Previous recipients of the EURORDIS Black Pearl Awards

2012-2017

2018

Patient Organisation Award – PHA Europe
Volunteer Award – Helene and Mikk Cederroth
Volunteer Award – Chris Sotirelis
Young Patient Advocate Award – Sammy Basso
European Rare Disease Leadership Award – Bruno Sepodes
Policy Maker Award – Elena Gentile
Visual & Audio Media Award – Special Books by Special Kids
Written Media Award – Serge Braun
Scientific Award – Prof Michele De Luca & Dr Thomas Hirsch
Company Award for Innovation – Novartis
Company Award for Patient Engagement – EFPIA, MSD, Bayer, UCB
Lifetime Achievement Award – Alastair Kent

2012

Patient Organisation Award - Association Française des Myopathies
Volunteer Award - Michele Lipucci Di Paola
European Rare Disease Achievement Award - Kerstin Westermark
Policy Maker Award – Andrea Vassiliou
Scientific Award - Professor Alain Fisher and Professor Maria Grazia Roncarolo
Media Award – BBC
Company Award – CSL Behring
Company Award – Shire
Company Award – Sigma Tau Pharmaceuticals, Inc

2017

Patient Organisation Award - Dravet
Volunteer Award – Elizabeth Vroom
Policy Maker Award – Frédérique Ries
Scientific Award – Lucia Monaco
Media Award – Aldo Soligno
Company Award – GlaxoSmithKline
Lifetime Achievement Award – Anders Olauson

2013

Patient Organisation Award – Alström Syndrome UK
Volunteer Award – Lesley Greene
European Rare Disease Leadership Award – Dr Ruxandra Draghia-Akli
Policy Maker Award – Françoise Grossetête
Scientific Award – Dr Ségolène Aymé
Media Award – Andrew Jack
Company Award – Celgene Corporation
Company Award – Prosensa
Company Award – Genzyme, a Sanofi Company

EURORDIS
Black Pearl
AWARDS

2016

Patient Organisation Award – UNIQUE – The Rare Chromosome Disorder Support Group
Volunteer Award – Tsveta Schyns-Liharska
European Rare Disease Leadership Award – Antoni Montserrat Moliner, Jarek Waligóra, Michael Hübel
Policy Maker Award – Christian-Silviu Buşoi
Scientific Award – Prof. Dr. Peter N. Robinson
Media Award – France Télévisions – AFM-Télethon, France
Company Award – Actelion, Switzerland
Lifetime Achievement Award – Renza Barbon Galuppi, Italy

2014

Patient Organisation Award – Allianz Chronischer Seltener Erkrankungen
Volunteer Award – Lise Murphy
European Rare Disease Leadership Award – Paola Testori Coggi, Prof Guido Rasi, Prof Luca Pani
Policy Maker Award – Antonyia Parvanova
Scientific Award – Professor Hans-Hilger Ropers
Media Award – Rick Guidotti
Company Award – Sobi
Company Award – Orphan Europe

2015

Patient Organisation Award – Children with SMA
Volunteer Award – Rosa Sánchez de Vega
European Rare Disease Leadership Award – Prof Josep Torrent-Farnell
Policy Maker Award – Glenis Willmott
Scientific Award – Prof Kate Bushby
Media Award – Peter O'Donnell
Company Award – Pfizer, Inc.
Lifetime Achievement Award – Abbey Meyers

2017

EURORDIS Volunteer Award – Elizabeth Vroom

Elizabeth Vroom is mother to a son with Duchenne Muscular Dystrophy (DMD) and founded the Duchenne Parent Project in the Netherlands, of which she remains president to this day. In 2002, Elizabeth co-founded the United Parent Projects Muscular Dystrophy (UPPMD) through which she helped establish World Duchenne Awareness Day."Receiving an award is very special, but even more so because this comes from a community I treasure and consider as family. I am very honoured to receive the Volunteer Award for 2017 as I know how many people like me work hard to change the lives and future of patients with rare diseases."
Elizabeth Vroom

Media Award – Aldo Soligno

In October 2014, Aldo Soligno led the project 'Rare Lives', a powerful photographic storytelling tool that gives an insight into the daily lives of people living with a rare disease. "Spending days and weeks with families and patients suffering from a rare disease has meant so much to me to see how opposites can meet and turn into one another: weakness into strength, sadness into joy, despair into determination. Seeing this happen simultaneously in 7 different European countries has confirmed to me that there are no boundaries for scientific research and solidarity and they must not exist for rare disease patients too. I really hope this will be the legacy of my project."
Aldo Soligno

Lifetime Achievement Award – Anders Olauson

Anders Olauson has devoted the past 30 years of his life to the rare disease cause. In 1989 he founded the Ågrenska Centre, a national competence centre in Sweden which provides support for people affected by rare diseases. President of the European Patients' Forum from 2005 to 2015, Anders remains Honorary President of the organization. "I think it's fantastic that I just want to share it with so many people to have this award. It's a recognition that all the work you have done, all the travels and all the things and energy you have put in, that it has given a result and that result has been so greatly appreciated. I am totally honoured. Having a rare disease affects all aspects of life; experiences from thousands of families worldwide tell the same story. All areas of life – healthcare, social services, schools, insurances and labour – must work together. Life is holistic, as should care and support be. Working together at the United Nations will make this possible." *Anders Olauson*

