



Rare Disease Day

**Focus on Rare Diseases in Ireland;  
What is the National Plan?**

European Public Information Centre,  
European Union House, 18 Dawson St, Dublin 2

Wednesday 25th February 2009

9.30am **Opening: A Patient's Perspective**  
Ms Doris Noe, Pompe Patient

**Session 1**

- 9.45am – 9.50am **Welcome**  
Chair: Ms Eibhlin Mulroe, Chief Executive, IPPOSI
- 9.50am – 10.05am **Accessing Treatment and the Experience of Rare Disease Patients in the UK**  
Mr Eric Low, Chief Executive, Myeloma UK
- 10.05am – 10.20am **Department of Health and Children and its role in developing a plan for Rare Diseases**  
Dr John Devlin, Deputy Chief Medical Officer, Department of Health & Children
- 10.20am – 10.40am **Plan for Rare Diseases in Europe**  
Ms Christel Nourissier, General Secretary, European Rare Diseases Organisation (EURORDIS)
- 10.40am – 10.55am **What can Ireland learn from other EU countries? – an industry perspective**  
Mr Brendan Martin, UK & Ireland Business Unit Director, Genzyme Corporation
- 10.55am – 11.10am **Discussion**
- 11.10am – 11.25am **Coffee**

**Session 2**

- 11.25am – 11.30am Chair: Ms Avril Daly, Chairperson, GRDO
- 11.30am – 11.45am **A Case Study: The Importance of Research into Rare Diseases in Ireland**  
Dr Patrick Harrison, University College Cork (UCC)
- 11.45am – 12.00pm **What can Ireland learn from other EU countries? – a patient perspective**  
Mr Larry Warren, Chief Executive, Alpha1
- 12.00pm – 12.15pm **A Case Study: Diagnosis is difficult and for many Rare Diseases there is no cure**  
Dr Aisling Ryan, Consultant Neurologist, Cork University Hospital
- 12.15pm – 12.30pm **Economics of Orphan Medicines**  
Mr Paolo Morgese, Market Access Manager, Health Policy & Market Access – Europe, Merck Serono SA
- 12.30pm – 12.50pm **Discussion**
- 12.50pm – 1.00pm **Closing Comments**  
Mr John McCormack, Chairperson, MRCCG
- 1.00pm **Lunch**

## Speakers' Biographies



### Ms Doris Noe

Originally from Germany, my passion for nature conservation brought me to Northern Ireland in 1985. I live with my husband and two children in County Down. My stamina has reduced and my mobility has been compromised since my 20s due to Pompe Disease; a lysosomal storage disorder that I suffer from. My hero in life is my brother Peter who experienced an earlier and more severe progression of Pompe.

I like his philosophy of embracing this condition as an integral, important part of his life. He died of Pompe in 2005 before Enzyme Replacement Therapy was approved whereas I have benefited from this new treatment for over a year now. Patient groups played a vital part in moving along the research and development of this drug.



### Ms Eibhlin Mulroe, Chief Executive, IPPOSI

Ms Eibhlin Mulroe has a BSSC Hons in Politics from Queens University Belfast and is an MBA graduate from the Smurfit Business School, University College Dublin. She spent the first half of her career to date working in Irish Politics and then the NGO sector where she worked as CEO of the Asthma Society of Ireland. Ms Mulroe moved to the private sector for a time and served as an Account Manager for MedMedia on the campaign side of the business, managing campaigns for major pharmaceutical and healthcare companies in Ireland. In 2007, she became the first CEO of the Irish Platform for Patients' Organisations, Science and Industry (IPPOSI).

### Mr Eric Low, Chief Executive, Myeloma UK

Eric established Myeloma UK twelve years ago after volunteering in the United States for the International Myeloma Foundation and realising there was a need for an organisation to provide information and support to people affected by myeloma in the UK.

Since then Myeloma UK has grown and flourished due to Eric's limitless commitment and enthusiasm. It now provides a broad and innovative range of services, covering every aspect of myeloma, from information and support, to improving standards of treatment and care through education, research, campaigning and awareness raising. The organisation reaches over 12,000 people affected by myeloma and is staffed by a small but effective team, managed by a Board of Directors and guided by Expert Advisory Boards.

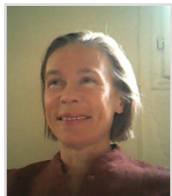
As Chief Executive, Eric has been instrumental in developing the organisation's 'bench to bedside' strategy, which stands as an optimal integrated model and is the only one of its kind in the UK. Amongst a very busy portfolio of projects Eric's principle focus at the moment is in establishing a national early phase trial network and a myeloma genetics and drug development research programme.

Eric also spends much of his time out and about meeting with representatives from the myeloma community, tirelessly advocating and campaigning on behalf of myeloma patients for the very best standards of treatment and care. Through his passion and enthusiasm he has significantly raised the profile of myeloma in the medical, political and media arena.

