

RARE NEUROLOGICAL DISEASES OF CHILDHOOD:

“WE TREAT THE CHILD TO TREAT THE ADULT”



2 December 2010, 11:00–13:30

European Parliament, Room J6Q1
Brussels, Belgium



WELCOME

We are pleased to welcome you today to this meeting at the European Parliament that is focused on rare neurological diseases of childhood.

The main aim of the meeting is to acknowledge the growing interest of the European Union (EU) Commission in both rare and neurological disorders. By holding this meeting, we wish to demonstrate the unity of intent of family associations, the biotechnology and pharmaceutical industries, and the scientific community in stimulating interest in rare neurological diseases. It is our belief that lessons learned from in-depth research conducted into these rare genetic neurological disorders of childhood will also inform treatment of more common neurological disorders.

The Brains for Brain Foundation, together with the European Brain Council and the Lysosomal Storage Disease (LSD) Patient Collaborative, in collaboration with the Veneto Region, sponsored by Mrs Amalia Sartori MEP, has therefore organized this meeting to give relevant stakeholders the opportunity to share views on current challenges, as well as to formulate new research strategies to improve therapy and also quality of life for patients and families affected by rare neurological disorders.

Acknowledgements

The organizers would like to thank Shire Human Genetic Therapies for providing support that has enabled them to hold this meeting.

The organizers would also like to thank Professor Edmond Wraith for his important contribution to this initiative and for his continuous support throughout the years. They also wish to thank all of the patients and families affected by rare neurological disorders for their enthusiastic support of endeavours to develop new and more effective therapies for these conditions.



PROGRAMME

11:00–11:25 Opening address

Mrs Amalia Sartori, Member of the European Parliament
Mr Stefano Beltrame, Diplomatic Councillor, Veneto Region
Dr Mary G Baker, President of the European Brain Council
Professor Maurizio Scarpa, Brains for Brain Foundation
Moderator: Dr Michael D Rogers

11:25–11:40 Keynote lecture

Brain, rare neurological diseases and European healthcare systems Mr John Bowis OBE, UK

11:40–12:10 Theme 1: Brain and rare diseases – the patients' perspective

Promoting awareness of brain diseases in Europe: enabling the dialogue between science and society Dr Mary G Baker MBE, UK

Living with a person affected by a rare disease: impact on the family and carers Mrs Christine Lavery MBE, UK

Rare diseases: the clinician is best placed to connect science and the patient Professor Dr François JM Eyskens, Belgium

12:10–12:45 Theme 2: Brain and rare diseases – the doctors' and scientists' perspectives

The burden of diagnosing rare brain diseases: medical and ethical implications Professor Dr Frits A Wijburg, Netherlands

The impact of lysosomal storage diseases on brain function Professor Tony Futerman, Israel

Health economics of orphan drugs Professor Michael Drummond, UK

The voice of industry: Assobiotech for personalized medicine Dr Riccardo Palmisano, Italy

12:45–13:00 Keynote lecture

The burden of rare neurological diseases of childhood in Europe: what is the future? Professor Timothy M Cox, UK

13:00–13:30 Panel discussion: can brain research help our society?

Consensus statement on European brain research and rare neurological diseases

13.30 Lunch

PARTICIPANTS FROM THE EUROPEAN PARLIAMENT AND THE ORGANIZING ORGANIZATIONS

Mrs Amalia Sartori MEP

Amalia Sartori serves as Member of the European Parliament for the North-East constituency with the Popolo della Libertà Party and as a Member of the Bureau of the European People's Party. She is a member of the European Parliament's Committee on Industry, Research and Energy and serves as Vice-Chair of the Euro-Latin American Parliamentary Assembly. Mrs Sartori is also a member of the Delegation for Relations with the Countries of the Andean Community. In addition, she sits as a substitute in the Committee for Internal Market and Consumer Protection, in the Special Committee on the Financial, Economic and Social Crisis and in the Delegation for Relations with the United States.



Before her first election to the European Parliament in 1999, Mrs Sartori served as a member and then Vice-Chair of the Veneto Regional Executive and subsequently as Chair of the Regional Council. As member of the Regional Executive of Veneto (Veneto Region), she was responsible for roads and transport. Her commitment to infrastructure, resource management and regulation led her to be appointed Chairwoman of the international airport in Venice and subsequently Chairwoman of the Veneto, Emilia Romagna and Piemonte inter-regional board for the management of the Po River-Veneto waterways system. Among her accomplishments, Mrs Sartori drew up the first Regional Transport Plan for Veneto, and devised the Snow Plan and the Plan for Tourist Ports. During the course of her career, Mrs Sartori has also been responsible for major institutional initiatives promoting the interests of women in organizations, in the family sphere and in society at large.