Scientific Award – Lucia Monaco

Lucia Monaco is Chief Scientific Officer at the Fondazione Telethon and has made a significant impact in the field of rare disease in Italy and abroad, in particular through her strong commitment to the IRDiRC initiative. Her personal enthusiasm, guidance and dedication inspires many scientists and clinicians to become involved in state-of-the-art research for rare diseases such as the development of gene therapies. "I feel deeply honoured receiving this award, which I believe really recognizes the value of the organization which I have the fortune to work with, as well as the value and the extraordinary results of our scientists. The Fondazione Telethon is a charity which is committed to fight rare genetic diseases and to do this we support excellent research, but our mission is to bring the results of this research to patients. The results we have seen over the past few years could not have been achieved without a strong collaboration among all of the stakeholders, colleagues, patients and their families." *Lucia Monaco*

Policy Maker Award – Frédérique Ries

Frédérique Ries, Member of the European Parliament since 1999, has been a pioneer in the implementation of patient-centric health policies that focus on Union-added value, especially in the field of rare diseases. Her most recent work, a project proposal that promotes a continuous bottom-up research approach including patients, demonstrates Ries' consistent dedication towards tackling rare diseases in the European Union.

Company Award – GlaxoSmithKline

Since 2010, GSK has designated rare diseases as one of its core therapeutic areas. In 2016, GSK brought to market an innovative gene therapy to treat patients with the rare disease adenosine deaminase severe combined immunodeficiency syndrome (ADA-SCID). "It is a huge privilege and a great honour, as well as humbling to accept this award on behalf of GSK. Since coming into the rare disease universe, I have felt overwhelmed by the welcome that you get, that everyone has a single-minded goal which is all around patient treatment and patient outcome. You don't have to look very far to be inspired in this world. This award goes to all the people who have the courage, determination and focus, and who put in all their energy to make a change for all people living with severe rare disease. And of course to all the patients and families who drive us each day." *Martin Andrews*

Patient Organisation Award – Dravet

The Dravet Syndrome European Federation was founded in 2014 by eight patient organisations in seven countries. They now work together to raise awareness and to fight the impact of Dravet Syndrome—also known as Severe Myoclonic Epilepsy of Infancy (SMEI)—on patients and their families."It is extremely important to us to receive this recognition. Personally for me EURORDIS is like my family. I attended the summer school 3-4 years ago and I have learnt so much from all that EURORDIS is doing. We are a new federation that has developed over the past 3 years. We created this federation with the determination of working on new treatments for Dravet, working in three pillars: Removing the barriers for research such as developing a new animal model to help facilitate treatment and how new drugs can be tested for Dravet. Digital transformation for our patients and preparing important and the right data for our clinicians, researchers, physicians and patients. And lastly collaboration between different multi-stakeholder groups. These are the 3 pillars in which we base our work." Julian Isla

2016

Policy Maker Award – Christian-Silviu Buşoi

Cristian-Silviu Busoi, Member of the European Parliament since 2007, has consistently demonstrated a strong vision of patient centric, quality and accessible medical systems across Europe in this position. A physician and former lecturer in Public Health and Health Management at the Victor Babes University of Medicine and Pharmacy. As a member of the ENVI Committee within the European Parliament, he has used this platform to champion patients' rights in each country, launching the public "Patients' rights' campaign, supporting a multi-centre collaboration at EU level to encourage partnerships between all rare disease stakeholders. In holding parliamentary events in support of rare cancers and rare diseases, and co-hosting the Rare Disease Day policy event to improve access to therapies for rare diseases, he has shown devotion passion in addressing the needs of rare disease patients across Europe, making him a truly deserving winner of the EURORDIS Policy Maker Award 2016.

Volunteer Award – Tsveta Schyns-Liharska

Tsveta Schyns-Liharska has a Phd and Post doc in genetics from Wageningen University and the Free University in Amsterdam, respectively. As a parent of a daughter affected with the rare disease alternating hemiplegia, Tsveta has dedicated a considerable amount of time to caring for her daughter and to volunteering for the rare disease community. Tsveta's volunteer activities include being a patient representative on the Paediatric Committee (PDCO) of the European Medicines Agency since 2008 and for 8 years dedicating a massive amount of time and work as Scientific Coordinator of the European Register for Multiple Sclerosis Project. A true achievement has been the founding and running, as Secretary General, of ENRAH and the work Tsveta has done for the EU Public Health Programme. This award serves to recognise all that Tsveta has accomplished in supporting so many rare disease organisations on a volunteer basis and to recognise her long and faithful service as a EURORDIS volunteer.

Media Award – France Télévisions – AFM-Télethon, France

The EURORDIS Media Award recognises the long-standing support, of France Télévisions in broadcasting live the French Telethon. Millions of people have taken part and donated to the AFM-Téléthon cause, making it possible to support research and create the Institute of Biotherapies for Rare Diseases, with laboratories such as the Myology Institute, Généthon, I-Stem et Atlantic Gene Therapies. Généthon, for example, stands out through its unique ability to develop, produce and test its own innovative gene-based medicines for rare diseases, the creation of which has been made possible from the proceeds of the AFM-Téléthon. Moreover, thanks to the French Telethon, it has been possible to inform the general public about rare diseases, promote changes in the legal framework in France and in Europe and improve the daily life of patients.

Patient Organisation Award – UNIQUE – The Rare Chromosome Disorder Support Group

UNIQUE has been a source of mutual support and self-help to families of children with a rare chromosome disorder since it was founded by Edna Knight MBE in the UK in 1984 as the Trisomy 9 Support Group. Starting with 1192 families, to now representing over 14,000 families world-wide in over 90 countries, UNIQUE as an organisation has worked hard to raise awareness of rare chromosome disorders to professionals and to the general public so that they too have an appreciation of the extraordinary challenges their members face. This incredibly well-deserved award serves to recognise the efforts and successes that have resulted from the hard work and determination of UNIQUE.