### Ms Christel Nourissier, General Secretary, EURORDIS

Christel faced a 16 year wait before her daughter's condition was diagnosed as there was no professional knowledge about, or understanding of her symptoms and behavioural problems. There was also a lack of recognition of her special needs in school. There was no patient group for Amélie's condition until 1997. Today her strongest wish for the future is that no family living with a rare disease in Europe would have to face such issues.

Christel Nourissier was elected Director of EURORDIS, Rare Diseases Europe, in 2000, representing the newly established Alliance Maladies Rares in France. She became Secretary General in 2003. Since 2000, she has been involved in all Eurordis European projects, in the organisation of the European Conferences on rare diseases Paris 2003, and Luxembourg 2005 with the support of the European Commission. She has been actively associated to the growth of Eurordis, particularly in the new Member States, and represents people living with rare diseases in many European institutions and platforms. She is also advocating for patients' rights and for the rights of those with disabilities.



### Mr Brendan Martin, UK & Ireland Business Unit Director, Genzyme Corporation

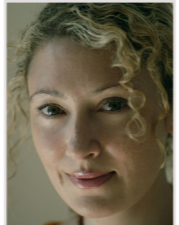
Brendan Martin is UK & Ireland Business Unit Director with Genzyme and a founding member and Treasurer of IPPOSI. He oversees the commercial management of treatments for ultra rare genetic disorders. In 2002 he joined Genzyme in Ireland to establish access to these treatments for Irish patients, and took up his current position in Oxford UK in 2007. Prior to Genzyme, Brendan worked in Australia with Boehringer Mannheim as Marketing Manager for Diabetes Care and Point of Care Diagnostics.

In his earlier career he trained in Analytical Science in DCU and having emigrated to Australia, worked in endocrinology with particular focus on treating human infertility. Throughout he has always engaged actively with patient groups and sought to promote understanding and collaboration between patient associations, scientists and industry.

### Ms Avril Daly, Chairperson, GRDO

Avril has worked for the Irish charity Fighting Blindness for over eight years. As Head of Public Affairs she is responsible for raising awareness of retinal degenerative diseases among the general public, health care professionals and policy makers. She also works on the promotion and development of the charity's significant research portfolio. Avril is Chairperson of the Genetic and Rare Disorders Organisation (GRDO), the Irish umbrella group supporting Irish families affected by genetic and rare disorders. She works closely with the European Rare Disease Organisation (EURORDIS) and is currently an advisor to EUROPLAN, an EU Commission initiative to establish national plans for Rare Diseases throughout the EU's Member States.

Avril is a board member of the European Platform for Patients' Organisations, Science and Industry (EPPOSI) and is a member of NanoMed an EU round table on the future of Nanotechnology in healthcare. She is also working on EuroGenGuide which aims to provide information about genetic testing, counselling and research to the public and health professionals across Europe. Through her work with Fighting Blindness she is a member of the Medical Research Charities Group (MRCG), the Irish Platform for Patients' Organisations, Science and Industry (IPPOSI), and Retina International. Before working for Fighting Blindness Avril held positions in publishing, event management and public relations.



### Dr Patrick T. Harrison, University College Cork

Mutations in the human genome can cause a wide range of health disorders and predispose us to a variety of diseases. The long term goal of Dr Harrison's research is to develop a generic approach to repair mutations in the human genome. Conventional approaches to genome modification have a success rate of only 1 in a million cells; whilst useful in research, it is of little therapeutic value. Dr Harrison's lab is using a novel class of synthetic proteins known as ZFNs which can repair mutations in as many as 1 in 5 cells in a single treatment.

Dr Harrison's group has successfully developed ZFNs that can target the gene defect in many cystic fibrosis patients, and is now designing ZFNs to target the genetic mutations that cause cystinosis, a rare genetic multisystem disorder that invariably leads to kidney failure. Once gene repair has been demonstrated in isolated cells in culture, the next step is to determine the feasibility of delivering the gene repair system using virus vectors. This work will be carried out in collaboration with Dr Martina Scallan in the Microbiology Department, UCC, using facilities they established for the production of recombinant virus vectors, which have been used in the study of models of Parkinson's disease and certain types of cancer.

Dr Harrison is a Senior Lecturer in the Department of Physiology and BioSciences Institute, UCC, and national contact for the newly formed European Association for Predictive, Preventive & Personalised Medicine ([www.epmanet.eu](http://www.epmanet.eu)). His research is supported by Cystinosis Ireland, the Health Research Board and Science Foundation Ireland.

#### Recent Publications

Harrison PT (2008). Application of Gene Therapy in Diabetes Care. *Infect Disord Drug Targets*. 8:129-33.

