disease, some patients present in early infancy with rapidly progressive cardiomyopathy, muscle weakness, and die of cardiorespiratory failure before age 1. Others present later in childhood or adulthood with progressive muscle weakness that leads to wheelchair and respirator dependency and premature death.

Genzyme's commitment:

Through innovation and partnership, the development and approval of a therapy for Pompe disease that would be accessible to Pompe patients in medical need.

Drug development pathway:

The drug development pathway for orphan genetic diseases is a complex one. It begins with the discovery of the pathogenesis of disease and gene localization, followed by the development of a candidate drug or treatment. Pre-clinical and subsequent clinical trials need to be conducted. The next steps involve the scale up of the production of the successful drug or treatment, the global regulatory filing and review process. If successful, this results with the marketing approval and if appropriate, the definition of post-approval commitments. The final phase includes lobbying for national reimbursement and therapy access, and the long term management of patients. With so many steps involved, the path from early research to a widely available treatment can be a long and expensive one. It takes more than 8 years, on average for a new biotech product to go through clinical development and regulatory review. As an example, the development of

Myozyme® for Pompe disease cost \$500M, not including post authorization costs and cost of failures.

Key challenges in such a process for a rare disease, could include the identification and location of suitable patients for a clinical trial; making decisions regarding the trial design, duration and clinically meaningful endpoints; the relocation of patients to clinical trial centres etc. Gaining marketing approval to commercialise a therapy once clinical trials are completed is an extensive process. All of the clinical trial data must be filed with the appropriate regulatory authorities around the world eg EMEA or USA FDA. On average, there is a review time of 12-14 months. Some authorities may require expert panel discussions and post approval commitments such as continued collection of data on the disease and on the effects of the new therapy.

Prior to approval, there are a number of ways a patient may have access to therapy. For instance participation in clinical trials, or in the case of Pompe disease where some patients could expect a rapid progression to irreversible organ damage and death without treatment, the ethical and medical case was made for pre-approval access to therapy, while bearing in mind the limited supply of such therapy.

Approval and beyond

In the spring of 2006, both the European and US regulatory authorities (EMEA and FDA) approved the application of alglucosidase alfa (Myozyme®) for long-term enzyme replacement therapy in patients with a confirmed diagnosis of Pompe disease. This was a major milestone in the search of a therapy for this so far untreatable disorder and Genzyme continues with the regulatory filing process for treatment approval and access internationally. Currently, hundreds of Pompe patients around the world are receiving Myozyme®, the majority of which are older children and adults. As the experience in treating Pompe patients is growing, so too will the understanding of the disease.

As is the case with other rare diseases, symptoms are often similar to many other conditions, so valuable time is often lost from the onset of symptoms until an accurate diagnosis is made. The availability of a treatment for Pompe disease, heightens the need for better disease recognition and prompt diagnosis so that treatment can

Drug Development for Pompe Disease

begin before irreversible muscle damage occurs. This can be achieved with Pompe disease awareness, training and educational programs, not only for health care professionals involved in the diagnosis and management of Pompe patients, but also for the general public. In addition to awareness campaigns, Genzyme will maintain a presence at key scientific conferences, in Europe and around the world, in order to network with other stakeholders. The Pompe Registry, is a global, observational, internet-based programme, established to increase the understanding of the natural course of Pompe disease and to monitor patient outcomes. An additional goal of the Pompe Registry is also to extend the knowledge of long-term therapy (safety & efficacy).

The patient organisation's crucial role in the drug development pathway

Patient organisations often partner with industry to reach their shared mission to improve patient care, raise disease awareness; accelerate the development of safe and effective treatments; increase access to treatments, services and information; and foster the development of a sustainable healthcare infrastructure.

As an example, the International Pompe Association (IPA), was closely involved in the development of a therapy for Pompe disease. This included their involvement in research to document the natural history of the disease, recruitment of patients for clinical trials, review of clinical protocols and to share the patient perspective at regulatory panel meetings. Genzyme and IPA continue to maintain this mutually beneficial collaboration.

Conclusion

There are many challenges and potential obstacles along the pathway from the identification and approval of an effective new treatment, to providing access to patients. The process is a resource-intensive, and time-consuming endeavor. To achieve the shared goals of patient associations, health care professionals, regulatory authorities and industry, close partnerships and effective communication are vital.