Scientific Award – Prof. Dr. Peter N. Robinson, Institute for Medical Genetics, Universitätsklinkum Charité, Germany Support Group, U

Professor Peter N. Robinson is a Professor for Medical Genomics at the Charité Universitätsmedizin Berlin in Germany, as well as Research Group leader at the Institute of Medical Genetics and Human Genetics of the Charité – Universitätsmedizin Berlin. Amongst other activities, Peter has developed the Human Phenotype Ontology (HPO), as well as a number of algorithms for disease gene prediction and next-generation sequencing data. His developments contribute also to correlate animal models and human diseases. Peter's team's output in recent years has included the development of a novel treatment strategy for Marfan syndrome in mice based on antagonism of a class of bioactive motivs that are common in fragments of elastin and fibrillin-1, the identification of novel disease genes for a form of ataxia (CA8) and hyperphosphatasia with mental retardation syndrome (PIGV). Peter Robinson has concentrated his diverse background and skills to improve the understanding and the diagnosis of inherited diseases.

Company Award – Actelion, Switzerland

Established in 1997, Actelion focuses on the discovery, development and commercialization of innovative drugs for diseases with significant unmet medical needs. The Company has, in particular, made a difference for pulmonary arterial hypertension (PAH) in Europe and globally. Actelion's pipeline reflects continued commitment to address unmet medical needs, and they have engaged with and supported patient organisations since their founding. An Emerald member of the EURORDIS Round Table of Companies (ERTC), Actelion has supported several key EURORDIS initiatives, including: the EurordisCare survey, EURORDIS Membership Meetings, and the Black Pearl Evening, which helps make possible EURORDIS' actions to end isolation of people living with a rare disease, empower leaders of the rare disease patient community, and raise awareness of all rare diseases.

European Rare Disease Leadership Award – Antoni Montserrat Moliner, Jarek Waligóra, Michael Hübel

EURORDIS has decided to jointly present the European Leadership Award 2016 to three key pioneers from the Directorate General of Health and Food Safety (DG-SANTE) within the European Commission. **Antoni Montserrat Moliner** has championed patient engagement, particularly in his involvement in EUROPLAN and National Plans. Montserrat has driven encouraging actions in rare cancers, ensuring such patients can benefit from advances in both the cancer and rare disease fields. A clinical genetic paediatrician by training, **Jarek Waligóra** has tirelessly brought his medical expertise to his position of Policy Officer for rare diseases at the European Commission (EC). Waligóra specifically worked on the EC report on the implementation of the Council Recommendation on Rare Diseases, and has been pioneering in setting the policy agenda for the first and the current Rare Disease Joint Actions. **Michael Hübel**, Head of the Unit of Programme Management and Diseases at DG-SANTE within the European Commission, has been instrumental in initiating and supporting policies around rare diseases, notably in the establishment of the Commission Expert Group on Rare Diseases and the Commission Expert Group on Cancer Control.

Lifetime Achievement Award – Renza Barbon Galuppi, Italy

Renza Barbon Galluppi is believed to be a "wonder woman" for her ability and strength to be where she is needed at the right moment, even though it means attending conferences, meetings and workshops in three different cities in two days. It has been calculated that in the past few years she has spent an average of 15 hours a day in activities related to Rare Diseases advocacy and to answering individual patients or Patient Organisations and organizing projects to train Rare Disease patients' representatives on key topics. It all started with the diagnosis of a typical type of hyperphenylalaninemia given to two of her three children. The delay in the delivery of the diagnosis to her eldest daughter and its consequences led her first to start the collaboration with the Patient Organisation involved in metabolic diseases but shortly after with UNIAMO, the Italian Federation of Rare Diseases, to contribute to addressing all the transversal needs.In the past 10 years, Renza has contributed to stressing the importance for patients with Rare Diseases to share their experiences within associations and promote their integration into the community in every facet of life. In particular, she has advocated for patient representatives to be part of the expertise and decision making process, and has committed to the social innovation project, 'Ristoro Fantasia', overcoming mental and social barriers within young patients affected by rare diseases.

2015

European Rare Disease Leadership Award – Professor Josep Torrent-Farnell

Professor Josep Torrent-Farnell is a qualified Pharmacist and a specialist in Internal Medicine, and is a member of the Scientific Advice Working Party (SAWP) at EMA, Professor of Clinical Pharmacology and Therapeutics at the Autonomous University of Barcelona, and former Director General of the Fundació Doctor Robert, Advanced Centre of Services and Training for Health and Life Sciences. Josep has consistently demonstrated extraordinary leadership in the field of rare diseases, beginning with his membership of the Committee for Orphan Medicinal Products (COMP) at the European Medicines Agency in 2000 where he later went on to serve as First Chairperson for 6 years. He is also a member of the European Task Force for Rare Disease (DG SANCO). EURORDIS is delighted to present Professor Torrent-Farnell with the European Rare Disease Leadership Award as a symbol of his tireless devotion to, and his pioneering leadership of, the rare disease community. Not only has he determinedly supported the promotion of orphan drugs at conferences around Europe and the World but he has been instrumental in encouraging dialogue between EMA and FDA, was key to the creation of the Catalonian Alliance of People with Rare Diseases and became the 1st Chairperson of the Therapeutic Scientific Committee of IRDiRC. Josep continues to be a long-standing supporter of EURORDIS and the projects we undertake such as the Summer School and the Round Table of Companies Workshops and participates in collaborations with over 70 patient organisations.