Professor Maurizio Scarpa MD PhD

Maurizio Scarpa is Head of the Lysosomal Unit at the Centre for Rare Disorders, Department of Paediatrics, University of Padova, Padova, Italy. After gaining his medical degree and doctorate from the University of Padova Medical School, he specialized in paediatrics at the same institution. He was awarded postdoctoral fellowships at the European Molecular Biology Laboratory (EMBL) in Heidelberg, Germany, investigating molecular biology and gene expression, and at the Howard Hughes Medical Institute, Institute for Molecular Genetics, Baylor College of Medicine, Houston, TX, USA, studying genetics and gene therapy. He was then appointed Consultant in Molecular Biology at the Middlesex Hospital and the London Hospital in London, UK.



Professor Scarpa is Director of the Genetics and Biochemistry PhD Programme in the Department of Paediatrics at the University of Padova and Director of the Office of Foreign Affairs of the University of Padova Medical School.

He has vast clinical experience in the diagnosis and treatment of lysosomal storage diseases (LSDs) and neurometabolic diseases, and is especially interested in developing diagnostic assays for LSDs and new approaches to their treatment. Currently, Professor Scarpa is coordinating the Shire Human Genetic Therapies Central Nervous System (CNS) Working Group and, together with Dr David Begley from King's College London, UK, he is founder and Scientific Coordinator of the Brains for Brain Foundation and Research Consortium, a pan-European task force on brain and neurodegenerative LSDs. He is also Vice-President of the Brains for Brain Foundation.

Mary G Baker MBE PhD

Mary Baker is President of the European Federation of Neurological Associations, President of the European Brain Council, Consultant to the World Health Organization (WHO) and Chair of the Working Group on Parkinson's Disease, formed by the WHO in May 1997.



In 2008, the Council of Europe re-appointed Dr Baker as a patient representative to serve on the Management Board of the European Medicines Agency (EMA), and in the same year she was appointed to the Innovative Medicines Initiative Joint Undertaking (IMI JU) Scientific Committee. In 2007, Dr Baker joined the Council of the Association of the British Pharmaceutical Industry (ABPI) and she is a member of the ABPI. Other positions include Director at Large for the World Stroke Association, a former patient editor of the *British Medical Journal* and currently Chair of the BMJ Patient Advisory Group.

Dr Baker is also Patron of the European Parkinson's Disease Association (EPDA) and former President of EPDA, a position she was elected to in 1992 when the EPDA was first formed. She retired as Chief Executive of the Parkinson's Disease Society of the UK in 2001, after holding this post for 18 years.

In 2009, Dr Baker received the British Neuroscience Association Award for *Outstanding Contribution to British Neuroscience and for Public Service* and, in 2003, an honorary doctorate was conferred upon her by the University of Surrey in recognition of her work relating to Parkinson's disease.

David J Begley PhD

David J Begley is Senior Lecturer in Physiology at King's College London. He heads up a laboratory in the Pharmaceutical Sciences Division of King's College London investigating the blood-brain barrier and drug delivery to the central nervous system (CNS), with special emphasis on lysosomal storage diseases (LSDs). He is author of more than 60 key peer-reviewed papers and 16 book chapters. Dr Begley was the Friedrich Mertz Stiftungsgast Professor at Johann Wolfgang Goethe-Universität, Frankfurt, for the academic year 1997–1998 and was sabbatical visiting Academic in Residence at GlaxoSmithKline in 2005–2007. He was organizer and Chairman of the Gordon Conference on 'Barriers of the CNS', held in New Hampshire in 2002.



He lectures frequently to worldwide audiences on the blood-brain barrier and receives research support from national research councils, the pharmaceutical industry and charitable foundations. With Professor Maurizio Scarpa of the University of Padova, Italy, he has recently set up the Brains for Brain Foundation and Research Consortium, a pan-European task force dedicated to the study and treatment of neurodegenerative LSDs.