Lee, CM, Flynn, R, Harrison, PT (2008). Towards in vivo correction of the cfr  $\Delta F508$  allele: design and synthesis of two zinc finger nucleases for homology-directed repair. *Proc Physiol Soc* 11:C100.

Collins SA, Guinn BA, Harrison PT, Scallan MF, O'Sullivan GC, Tangney M. (2008). Viral vectors in cancer immunotherapy: which vector for which strategy?. *Curr Gene Ther*. 8:66-78.



### Mr Larry Warren, CEO, Alpha1 Foundation & President Alfa Europe

Mr Larry Warren is a native of Dalkey, Co. Dublin. He is married to Ann. He initially worked in the field of education -1967 to 1990. He was a school Principal for twelve years. In 1990 he entered the Health/Disability sector and worked in a managerial capacity in that sector.

1995 to 2001 he worked as CEO with the Cystic Fibrosis Association. He set up a complete dynamic database/registry of CF for Ireland.

Since April 2001 he has worked as CEO with the Alpha One Foundation where he is driving a Targeted Detection Programme. The establishment of a Lung Transplant Unit was as a direct result of patient advocacy. He works closely with voluntary and statutory bodies as an advocate. He has organised various medical conferences with presenters from Ireland, UK, Europe and the USA. He negotiates with the Department of Health and Children and with other health agencies on relevant matters. In 2004, together with Prof Gerry McElvaney, he set up the Alpha One Suite at Beaumont Hospital, Dublin for research and patient care. He established a Registry of Alpha – 1 Patients at the Suite. Larry also acts as consultant to the European Medicines Agency (EMA) on therapy studies and medicine development for Alpha-1 patients. He has a special interest in Rare Diseases and Organ Transplant and works with EU agencies on these issues also.

Larry is President of Alfa Europe Federation since 2004. As a founder member of this Federation he set up the legal and financial framework of the organisation. He was involved in organising two World Congresses for Alpha-1 Patients in Dublin (2004) and Rome (2005). The Federation comprises membership of most EU countries as well as Norway and Switzerland. The Federation is working on a number of projects and is building relationships with regulators, other patients, science and industry. Larry is also a member of:

Irish Platform for Patients' Organisations Science and Industry (IPPOSI), Irish Donor Network, and the Medical Research Charities Group (MRCG).



### Dr Aisling Ryan, Consultant Neurologist, Cork University Hospital

A graduate of UCC, Dr Ryan received her MB, BCh, BAO in 1995. She subsequently trained in general medicine in the Cork University Hospital and allied hospitals before specialising in Neurology. Following a period of specialist neurology training in Dublin, she moved to Germany to undertake a PhD in neuromuscular channelopathies at the Dept of Physiology, University of Ulm.

Dr Ryan returned to Ireland in 2002 to complete her clinical neurology training. She subsequently spent a year at the Centre for Neuromuscular Disease, National Hospital for Neurology and Neurosurgery, Queen Square, London before returning to a post as Consultant Neurologist in Cork University Hospital.

#### Research Interests:

Neurological Channelopathies, Muscle disease, Peripheral Neuropathies



### Mr Paolo Morgese, Market Access Manager, Health Policy & Market Access – Europe, Merck Serono SA, Geneva

Paolo Morgese holds undergraduate and postgraduate degrees in "Economics" from University of Siena (Italy) and an MSc degree in "Financial and Business Economics" from Essex University (United Kingdom). From 1999 to 2006 he worked at the Italian Competition Authority, the Italian Committee for Evaluation of Research and the Italian Agency for Health Services working on analysis of Healthcare systems and Health Technology Assessment. More recently Paolo has worked as health economist in the medical devices industry at Kyphon International. Currently, he is Market Access Manager for Europe at Merck Serono SA, the pharmaceutical division of the German Group Merck KgaA. Paolo represents Merck Serono at the Market Access Delay WG at EFPIA and is a Member of HTAi (Health Technology Assessment International).

### Mr John McCormack, Chairperson, MRCG

Mr John McCormack was appointed CEO of the Irish Cancer Society in October 2002. He was formerly Company Secretary and Financial Controller with the Society, is a member of the Institute of Certified Public Accountants in Ireland and holds an MBA from Dublin City University.

In 1991, he founded the Irish Charities Tax Reform Group which has lobbied successfully for the introduction of tax relief on corporate donations.

Two of Mr McCormack's key areas of interest are men's health and palliative care. In November 2003, he was instrumental in launching the first ever Men's Cancer Action Week.

Mr McCormack was also involved in lobbying for the implementation of a nationwide cervical cancer screening programme and for the implementation of the recommendations of the 2001 Report of the National Advisory Committee on Palliative Care.

The Irish Cancer Society is the national charity for cancer care, dedicated to eliminating cancer as a major health problem and to improving the lives of those living with cancer.