Policy Maker Award – Glenis Willmott

Glenis Willmott, Labour Member of the European Parliament for the East Midlands in the UK since 2006 and threetime re-elected leader of the European Parliamentary Labour Party, has demonstrated outstanding dedication and commitment in addressing the needs of patients in the European Union. Ms Willmott is an active member of various committees and forums such as the Environment, Public Health and Food Safety Committee, the Delegation for relations with Canada, and the MEPs against Cancer Forum. Yet most remarkable of all is the instrumental role Ms Willmott has played in the passing of key legislation through her work as Rapporteur for the "Regulation on Clinical Trials on medicinal products for human use" and Shadow Rapporteur for the Regulation establishing a "Health for Growth Programme". These two pieces of EU legislation have a tremendous impact on the lives of the estimated 30 million people living with a rare disease in Europe.

Volunteer Award – Rosa Sánchez de Vega

As a rare disease patient herself, and the mother of a son of the same condition, Rosa Sánchez de Vega is a truly remarkable woman who has successfully managed to channel her difficulties and struggles with Aniridia into a positive force for change. She founded the Spanish Aniridia Association, for which she served as president until 2008. In 1999, she went on to co-found the Spanish Alliance for Rare Diseases (FEDER), serving first as Vice President and then President until 2010. Rosa joined the EURORDIS Board of Directors in 2003 and has served as Vice President from 2006 until 2013. She has been recently appointed President of the European Federation of Aniridia, Aniridia Europe. Ms Sánchez de Vega is an incredibly deserving recipient of the Volunteer Award as she has worked tirelessly and selflessly, often putting the rare disease cause beyond her own needs, in order to shine a light on rare diseases and improve the lives of others.

Media Award – Peter O'Donnell

Peter O'Donnell is a prominent writer and editor in the rare disease field currently working as Associate Editor of the European Voice. His impressive career has spanned over twenty years and various countries and has included working for prestigious newspapers such as The Financial Times, The Sunday Times, Reuters, the Economist Intelligence Unit and United Press International. He has worked as an editor, editorial adviser and speechwriter for numerous clients in the corporate, political and academic world and has frequently chaired EU-level policy debates and lectures on EU affairs. This has made him very well placed to be able to write and report forthrightly on the various complicated and rapidly-evolving issues surrounding rare diseases such as policy, at which he is incredibly skilled.
In October 2013 he spoke at the Lunch Debate on Data Protection at the European Parliament. It is for these reasons that EURORDIS is delighted to award him this year's Media Award.

Patient Organisation Award – Children with SMA – Vitaliy Matyushenko

"Children with SMA" is a voluntary, non-profit foundation which has undertaken the incredibly difficult mission of supporting those affected by, or involved with, Spinal Muscular Atrophy (SMA) in Ukraine. Among genetic diseases, it is a leading cause of death among children under age two. For almost 10 years, Children with SMA has worked tirelessly on behalf of those who suffer from SMA, promoting knowledge around the disease and encouraging dialogue between legislators, doctors, researchers and patients. This incredibly well-deserved award serves to recognise the efforts and successes that have resulted from the hard work and determination of Children with SMA in Ukraine such as its contribution to the adoption of the law for Rare Diseases in Ukraine in April of this year and the foundation of the Ukrainian National Alliance.

Company Award – Pfizer, Inc.

Pfizer is one of the world's premier pharmaceutical companies, and has demonstrated commitment to the rare disease cause - with 22 approved products to treat rare diseases worldwide including 4 in Europe. In 2010, Pfizer established its own Rare Disease Research Unit (RDRU), with the objective of taking an innovative and collaborative approach to the development of new medicines to create novel therapeutics across the spectrum of rare diseases. The current pipeline includes clinical and pre-clinical programmes in several rare diseases including sickle cell disease, haemophilia, muscular dystrophies, cystic fibrosis, and more. Through this award, EURORDIS recognises the role major pharmaceutical companies can play in the development of, and ensuring access to, innovative treatments for rare disease patients. The award also encourages companies to maintain a high level of corporate social responsibility by reassessing the value of medicines today so as to ensure that unmet medical needs are covered in the development of new treatments.

Scientific Award – Professor Kate Bushby

Professor Kate Bushby (MD FRCP) is a Professor of Neuromuscular Genetics and currently holds joint appointments between Newcastle University and the NHS. Her commitment to research in rare diseases, in particular inherited neuromuscular diseases, has been evidenced through her impressive publication list, clinical activities and involvement in policy actions. Prof Bushby is actively involved in many European projects including being a founding co-ordinator of the TREAT-NMD. Kate has played a leading role in the European and national rare disease policy area, acting as vice chair on the EUCERD from 2010 to 2013, which was mandated to assist the European Commission in the implementation of rare disease activities in all member states and she still acts in the capacity of invited expert on the new Commission Expert Group on Rare Diseases. In November 2014, Kate Bushby launched the John Walton Muscular Dystrophy Research Centre. The Centre will focus on three key research areas: translational research, innovative clinical trials and international networking. Kate has continuously demonstrated her true commitment to patients at many levels.

Lifetime Achievement Award – Abbey Meyers

Abbey Meyers is an extraordinary woman: once a housewife and mother from Connecticut, USA, Meyers was drawn into the world of political advocacy, fundraising and organisation development when it became painfully apparent through her experience as the mother of a child with Tourette syndrome, that patients with rare diseases were being neglected in favour of more common diseases that affected larger patient populations. Meyers founded the National Organization for Rare Disorders (NORD) in the USA, organisation dedicated to helping people with rare diseases and to the identification, treatment and cure of rare diseases through education, advocacy, research and service. Indeed, NORD was the precursor and inspiration for the creation of EURORDIS. This Lifetime Achievement Award serves to honour all that Ms Meyers has done for the rare disease community in the USA and throughout the world, her personal and tireless dedication to the cause and her instrumental role in the passage of landmark policies such as the Orphan Drug Act of 1983, which has served as the model for rare disease legislation beyond the USA.