CONTRIBUTING TO THIS INITIATIVE

Professor J Edmond Wraith MB ChB FRCPCH

Edmond Wraith is Professor of Paediatric Inherited Metabolic Medicine and Consultant Paediatrician in the Department of Genetic Medicine based at St Mary's Hospital, Manchester, UK. He specializes in inherited metabolic diseases and, in particular, in lysosomal storage diseases (LSDs) in children. He directs the nationally commissioned LSD service based at St Mary's Hospital and the Royal Manchester Children's Hospital.

Professor Wraith graduated from Sheffield University in 1977 and received his postgraduate qualification in medicine (MRCP) in 1980. In 1993, he became a Fellow of the Royal College of Physicians and, in 1997, a Fellow of the Royal College of Paediatrics and Child Health. He has served on the Editorial Board of the *Archives of Disease in Children* and is a Communicating Editor of the *Journal of Inherited Metabolic Disease*. In addition, Professor Wraith is a medical adviser to the UK Society for Mucopolysaccharide and Related Diseases and the Niemann–Pick Disease Group.

He is currently involved in several research projects, including investigations of cell-based and enzyme replacement therapies for the treatment of a number of LSDs and the use of substrate reduction therapy in patients with neurodegenerative disease. Professor Wraith has written over 200 articles, abstracts and book chapters on topics related to inborn errors of metabolism in neonates, children and adults.



MODERATOR OF THE MEETING

Michael D Rogers PhD

Michael D Rogers is a Consultant working at the interface of risk, ethics and the law, for a variety of clients in academia, foundations and private companies. He was previously the Adviser for Science and Ethics within the European Commission's Bureau of European Policy Advisers, which reports directly to the President of the European Commission. Within this multidisciplinary group, Dr Rogers was responsible for advice concerning science and society and was Secretary of the European Group on Ethics in Science and New Technologies. He has published a number of papers on the role of the precautionary principle in technological risk management under scientific uncertainty.



Dr Rogers has a PhD in physics from the University of Wales, Wales, UK. He has had a wide range of national and international experience in government service, academia and industry. Before joining the European Commission, he was for 5 years a member of HM Diplomatic Service in Tokyo. In 1990, he served as Counsellor (Science and Technology) at the European Commission Office in San Francisco, CA, USA. In 1999–2000, he was the European Union Visiting Fellow at Duke University, Durham, NC, USA. He is Visiting Professor in the School of Public Policy of the University of Tokyo (2007, 2008 and 2010) and is President of the International Council on Amino Acid Science (ICAAS). He has recently been appointed Chairman of the International Glutamate Technical Committee.



BRAIN, RARE NEUROLOGICAL DISEASES AND EUROPEAN HEALTHCARE SYSTEMS

Mr John Bowis OBE

John Bowis is President of Health First Europe and Vice-President of the European Health Forum Gastein. He is also Chairman of the Health Advisory Board of GlaxoSmithKline and Adviser to FIPRA, to Policy Action and to Hanover, and a board member of TuBerculosis Vaccine Initiative (TBVI), Global Initiative on Psychiatry, Global Alliance of Mental Illness Advocacy Networks–Europe, European Men’s Health Forum, European Health Academy, European Institute of Health, Sane, Mental Disability Advocacy Centre and Maastricht University European Health Faculty. In addition, he is a Patron of the Fund for Epilepsy, Vice-President of Diabetes UK, Vice-President of the Conservative Europe Group, Ambassador for the National Aids Trust and Ambassador for the Alzheimer’s Society.



Between 1999 and 2009, John Bowis was Member of the European Parliament for London and rapporteur for the Parliament on the European Centre for Disease Prevention and Control, mental health, neglected diseases and cross-border health. Prior to this, in 1997–1999 he served as International Policy Adviser to the World Health Organization on the global campaigns ‘Nations for Mental Health’ and ‘Out of the Shadows’ (epilepsy). Between 1987 and 1997, he was a Member of the UK Parliament, and Minister for Health & Social Services and then Minister for Transport.

John Bowis was awarded the OBE in 1981. He is an Honorary Fellow of both the Royal College of Psychiatrists and the Royal College of Physicians.

11:40–12:10 THEME 1: BRAIN AND RARE DISEASES – THE PATIENTS’ PERSPECTIVE

LIVING WITH A PERSON AFFECTED BY A RARE DISEASE: IMPACT ON THE FAMILY AND CARERS

Mrs Christine Lavery MBE

Christine Lavery was appointed Chief Executive of the Society for Mucopolysaccharide and Related Diseases (MPS) in 1993. During her time at MPS, she has taken the charity to new heights, managing a research budget of over £500,000 per year and a UK-wide advocacy service providing needs-led support to over 1200 children and adults with MPS, their families and professionals, in the areas of home adaptations, special educational needs, access to new therapies, respite care, palliative care and pre- and post-bereavement support. Mrs Lavery has served on the Department of Health Advisory Board on Genetic Testing and is currently the patient representative in the Department of Health National Specialized Commissioning Advisory Group.



