AWARD RECIPIENTS 2012-2022

EURORDIS

Black Pearl

AWARDS

PREVIOUS BLACK PEARL AWARDEES

Members Award

Childhood Cancer International Europe - 2022

AKU Society UK – 2021

ALAN Maladies Rares Luxembourg - 2020

ALS Liga Belgium – 2019

Pulmonary Hypertension Association Europe – 2018

Dravet Europe – 2017

UNIQUE – 2016

Children with SMA – 2015

Allianz Chronischer Seltener Erkrankungen – 2014

Alström Syndrome UK – 2013

Association Française des Myopathies – 2012

Volunteer Award

Graham Slater - 2022

Ingunn Westerheim, Rebecca T Skarberg – 2021

Claudia Crocione – 2020

Richard West, Russell Wheeler – 2019

Chris Sotirelis, Helene and Mikk Cederroth – 2018

Elizabeth Vroom – 2017

Tsveta Schyns – Liharska – 2016

Rosa Sánchez de Vega – 2015

Lise Murphy – 2014

Lesley Greene – 2013

Michele Lipucci Di Paola, PhD – 2012

PREVIOUS BLACK PEARL AWARDEES

Policy Maker Award

Professor Hans-Georg Eichler - 2022

Kateřina Konečná – 2021

Dr Vytenis Andriukaitis – 2020

Dr Edmund Jessop – 2019

Elena Gentile – 2018

Frédérique Ries – 2017

Christian - Silviu Buşoi – 2016

Glenis Willmott – 2015

Antonyia Parvanova – 2014

Françoise Grossetête – 2013

Androulla Vassiliou, JD – 2012

Scientific Award

Prof. Franz Schaefer - 2022

Prof. Alain Hovnanian – 2021

Prof. Annemieke Aartsma-Rus – 2020

Professor Philip Van Damme – 2019

Professor Michele De Luca and Doctor Tobias Hirsch – 2018

Dr Lucia Monaco – 2017

Professor Dr Peter N. Robinson – 2016

Professor Kate Bushby – 2015

Professor Hans-Hilger Ropers – 2014

Dr Ségolène Aymé – 2013

Professor Alain Fisher and Professor Maria Grazia Roncarolo – 2012

PREVIOUS BLACK PEARL AWARDEES

Media Awards

Ewenflix - 2022

Ana Ilic (Written) – 2021

Noémie Desquiotz-Sunnen (Visual and Media) – 2021

Tomasz Śliwiński & Magda Hueckel
(Visual and Audio) – 2020

Anne-Dauphine Julliand (Visual and Audio) – 2019

Bojana Mirosavljević (Written) - 2019

Serge Braun, Christopher Ulmer – 2018

Aldo Soligno – 2017

France Télévisions - AFM – Télethon – 2016

Peter O'Donnell – 2015

Rick Guidotti – 2014

Andrew Jack – 2013

BBC – 2012

Company Awards

Lysogene (Innovation Award), Spinal Muscular Atrophy (SMA)-NBS Alliance
(Patient Engagement Award), Aparito (Health Technology Award) – 2022

Orchard Therapeutics (Innovation Award), Takeda (Patient Engagement Award),
Epihunter (Health Technology Award) – 2021

Healx (Innovation Award), Boehringer Ingelheim
(Patient Engagement Award), Tobea (Health Technology Award) – 2020

Chiesi (Innovation Award), The HERCULES Project
(Patient Engagement Award), Air Liquide Medical Systems
(Health Technology Award) - 2019

Novartis (Innovation Award), EFPIA, Bayer, UCB and MSD
(Patient Engagement Award) – 2018

GSK – 2017

Actelion – 2016

Pfizer – 2015

Sobi, Orphan Europe – 2014

Celgene Corporation, Prosensa, Genzyme – 2013

CSL Behring, Shire, Sigma-Tau Pharmaceuticals – 2012

PREVIOUS BLACK PEARL AWARDEES

European Rare Disease Leadership Award

Dr Anne-Sophie Lapointe - 2022

Dr Enrique Terol, Victoria Hedley, Dr Birutė Tumienė – 2021

Dr Daria Julkowska – 2020

Professor Till Voigtländer – 2019

Professor Bruno Sepodes – 2018

Antoni Montserrat Moliner, Jarek Waligóra,
Michael Hübel – 2016

Professor Josep Torrent-Farnell – 2015

Paola Testori Coggi, Professor Guido Rasi,
Professor Luca Pani – 2014

Dr Ruxandra Draghia – Akli – 2013

Kerstin Westermark, MD, PhD – 2012

Lifetime Achievement Award

Dr William Gahl - 2022

Milan Macek – 2021

Michael Griffith - 2019

Alastair Kent – 2018

Anders Olauson – 2017

Renza Barbon Galuppi – 2016

Abbey Meyers – 2015

Marlene Haffner - 2014

Eva Luise Köhler - 2013

Young Patient Advocate Award

Danielle Drachmann - 2022

Rachele Somaschini – 2021

Jana Popova – 2020

Laëtitia Ouillade – 2019

Sammy Basso – 2018

Social Media Award

Milica and Noa - 2022

2022

EURORDIS BLACK PEARL AWARDS

2022

MEMBERS AWARD

Childhood Cancer International Europe

The EURORDIS Members Award is awarded to Childhood Cancer International Europe (CCI-E), the largest umbrella-organisation for childhood cancer in Europe, counting 67 patient-organisations in 34 countries.