2014

Policy Maker Award – Antonyia Parvanova

Dr Antonyia Parvanova, Member of the European Parliament since 2009, has shown an outstanding commitment to rare diseases: Dr Parvanova has since 2007 advocated for a 'Europe for Patients' emphasising the importance of improving access to medicinal products, and the benefits of cross border healthcare: Such relentless advocacy activity has allowed this topic to rise in the European political agenda leading to the adoption of the Directive on the application of Patient's Rights in Cross-border Healthcare. She worked as a paediatrician, expert and researcher in the field of healthcare management in the United Kingdom before launching her political career as an elected member of the Bulgarian Parliament in 2001 and then again in 2005. Alongside her persistent commitment to healthcare within the European Union, Dr Parvanova also strongly advocates for women's rights and gender equality, and to remove visa regulations imposed by the US and Canada upon Eastern European Countries.

European Rare Disease Leadership Award – Paola Testori Coggi

Paola Testori Coggi, biologist by education, was nominated as Director General for Health and Consumer Protection at the European Commission in 2010. In this position, she has facilitated European cooperation in the field of rare diseases through several important steps. For instance, the approval of the rare disease priority within the Health for Growth Programme; the adoption of a fundamental piece of legislation to facilitate patient mobility through the Directive on Patients' Rights in Cross-border Healthcare, crucial for rare diseases patients; as well as by taking the responsibility for the European Medicines Agency within her Directorate General.

European Rare Disease Leadership Award - Professor Guido Rasi

Professor Guido Rasi MD became Executive Director of the European Medicines Agency (EMA) in 2011 and has been pivotal in increasing the transparency of the agency's work. He has encouraged more dialogue between patients, heads of scientific committees, and health technology assessment (HTA) bodies, and promotes access to clinical trial data. Professor Rasi's overarching leadership and drive for collaboration between all rare disease stakeholders has undoubtedly facilitated the road to the authorisation of orphan products in Europe.

European Rare Disease Leadership Award - Professor Luca Pani

Professor Luca Pani MD, D.Psych undertook the role of Director General of the Italian Medicines' Agency (AIFA) in 2011. In this position he has emphasised the importance and urgency in creating orphan medicinal products, ensuring investment into rare disease research and granting the access to sustainable and successful treatments for those living with a rare disease. As a leader, Professor Pani has used initiative and experience to drive the equal access to cures for rare diseases, not only in Italy, but in Europe as a whole.

Media Award – Rick Guidotti

A graduate of New York's School of Visual Arts, Rick Guidotti led a successful career as a fashion photographer; every day he photographed what society deemed the most beautiful people in the world. This perspective was called into question after a chance encounter with a beautiful young girl living with the rare disease Albinism at a New York City bus stop. When he began to research the disease, he was troubled by the dehumanisation of people with rare diseases in medical text books. Since then, Rick Guidotti has launched a non-profit organisation, *Positive Exposure*, to change public perceptions of people living with genetic, physical and behavioural differences. The association runs educational and advocacy programs, organises exhibitions in public places and works with other NGOs and medical societies to give "positive exposure" to the beauty of those living with rare diseases. It is in the essence of his approach to changing the perception of beauty on a global scale for people with rare diseases that EURORDIS awards the EURORDIS Media Award 2014 to photographer Rick Guidotti.

Patient Organisation Award – Allianz Chronischer Seltener Erkrankungen

achse
Allianz Chronischer Seltener Erkrankungen

ACHSE represents 120 rare disease patient organisations in Germany and has been instrumental in strengthening the voice of rare diseases and turning Germany into one of the most committed Member States in the European Union for rare diseases in the fields of research, information, healthcare organisation and drug development. It has contributed to the progress NAMSE, which was presented to the German Health Minister in September 2013. The organisation is very active in raising awareness of rare diseases in Germany, having participated in Rare Disease Day since its launch in 2008, and by enlisting Eva Louise Köhler, the former First Lady of Germany, as an official patron of the organisation. ACHSE is exemplar of a valuable and effective platform for rare diseases, making them most deserving recipients of the EURORDIS Patient Organisation Award 2014.

sobi

Company Award – Sobi

Sobi, an international healthcare company dedicated to rare diseases, focuses on developing innovative treatments across four key therapeutic areas: haemophilia, inflammation/ autoimmune diseases, inherited metabolic diseases and oncology. Sobi is particularly sensitive to the need for targeted treatments that are accessible to paediatric populations. Sobi's track record of creating successful dialogue with patient communities demonstrates their objective for a transparent and progressive rare disease framework. Sobi is represented on several European Union committees such as the Commission Expert Group on Rare Diseases (replacing the EUCERD) and the working group on Mechanism of Coordinated Access to Orphan Medicinal Products (MOCA-OMP). This award recognizes the excellence and consistency of Sobi's work in the rare disease community.