Between 1984 and 1993, Mrs Lavery was employed as National Development Officer at Contact a Family, a national UK charity. During this time, she worked with parents and carers to set up over 300 patient support groups for a wide range of specific diseases. In 1991, with the help of a researcher, Mrs Lavery wrote the first edition of the Contact a Family Directory of Rare Diseases and Support Networks, which is now available on the Internet.

Prior to 1984, Mrs Lavery lived in Japan for 4 years with her husband and two children, the eldest of whom was terminally ill with Hunter disease. During her husband's tour of duty as First Secretary at the British Embassy, Mrs Lavery worked voluntarily for Save the Children and the International Year of the Child, and organized the first diplomatic sponsored climb of Mount Fuji.

Mrs Lavery was awarded an MBE for her services to metabolic diseases by HM Queen Elizabeth II in the New Year's Honours List for 2002, and at the 2006 International Symposium on Mucopolysaccharide and Related Diseases received 'a lifetime award' from the International MPS Community.

Abstract

Being diagnosed with a rare disease or being the parent of a child diagnosed with a rare disease is usually a devastating and life-changing experience. Apart from the challenges of the particular condition, no-one around you has heard of the disease – and that often includes the medical teams and local services. An effective patient organization like the Society for Mucopolysaccharide Diseases can become a lifeline for patients and their families, providing individual social care support and accessible and up-to-date information on specific diseases, and on clinical experts, treatment options and, when appropriate, clinical trials. The MPS Society is playing a vital role in funding research that may lead to clinical benefit and new therapies for its members, and has already provided in excess of £5 million of research funding over the past 15 years.

RARE DISEASES: THE CLINICIAN IS BEST PLACED TO CONNECT SCIENCE AND THE PATIENT

Professor François JM Eyskens MD PhD

François Eyskens studied medicine at the University of Antwerp before training in paediatrics at the Kinderziekenhuis Antwerpen. He then trained in metabolic diseases at the Wilhelmina Kinderziekenhuis, Utrecht. By 1987, he was involved in the neonatal mass screening programme for metabolic disorders in Antwerp at the Provinciaal Centrum voor Metabole Aandoeningen (PCMA). He obtained his PhD in 1997 for his thesis entitled 'Neonatal screening: the experience in Antwerp'.

Professor Eyskens is currently working as a Resident in Paediatrics at the University Hospital of Antwerp and coordinates the Center for the Study of Metabolic Diseases (CEMA). He is Associate Professor in Paediatrics at the University of Antwerp and Director of the PCMA Metabolic Laboratory, where screening and analysis of different metabolic compounds is performed. Professor Eyskens' specific areas of research interest include screening for metabolic diseases, organic acidurias, lysosomal storage diseases and cognitive dysfunction in metabolic diseases.