*Annamarie Dillon
Myozyme® Global Marketing, Genzyme*

Patient/Parent Neuromuscular Disease Organisation Membership

Extend WANDA Membership widely to new organisations – especially in developing countries and for single disease groups, update database as appropriate and develop automated, web-based application to join WANDA and web-based update of member information.

Industry Membership

Extend database to relevant pharmaceutical and biotechnology industry.

Institutional Membership

Extend database to relevant research and educational institutions (universities and pure research establishments) and include benevolent foundations, institutions and individuals.

Enhance Communication

Improve WANDA or joint WANDA/WCAF website/s with respect to information content, two way communication, etc. Exchange information via networks, newsletters, pamphlets, handbooks.

New organisations

Encourage the formation of new neuromuscular disorders organisations where none currently exist. Facilitate co-operation, amalgamation and collegiality as appropriate.

Patient Disease/Genetic databases

Encourage the establishment of voluntary patient disease/genetic databases by local organisations. Encourage member organisations to provide information about international patient/genetic databases and to encourage their patients to volunteer disease/genetic information to those international databases.

Biobanks

Provide information to member organisations about the existence of relevant biobanks and encourage the voluntary submission of appropriate biological samples (DNA, blood, muscle/nerve/brain tissue) to those repositories.

WANDA International Assembly, Naples 2010

Inform members about the Naples meeting of WANDA members/ patients/ parents/ carers to be held in conjunction with the 12th International Congress on Neuromuscular Diseases, July 17 – 22, 2010.

Existing societies

Promote existing societies and groups such as ENMC, AOMC, EAMDA, etc., to WANDA members and encourage the establishment of similar new groupings elsewhere.

Patient/ Parent/ Carer Involvement

Encourage the active participation of patients/ parents/ carers in their local organisations, regional groupings and international affairs. Become involved in fundraising, liaising, lobbying and driving research, education, etc.

Forum of Senators

Establish a Forum of Senators, senior experts who might be willing to provide expertise and opinion on matters of importance (medical, scientific, fundraising, organisational, administrative, etc.) to member organisations and individuals.

Accurate Reporting

Encourage accuracy, high ethical standards and honesty in reporting on research developments. Eliminate the hype and the spin but maintain the hope.

Brief Business Plan 2009-2010

To achieve its goals WANDA will search for capacity, build infrastructure and collaborate with all stakeholders.

Financial resources are expected to come from fundraising, from funded projects, from some specific charitable foundations and from major NMD associations.

The quadrennial assembly in Naples in 2010 will be a milestone and focus point for awareness, communication and promotion. From it the 2011-2012 Business Plan is expected to be developed.

	2009	2010
NET INCOME*	€18500	€50000
TOTAL EXPENDITURE		
Secretariat	€2000	€5500
Prospectus preparation and distribution	€7500	
Website reconstruction and maintenance	€2500	€1000
Preparation for Naples Assembly	€6500	€23500
Promotion, publications, reports		€7500
Various projects		€12500
ESTIMATED SURPLUS /(DEFICIT)	€0	€0

*Additional Information: Various contributions have been donated to WANDA in kind so that this financial survey does not reflect WANDA's activity level

Invitation to Naples, 2010.

The 6th International Assembly of the World Alliance of Neuromuscular Disorder Associations (WANDA) will be held in Naples, Italy, in conjunction with the 12th International Congress on Neuromuscular Diseases (ICNMD) between 17th and 22nd July, 2010.

As President of the 12th ICNMD, I warmly invite WANDA members, delegates and other interested participants to Naples to attend this quadrennial assembly. As in the past, it is set to inform patients, parents, carers and administrators of Neuromuscular Disease Associations about recent research developments in lay language and to allow communication and cooperation between local, national and international individuals and groups.

Naples is a modern city of around 4 million people with an ancient (founded 2,800 years ago) and historical, World Heritage Listed, heart set on a magnificent bay with Mount Vesuvius as a backdrop. It is rich in art, culture and gastronomy, the last, especially, for having invented the pizza. We can assure that you will receive the most cordial welcome to our city and are certain that your visit will be a most enjoyable one.



The Skyline of Naples looking across the Bay of Naples to Mount Vesuvius in the distance.

We look forward to meeting you here in 2010.

Sincerely,
Giovanni Nigro
President 12th ICNMD
Second University of Naples
Naples, Italy

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