Company Award – Orphan Europe

Orphan Europe, forming part of the Recordati group, has 25 years' experience in bringing orphan medicinal products to the market. Seven orphan products produced by Orphan Europe have been authorised to date and this rich portfolio of successful treatments is supported by the robust pipeline of medicines in development. Orphan Europe's support in the development of European Reference Networks EuroWilson, EPNET, E-IMD, E-HOD has helped to provide European guidelines for best care and treatment of rare disease patients. Strong alliances with patient organisations mark the central point of Orphan Europe's platform for therapy development, working alongside patient organisations for cystinosis, renal genetic diseases (AIRG), metabolic diseases and porphyrias. Orphan Europe also consistently supports networking and capacity-building for rare disease patient advocates via the EURORDIS Membership Meeting.

Lifetime Achievement Award – Marlene Haffner

For over 30 years, Dr Marlene Haffner has had an immeasurable impact upon the development of orphan drug therapies. Dr Haffner worked as the Director of the Office of Orphan Products Development at the United States of America Food and Drug Administration (FDA) for over twenty years, and she then applied her valuable knowledge and experience to assist the development of similar orphan drug programmes internationally. Dr Haffner's role in the FDA placed her in a unique position, bridging the gap between patient support groups and regulated industry with the common objective to develop successful orphan products. During her time at the FDA, over 300 products were brought to the market. These 300 medicines gave around 15 million people living with rare diseases in the USA alone access to treatment. After stepping down from the FDA, Dr Haffner spent two years as Executive Director of Global Regulatory Intelligence and Policy within Amgen, the largest biotech company in the world. She then founded Haffner Associates, of which she is now President. In this role, Dr Haffner applies her unmatched knowledge of the rare disease political landscape to consult and work together with patient advocacy groups and pharmaceutical and biotech companies of all sizes. She trained as an internist and haematologist, and spent five years as Director of the Office of Health Affairs at the center of Devices and Radiological Health. Due to Dr Haffner's admirable dedication within the field of Public Health, she rose to the rank of Rear Admiral in the United States Public Health Service (USPHS). The EURORDIS Lifetime Achievement Award is being awarded to Dr Marlene Haffner in recognition of her strong, lifetime dedication and commitment to addressing the needs of people with rare diseases.

Scientific Award – Professor Hans-Hilger Ropers

Hans-Hilger Ropers is Director at the Max-Planck-Institute for Molecular Genetics in Berlin and
Professor of Human Genetics at the Humboldt University. Dr Ropers has made many contributions to the molecular elucidation of monogenic disorders by positional cloning; his department forms part of the European MRX Consortium, and more recently, he has implemented very high resolution array CGH for the high-resolution detection of small unbalanced rearrangements in large cohorts of patients with monogenic and complex disorders. In total, Dr Ropers has published over 300 research articles. Between 1985 and 1993, H.H. Ropers served as Chromosome Chair and Co-Chair at several Human Gene Mapping Conferences. He is a member of HUGO since the year of its inception as well as member of the HUGO Council and the Human Genetics Meeting Scientific Program Committee. It is in recognition of Dr Roper's scientific excellence and untiring dedication to put single gene disorders into focus worldwide that we award Dr Hans-Hilger Ropers the EURORDIS Scientific Award 2014.

Volunteer Award – Lise Murphy

As an individual affected by the rare disease Marfan syndrome, Lise Murphy has had the experience this inherited disease across three generations, with her father and son also affected. In 2003, Lise Murphy was invited to be a member of the Board of Directors of the Swedish Marfan Organisation, (Svenska Marfanföreningen), and in 2004 became its Chairperson. Reaching beyond her own disease community, Lise Murphy has helped to catalyse the rare disease movement as a whole in Sweden, and for two years (between 2004 and 2006) she served on the board of the Swedish Rare Disease Alliance. Notably, from 2007 Lise Murphy represented EURORDIS at the European Medicines Agency's Patient and Consumer's Working Party (EMA-PCWP), and between 2010 and 2013 Lise Murphy held the responsibility of being co-chair of this working party. She has been instrumental in closing the gap between patients, health care professionals and pharmaceutical agencies, demonstrating the importance of patient dialogue and her unique brand of enthusiasm, energy and Swedish directness has added a special touch to all that she has done.

2013

Policy Maker Award – Françoise Grossetête

Ms Françoise Grossetête has been a Member of the European Parliament for almost 20 years. With her leadership as MEP and Rapporteur on several legislations, two essential EU Regulations have been adopted on Orphan Medicinal Products in 1999 and the Regulation on Medicines for Paediatric Use in 2006. Her interventions in favour of the EU Regulations on Advanced Therapy Medicinal Products and in the discussions around rare disease patients' mobility within the negotiations on the Cross Border Healthcare Directive, have always been instrumental in achieving the best possible outcomes for rare diseases patients. As a Member of the Committee on Environment, Public Health and Food Safety and as a Substitute of the Committee on Industry, Research and Energy, she has supported several amendments boosting research and securing Public Health projects in areas directly or indirectly making an impact on the rare disease field.

Volunteer Award – Lesley Greene

Lesley Greene is a true pioneer of the rare disease movement in Europe. In 1980, upon the diagnosis of her firstborn daughter at age 15 months, Lesley and her husband Peter Greene established the charity Research Trust for Metabolic Diseases in Children (RTMDC), dedicated to this group of disorders. RTMDC is now known as CLIMB (Children Living with Inherited Metabolic Diseases). In 1995, Lesley was invited to join Abbey Myers (Founder of NORD) in Brussels, as a patient representative, to discuss the feasibility of developing an orphan drug legislation in Europe. After which RTMDC collaborated with other patient groups across Europe to support the adoption of the Regulation in 1999. Lesley is still active with respect to the Orphan regulation via her role as patients' representative on the Committee for Orphan Medicinal Products, where she has served since 2009, and in her current position as Vice-Chair of the Committee since 2012. Lesley was elected as a Founder Director of EURORDIS in 1997 and from 2001 to 2003 she was EURORDIS' President.