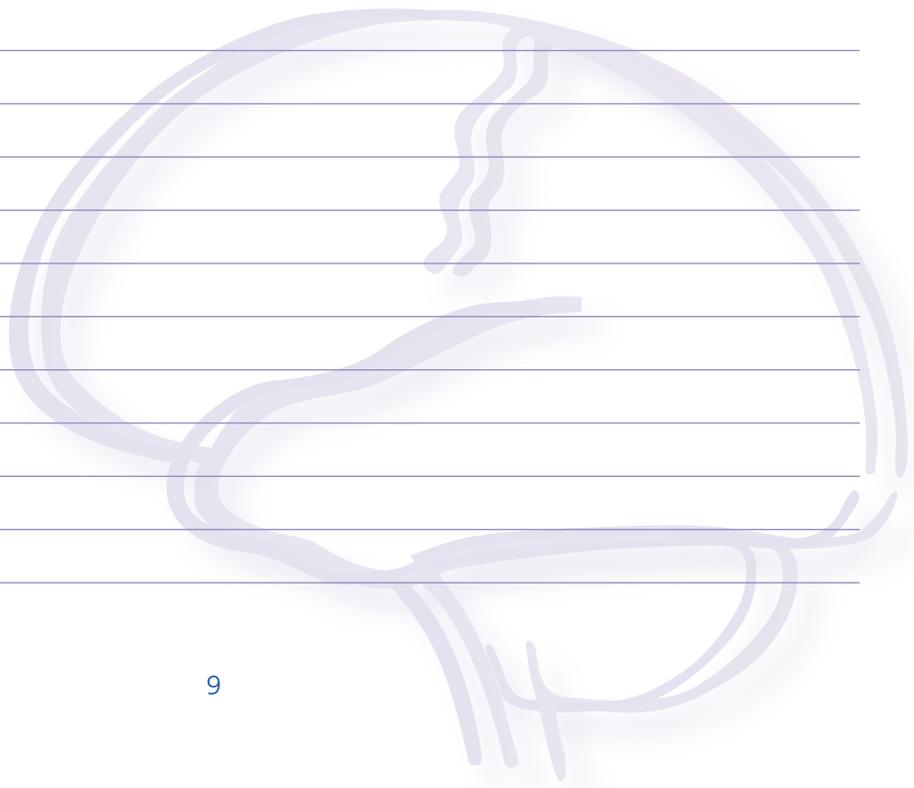
Abstract

Metabolic diseases are rare inherited disorders that manifest at various ages. These diseases may be detected in neonatal mass screening programmes, but more often the diagnosis relies on recognition of signs and symptoms (either specific or nonspecific) by a physician. Sometimes, it takes years for a patient to receive the correct diagnosis, despite numerous consultations and examinations. Brain involvement is present to varying degrees in patients with metabolic diseases. This may include severe neuronal loss, white matter lesions, vascular or metabolic stroke and cognitive dysfunction. Many of these diseases are treated by dietary measures, while some are treated with drugs. The specific drugs used in treatment belong to a group designated 'orphan drugs'. After initial enthusiasm ("finally, we can treat these patients"), it is now becoming apparent that most of these therapies do not cross the blood–brain barrier in sufficient quantities to achieve anything more than limited therapeutic efficacy in the central nervous system. A good doctor–patient relationship and appropriate care is therefore central to alleviating physical and psychological suffering and limiting frustration, so that the quality of life of these patients is influenced positively.

NOTES



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12.10–12.45 THEME 2: BRAIN AND RARE DISEASES – THE DOCTORS' AND SCIENTISTS' PERSPECTIVES

THE BURDEN OF DIAGNOSING RARE BRAIN DISEASES: MEDICAL AND ETHICAL IMPLICATIONS

Professor Frits Wijburg MD PhD

Frits Wijburg studied medicine at the University of Amsterdam and specialized in paediatrics at the Academic Medical Center (AMC) in Amsterdam, the Netherlands. He obtained his PhD in 1993 from the University of Amsterdam for his thesis on 'Mitochondrial respiratory chain disorders'. In 1992–1994, he trained in paediatric gastroenterology and nutrition at the University Medical Center in Groningen, the Netherlands. He then returned to the AMC to train in inherited metabolic diseases.

In 2004, he became a full Professor and Chairman of the Division of Clinical Metabolic Diseases of the Emma Children's Hospital at the AMC, one of the largest units for metabolic disorders in the Netherlands. His clinical and research groups focus on lysosomal storage diseases, with special emphasis on optimal timing and efficacy of treatment, as well as on inborn errors of intermediary metabolism. Professor Wijburg is a member of numerous national and international committees on metabolic disorders and has published over 135 peer-reviewed publications on this subject.



Abstract

More than 300 different inherited disorders are currently known to cause brain disease in children. Although the individual disorders are rare, their combined prevalence is significant, resulting in a high burden for society. Many of these diseases are characterized by a relentless progressive course, leading to severe mental and motor handicaps, dementia and early death. While improved technology has facilitated reliable diagnosis in most of these patients, a lack of knowledge about the underlying pathophysiological mechanisms still hampers the development of disease-modifying treatment. For parents, the stark contrast between technologically advanced diagnostic tests on the one hand and the complete lack of therapeutic options on the other hand is incomprehensible and intolerable. Diagnosis is often followed only by supportive and palliative care.

The family may be offered prenatal testing during subsequent pregnancies but, because many of these disorders are characterized by an initial symptom-free interval, it is not uncommon that another child has already been born into the family before an older child has been diagnosed. As a result, families may bear the heavy burden of having two or more children with a debilitating and ultimately fatal brain disease. Earlier diagnosis through newborn screening might prevent this. However, newborn screening is generally considered permissible only if a disease-modifying treatment is available.

THE IMPACT OF LYSOSOMAL STORAGE DISEASES ON BRAIN FUNCTION

Professor Tony Futerman BSc PhD

Tony Futerman received his BSc in Biochemistry from the University of Bath, UK, in 1981, then moved to the Department of Neurobiology of the Weizmann Institute of Science, Israel, for his doctoral studies. In the course of his PhD research, he demonstrated that acetylcholinesterase, a key enzyme in terminating neuronal signalling in the brain, is attached to the cell membrane via a novel mechanism. After 3 years of postdoctoral studies at the Carnegie Institution in Baltimore, MD, USA, he joined the Weizmann Institute, where he is currently a full Professor in the Department of Biological Chemistry.



Professor Futerman chaired the 2006 Gordon Conference on 'Glycolipid and Sphingolipid Biology' and will chair the 2011 Gordon Conference on 'Lysosomal Diseases'. He also serves on the editorial board of the Journal of Biological Chemistry. In addition to authoring approximately 150 scientific papers, he has edited the book *Ceramide Signaling* and co-edited a book on *Gaucher Disease*. At the Weizmann Institute, he is the incumbent of the Joseph Meyerhoff Professorial Chair of Biochemistry and serves as Director of the Nella and Leon Benozzi Center for Neurological Diseases.

Abstract

Lysosomal storage disorders (LSDs), of which more than 40 are known, are caused by the defective activity of lysosomal proteins, which results in intralysosomal accumulation of undegraded metabolites. Despite years of study of the genetic and molecular basis of LSDs, little is known about how accumulation of these metabolites causes pathology. After summarizing the biochemistry of LSDs, this presentation will discuss some of the downstream cellular pathways that are potentially affected in these disorders. The focus will be specifically on different cell populations in the brain, such as neurones, glia and microglia, and the presentation will explain how defects in these cells might lead to brain dysfunction. In addition, this talk will emphasize that much more basic research is required before we are in a position to understand fully the pathological mechanisms involved, a pre-requisite for finding new therapies for these devastating diseases.

HEALTH ECONOMICS OF ORPHAN DRUGS

Professor Michael Drummond BSc MCom DPhil

Michael Drummond is Professor of Health Economics and former Director of the Centre for Health Economics at the University of York, UK. He is particularly interested in the economic evaluation of healthcare treatments and programmes. He has undertaken evaluations in a wide range of medical fields, including care of the elderly, neonatal intensive care, immunization programmes, services for people with AIDS, eye healthcare and pharmaceuticals. He is the author of two major textbooks and more than 600 scientific papers, has acted as a consultant to the World Health Organization and was Project Leader of a European Union Project on the Methodology of Economic Appraisal of Health Technology. He has been President of both the International Society of Technology Assessment in Health Care, and the International Society for Pharmacoeconomics and Outcomes Research. He is currently a member of the Guidelines Review Panels of the National Institute for Health and Clinical Excellence (NICE) in the UK, and is a Principal Consultant for i3Innovus. In 2008, he received an Honorary Doctor of Science from City University, London, UK.



Abstract

Most new health technologies (drugs, devices or procedures) deliver substantial health benefits, but usually add to healthcare costs. For this reason, many European Union (EU) member states have introduced procedures to assess the clinical efficacy and cost-effectiveness of health technologies in order to determine whether they should be funded. These procedures make an important contribution to the efficiency of European healthcare systems, but pose challenges for drugs for rare diseases. These drugs are almost never shown to be cost-effective by conventional standards, because the costs of research and development have to be recovered from sales to a much smaller patient population than is the case for drugs for more common diseases. In addition, it is harder to

assemble clinical evidence for rare diseases than for common diseases because of the small number of patients available for enrolment into clinical trials. Most member states exclude drugs for rare diseases from standard health technology assessments because of these and other special considerations, including the fact that the drugs are often for serious or life-threatening diseases for which no alternative treatments are available. It could also be argued that, on grounds of equity, people with rare diseases should have the same access to care as the general population. Finally, given the small number of patients with rare diseases in individual EU member states, there are strong arguments for European collaboration in clinical research.