European Rare Disease Leadership Award – Dr Ruxandra Draghia-Akli

Dr Draghia-Akli is Director of the Health Directorate at the Research & Innovation DG of the European Commission. She received an MD from Carol Davilla Medical School and a PhD in human genetics from the Romanian Academy of Medical Sciences. She also completed a doctoral fellowship at the University of Rene Descartes in Paris and a post-doctoral training at Baylor College of Medicine (BCM), Houston, Texas, USA, where she was also part of the faculty. She served as Vice-President of Research at VGX Pharmaceuticals (now Inovio) and VGX Animal Health. Her research activities have focused on molecular biology, gene therapy and vaccination. She is a global leader in the field of nucleic acid delivery for therapeutic and vaccination applications. Dr Draghia-Akli has demonstrated her commitment to research in the field of rare diseases via the framework programme FP7 and her unique leadership when launching the International Rare Disease Research Consortium (IRDiRC). The ambitious goal of this International Consortium is to develop 200 new therapies for rare diseases and the means to diagnose the most rare diseases by the year 2020.

Scientific Award – Dr Ségolène Aymé

Dr Segolene Ayme is a medical geneticist and Emeritus Research Director at the French National Institute of Health and Medical Research (INSERM). She developed Orphanet, the world's leading reference portal for expert validated rare disease and orphan drug information. Orphanet, funded by the INSERM, the French Ministry of Health, the AFM Téléthon and the European Commission (DG Public Health and DG Research), is considered the most comprehensive, reliable, up-to-date resource available for rare disease and orphan drug information. Available in six languages and with partners in 38 countries, Orphanet provides open-access data for 6,000 rare diseases – including clinical descriptions, related genes, research projects, patient organisations, medicinal products under development or approved, laboratory diagnostic services, centres of expertise, emergency guidelines, and more. Dr Ségolène Aymé also serves as Chair of EUCERD, heads the Scientific Secretariat of the IRDiRC, is Chair of the Topical Advisory Group for Rare Diseases, is responsible for revising the International Classification of Diseases at the World Health Organisation, and is Editor-in-Chief of the Orphanet Journal of Rare Diseases (www.ojrd.com). Dr Ségolène Aymé has contributed to dozens of scientific articles, participated in numerous rare disease-related projects and committees, and plays a key role in bringing the scientific expertise in National and in EU-level policies.

Patient Organisation Award – Alström Syndrome UK

Kay Parkinson is a unique leader who created an inspiring and exemplary patient organisation. After losing her two children because of late diagnosis of Alström disease, she studied law in order to better defend her children's interests when she launched the patient organisation. One of the key achievements of Alström UK is the development of patient led, NHS funded multi-disciplinary clinics for Alström Syndrome. Alström UK is a partner in the Euro-WABB project, an EU Rare Diseases Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes. The EURO-WABB Project is a collaboration of doctors, scientists and patient support groups from all over Europe. It is supported by the EU Directorate General for Health and Consumers (DG-SANCO) via its Executive Agency for Health and Consumers. The overall aim for this register is to be a key instrument to increase knowledge of these rare diseases, improve the lives of affected people through better management, and to develop clinical research. Alström UK is recognised by the EURORDIS Patient Organisation Award for its long-term commitment and outstanding achievements for Alström Syndrome patients.

Media Award – Andrew Jack

Andrew Jack has been a journalist for the Financial Times since 1990. Since 2004, he has specialised in health and pharmaceuticals, based in He was one of a group of journalists to be awarded the "1993 British Press Awards Reporting Team of the Year" accolade for coverage of the Robert Maxwell affair. Mr Jack has written articles for medical journals including the British Medical Journal and the Lancet. A geography graduate from St Catharine's College, Cambridge, Mr Jack was the Joseph Hodges Choate Memorial Fellow at Harvard University, Cambridge, Massachusetts; a New York City Government Urban Fellow; and a trustee of Pushkin House, a London-based centre for Russian culture. Mr Jack is being awarded the EURORDIS Media Award in recognition of his contribution to better the understanding of rare diseases and the issues surrounding these diseases through his articles written in the Financial Times during the past several years.

Company Award – Celgene Corporation

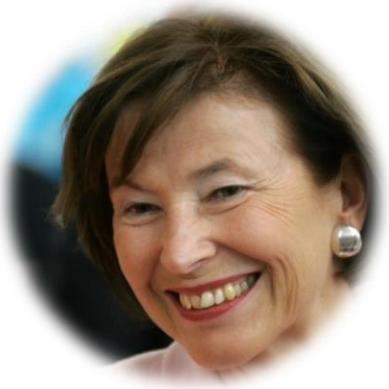
Since its inception in 1986, Celgene's leadership in the research, discovery, development and marketing of treatments for rare cancers has significantly improved conditions for rare disease patients. Their steadfast and significant reinvestment in research and development place it in a position to make a life-changing difference for thousands more rare disease patients in the future. The Company's commitment to innovation is reflected in the more than 200 clinical trials underway worldwide using compounds developed at Celgene. Celgene has created Celgene Patient Support® to assist patients worldwide in accessing products the Company has marketed in their respective countries. Celgene has a longstanding, supportive relationship with EURORDIS and other patients organisations in Europe and internationally. Most notably, the company supported the pilot and growth of RareConnect: the Online Patients Communities Project, which enables people affected by rare diseases to form communities across languages and geographic barriers.

Company Award – Prosensa

Prosensa has achieved several Orphan Drug designations and developed an impressive clinical portfolio in the short time since its founding in 2002. With the Company's commitment to "develop innovative, RNA based therapeutics to fill unmet medical needs for patients with genetic diseases," Prosensa has the potential to make a life changing difference for people living with rare diseases. Prosensa currently has several compounds in development for treating Duchenne Muscular Dystrophy (DMD), including a development in collaboration with GlaxoSmithKline for the development and commercialization of RNA based therapeutics for DMD. Prosensa has established partnerships with the patient community through strategic partnerships with muscular dystrophy focused patient organisations, including EURORDIS Members Duchenne Parent Project, Aktion Benni & Co e.v., and the AFM (French Muscular Dystrophy Association).

Company Award – Genzyme, a Sanofi Company

Genzyme is a pioneer in researching, developing and marketing medicinal products for patients living with diseases. Now part of Sanofi, Genzyme, a Sanofi Company continues to offer hope to people living with genetic diseases, endocrine and cardiovascular diseases. Strategies to increase access to Genzyme products include free drug programs and humanitarian initiatives. Genzyme began this practice in 1999, establishing the Gaucher Initiative, a humanitarian partnership to provide the Company's first product to Gaucher disease patients in developing countries. The Company is currently one of four co-funders of EURORDIS' work in the EpiRare project, a three-year project designed to address the need for rare diseases registration throughout Europe. Genzyme is also a long-time sponsor of EURORDIS Membership Meetings and an Emerald Member of the EURORDIS Round Table of Companies.



Lifetime Achievement Award – Eva Luise Köhler

As First Lady and now Former First Lady of Germany, Eva Luise Köhler has campaigned for the interests of people with chronic rare diseases and has taken over the patronage of the ACHSE. She is also the Chair of the Board of Trustees of the Eva Luise and Horst Köhler Foundation for people with rare diseases. Specifically, the Eva Luise and Horst Köhler Foundation supports research in the field of rare diseases. The foundation provides funding for basic and clinical research and awards 50,000 euros to a research team on an annual basis on the occasion of the official Rare Disease Day. The EURORDIS Lifetime Achievement Award is being presented to Mrs Köhler in recognition of her strong, lifelong dedication and commitment to addressing the needs of people living with a rare disease and for her contribution to the promotion of the rare disease cause in Germany.

2012

Patient Organisation Award - Association Française des Myopathies

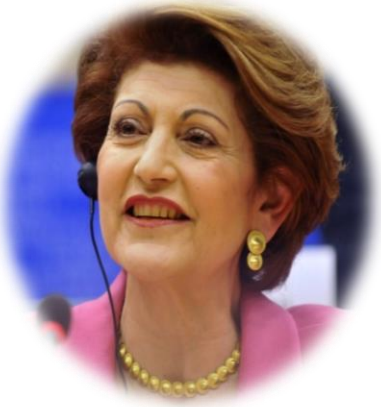
In recognition for its unmatched support for rare disease patients organisations throughout France and to EURORDIS, as well as outstanding success increasing awareness, raising funds and widely supporting research.

Volunteer Award - Michele Lipucci Di Paola, PhD

In recognition for his unshakable committed to improving conditions for people affected by rare diseases and particular dedication in Italy and on the European level.

European Rare Disease Achievement Award - Kerstin Westermark, MD, PhD

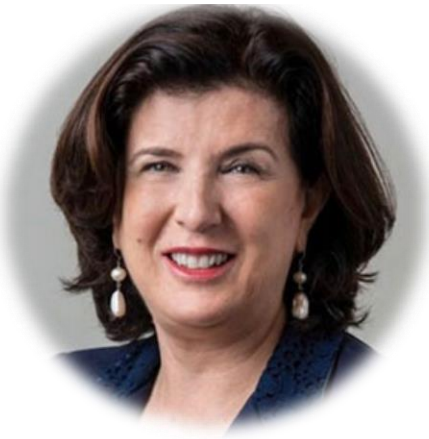
In recognition for her dedicated expertise and over a decade of support to those with rare diseases as the Swedish delegate and Chair of the Committee for Orphan Medicinal Products at the European Medicines' Agency.



Policy Maker Award – Andrea Vassiliou, JD

In recognition for her invaluable contribution to the rare disease community as Commissioner for Health and Consumer Policy from February 2008 to end of 2009. Her work to launch the Public Consultation, *Rare Diseases: Europe's Challenge*, the consequent adoption of the Commission Communication on Rare Diseases, and the Council Recommendation on Rare Diseases, has laid the groundwork to improve conditions for rare disease patients in all Member States of the European Union.

Scientific Award - Professor Alain Fisher and Professor Maria Grazia Roncarolo



As an outstanding example of scientific research and European collaboration that has resulted in the successful development of the first gene therapy for rare diseases related to Severe Combined Immuno Deficiencies, which has set the stage for the extension of gene therapy to other genetic diseases with a high unmet medical need and for which there is currently no cure

Media Award – BBC

In recognition of more than three decades of support raising awareness on rare diseases and covering rare disease issues from a patient perspective.

Company Award – CSL Behring

In recognition of the Company's long-standing commitment to rare disease patients, and most recently for its advances to benefit people living with primary immunodeficiency diseases and secondary immune-deficiencies.

Company Award – Shire

In recognition to Shire's pioneering initiatives, particularly for patients with Hunter Syndrome and Gaucher Disease, and for the company's on-going commitment to people living with rare diseases throughout Europe.

Company Award - Sigma Tau Pharmaceuticals, Inc

In recognition for being an early leader in developing medicines for rare diseases and for remaining an exemplary partner in the rare disease community.