THE VOICE OF INDUSTRY: ASSOBIOTEC FOR PERSONALIZED MEDICINE

Riccardo Palmisano MD

Riccardo Palmisano is Executive Vice-President of Assobiotech and Vice-President and General Manager of Genzyme, Italy. After graduating in medicine from the University of Parma in 1985, he joined the pharmaceutical industry, where he has held managerial positions of increasing responsibility. His professional career began at Farmitalia Carlo Erba, where he was Product Manager and Medical Manager, first in the cardiovascular area and then in the gastroenterological area. In 1988, he joined the Menarini Group, where he worked in sales and marketing to support the growth and development of this leading Italian pharmaceutical company during the first half of the 1990s. In January 1993, he took on responsibility for all Italian pharmaceutical operations (medical, marketing, sales, logistics and public relations) for the group headquarters based in Florence. This professional development culminated in mid-1995 with his appointment as General Manager of Lusofarmaco S.p.A., a subsidiary of the group, based in Milan. After gaining considerable experience working for these two major Italian pharmaceutical companies, he joined a multinational company, Shire Pharmaceuticals, in 2000. As General Manager and Managing Director of Shire Pharmaceuticals, he set up the Italian operations from scratch. In 2003, he was recruited by GlaxoSmithKline (GSK) as Vice-President Commercial Retail Market, to lead a thorough commercial restructuring project at their Italian branch. By December 2004, GSK Italy had climbed from thirteenth to first position among the multinational companies operating in Italy.



Since November 2005, Riccardo Palmisano has held the position of Vice-President and General Manager of the Italian affiliate of Genzyme Corporation (Cambridge, MA, USA). This role has broadened his experience and knowledge of biotechnology. His work encompasses the areas of orphan drugs for rare diseases, as well as advanced treatments in other areas, including onco-haematology, nephrology, niche endocrinological conditions and multiple sclerosis. In 2008, he became a member of the Executive Committee of Assobiotech and, subsequently, joined the Board of Directors. In 2010, he was elected Executive Vice-President. In Assobiotech, he also leads the Biotech Pharma Working Group. In 2008–2009, he launched a project in the field of preclinical research involving partnership between Genzyme, Italy, and a number of important stakeholders representing the Italian public and the non-profit research environment.

Abstract

Assobiotech, the Italian trade association of biotechnology industries, is affiliated to Federchimica and represents green (agriculture), white (industrial) and red (healthcare) biotechnology industries. In the life sciences arena, Assobiotech represents a variety of biotechnology companies operating in Italy ranging from start-up companies, through pure biotechnology multinational subsidiaries to branches of traditional Big Pharma that are highly focused on innovation, as well as science and technology parks. This peculiar characteristic, gives Assobiotech a clear overview of the entire drug development path from discovery of new chemical entities to establishing market access. This includes an understanding of future business models for sustainable innovation in a limited resources scenario. Key aims for the future are to: (1) focus on unmet clinical needs (including rare diseases and neurological disorders) and personalized medicines; (2) optimize the resources used to fund innovative research, through connectivity and collaboration, at least in Europe; (3) encourage partnerships among scientists, universities, public and non-profit research and private industry, with the aim of facilitating both discoveries/preclinical platforms and subsequent industrialization and registration. Improving the technology transfer process could result in more new therapies being available to patients. Therefore, Assobiotech is not asking the European Parliament or national governments for more funds, only for clearer rules and ways to expedite networking and partnering. It is necessary to capture the current momentum regarding research into rare diseases so that we do not miss the opportunity to create value for patients/public health and the European Union economy.

THE BURDEN OF RARE NEUROLOGICAL DISEASES OF CHILDHOOD IN EUROPE: WHAT IS THE FUTURE?

Professor Timothy M Cox MD PhD

Timothy Cox is Professor of Medicine in the University of Cambridge, UK – a foundation chair to which he was elected in 1988. As an internist with experience in pathology and a special interest in inborn errors of metabolism, he graduated from the University of London in 1971 with postgraduate qualifications in Biochemistry and Medical Science. In 1983–1984, he was Visiting Scientist in the Department of Biology at the Massachusetts Institute of Technology, Cambridge, MA, USA. As an educator, he founded the Cambridge MB/PhD programme in 1989, of which he is Honorary Director – this was the first programme of its kind in the UK. As Honorary Consultant Physician, in 1996 he founded the National Service Centre for the treatment and diagnosis of lysosomal storage diseases at Addenbrooke's Foundation NHS Hospitals Trust Cambridge; this is funded by the UK Department of Health.



Professor Cox, who is a Fellow of the UK Academy of Medical Sciences, has made discoveries relating to the genetic and molecular basis of several disorders, including haemochromatosis, porphyria and hereditary fructose intolerance; his therapeutic research is currently focused on neurodegenerative aspects of lysosomal storage disorders. He edited and authored Molecular Biology in Medicine and, with DA Warrell and JD Firth, is editor of the three-volume Oxford Textbook of Medicine; he has contributed over 220 original articles and invited chapters in textbooks and peer-reviewed journals.

Abstract

Principally as a result of the Human Genome Project, we possess a lexicon of approximately 6000 disease-related genes – including many that cause rare ('orphan') disorders. This biological revolution facilitates a molecular understanding of disease and promotes therapeutic innovation, but the spectacular science has been accompanied by a societal revolution in the way we understand the needs of those directly affected by rare disorders: the physical burden is accompanied by a moral responsibility to provide relief. The analogy of the broken machine, with diseased parts (or the whole) to be replaced or rejected, is outmoded. Darwinian concepts are democratic and rational: illness results from interactions between the individual and an environment to which they are temporarily maladapted. Constitutional factors are the product of selection and persist as determinants of species diversity and survival.

This golden age of medicine and biological science not only allows a better understanding of disease but also expedites our search for cures. However, challenges remain, and ensuring that delivery of corrective medicines to the injured brain and all its constituents is one challenge that merits intensive and continued effort.

Gene and stem-cell transfer promise more definitive correction of brain diseases, but their development is impeded by regulation and weak commitment. Greater political investment is mandatory if we are to realize the human value of discoveries in rare diseases affecting the most vulnerable in our society. This imperative depends on an unassailable Harveian principle of medicine: the study of unusual conditions provides unique insights into the treatment of common ones. Brain diseases are prevalent in Europe and it is essential that we invest now to reduce their immense future burden.

THE ORGANIZERS

Brains for Brain Foundation

The Brains for Brain Foundation was founded in 2007 by highly distinguished research scientists and clinical neurologists dedicated to increasing scientific understanding of rare paediatric neurological diseases, particularly neurodegenerative disorders, and improving their treatment. At present, over 60 universities (in 12 European Union [EU] member states, USA, Brazil and Australia) and 10 biotechnology companies are collaborating with the Brains for Brain Foundation to develop innovative therapeutic approaches, which may also be applicable to more common disorders such as Alzheimer's and Parkinson's diseases, brain tumours, and spinal lateral atrophy.

The Brains for Brain Foundation believes in, and actively encourages, focused and organized international collaboration in order to facilitate research that aims to meet the needs of patients with neurological diseases.



European Brain Council

The European Brain Council (EBC) is a coordinating body formed by European organizations in neurology, neurosurgery, psychiatry and neuroscience, as well as patient organizations and industry. It therefore represents a vast network of patients, doctors and research scientists; these stakeholders, together with its industrial partners, make it eminently suited to working in close partnership with EU commissions, the European Parliament and the World Health Organization (WHO), as well as other decision-making bodies. The EBC was officially founded on 22 March 2002 in Brussels, and has offices in Brussels and Florence.



Lysosomal Storage Disease (LSD) Patient Collaborative

The LSD Patient Collaborative represents those affected by lysosomal storage disorders in the UK. The group comprises representatives from the Association for Glycogen Storage Disease (UK), Batten Disease Family Association, Gauchers Association, Society for Mucopolysaccharide Diseases (MPS Society), Niemann–Pick Disease Group (UK) and the Save Babies Through Screening Foundation (UK). The mission of the group is to promote awareness of and interest in LSDs, advance standards of care, enhance the well-being of those affected, and establish a forum in which members can discuss matters of common interest and contribute to the development and dissemination of good practice. The key objective is to provide a strong voice with which to influence national policy.



Regione del Veneto

The Veneto Region, situated in the north-eastern part of Italy, is divided into seven provinces with about 4.8 million people living in 580 municipalities (2009). Veneto Regional Government is an autonomous, territorial organization which, together with the other 19 Italian Regions, the State and the Autonomous Provinces, makes up the Italian Republic. The Regional Government has legislative and administrative competence in the following areas: general organization; health and social services (education, promotion of cultural activities, social assistance, health protection, sports and leisure); economic development; and regional planning (public works, water supply, roadway and railway network, urban planning, environmental protection). To date, the regional university system is involved in more than 150 research projects funded by the Seventh Framework Programme.



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