The Award celebrates the organisation's outstanding advocacy of childhood cancers on a national and international level, as well as their active commitment to fulfilling their role as patient advocates in the European Reference Network for Paediatric Cancer and several other EU projects such as Harmony, PanCare and Accelerate.

Thanks to the work of CCI-E and their collaborations with local and international partners they have enhanced the ability of many medical professionals and health workers to recognise the early signs of childhood cancer enabling them to make accurate and early diagnoses. This award recognises that CCI-E is at the forefront of the establishment of an 'EU network of youth cancer survivors', as outlined in the Europe's Beating Cancer Plan. CCI-E has also been extremely supportive of EURORDIS, notably in relations to the BECA report, and stands shoulder to shoulder with the rare adult cancer patients' community.

2022

VOLUNTEER AWARD

Graham Slater

The EURORDIS Volunteer Award 2022 goes to Graham Slater, one of the first survivors of Esophageal Atresia (EA), for his outstanding contributions and dedication to the rare disease community and to EURORDIS.

The Award acknowledges Graham's incredible commitment to rare diseases and his work as Board Member of TOFS (UK EA support group), Chair of the Esophageal Atresia Global Support Group (EAT), partner of the UK National Health Service's Clinical Reference Group for specialised surgery in children, as well as lead ePAG at the European Reference Network for rare Inherited and Congenital (digestive and gastrointestinal) Anomalies (ERN ERNICA).

The award also recognises Graham's involvement in different EURORDIS working groups, long commitment to international collaboration in the rare disease domain and his leadership to ensure that all rare disease patients, regardless of where they live, can benefit from the sharing of good practices and the development of new surgical techniques and improved life-long after-care.

2022

LIFETIME ACHIEVEMENT AWARD

Dr William Gahl

The EURORDIS Lifetime Achievement Award goes to Dr William Gahl for the lifelong dedication he has shown in addressing the needs of people living with rare and undiagnosed diseases. Dr William Gahl graduated from the Massachusetts Institute of Technology and earned his M.D. and Ph.D. from the University of Wisconsin. He served as paediatric resident and chief resident at the University of Wisconsin hospitals and completed clinical genetics and clinical biochemical genetics fellowships at the NIH. Dr Gahl elucidated the basic defects in cystinosis and Salla disease and helped bring cysteamine to new drug approval by the Food and Drug Administration as the treatment for cystinosis. He has published over 530 peer-reviewed papers, trained 42 biochemical geneticists and cultivated international experts for dozens of rare diseases, syndromes and disorders. In 2008, he established the NIH Undiagnosed Diseases Program (UDP), which has made more than 300 rare disease diagnoses and discovered 30 new genetic diseases. Dr Gahl expanded the UDP to a national Undiagnosed Diseases Network and a worldwide Undiagnosed Diseases Network International. He established the American Board of Medical Specialties certification for medical biochemical genetics. Dr Gahl received the Dr Nathan Davis Award for Outstanding Government Service from the AMA, the Service to America Medal in Science and the Environment, and numerous other awards. In 2019, he was elected to the National Academy of Medicine.

With this award, we would like to acknowledge the key role Dr Galh has played in the creation of the National Institutes of Health (NIH) Undiagnosed Diseases Network (UDN) and its essential role in discovering many new genetic disorders. His unwavering support of the rare and undiagnosed disease cause and his patient-centred approach is demonstrated by the leadership role he played in the development of the International Network on Undiagnosed Diseases (UDNI). This notable achievement is bolstered by the work being done by the UDNI which he spearheaded as the Chair; the programme provides an admirable example of an international institution which brings together the undiagnosed community at the global level. UDNI has provided a structure and forum to exchange on undiagnosed cases for clinicians working around the globe.

The importance of all his efforts in the field of rare disease research cannot be overstated, and we believe him to be a truly deserving recipient of this award.

2022

POLICY MAKER AWARD

Professor Hans-Georg Eichler

The EURORDIS Policy Maker Award is awarded to Prof. Hans-Georg Eichler MD, MS for his dedication to making a real difference in the rare disease community through his academia, institution and regulatory experience.

Hans-Georg is currently the Consulting Physician of the Association of Austrian Social Security Bodies. Before holding this position, he was the Senior Medical Officer of the European Medicines Agency for more than 14 years, and Professor and Chair of Clinical Pharmacology at the Medical University of Vienna. He held a range of other full-time and honorary positions in academia, industry and government.

This award acknowledges the role he played at the forefront of the campaign for clinical research to be more innovative in order to reduce the time and money it takes for a drug to come to the market. In his role as Senior Medical Officer of the European Medicines Agency he showed incredible leadership advocating for drugs licensing.

The award also recognises his active engagement with the Clinical Trials Transformative Initiative (CTTI), his commitment to making clinical research more global and bringing information to those who need it most and the active role he has played within EURORDIS by co-chairing the 32nd ERTC workshop and being a Programme Committee member for ECRD 2022.

2022

EUROPEAN RARE DISEASE LEADERSHIP AWARD

Dr Anne-Sophie Lapointe

The EURORDIS European Rare Disease Leadership Award 2022 is awarded to Dr Anne-Sophie Lapointe for the outstanding leadership and dedication shown to the rare disease community and the positive impact she has made in advancing rare disease policy and partnerships both at a national and international level.

Dr Lapointe's involvement in the rare disease field started in 1998 as a caregiver of two boys living with a rare disease. She then served as President for the French Lysosomal diseases association, Vaincre les Maladies Lysosomales, and as a board member at EURORDIS and the French national rare diseases alliance for 6 years.

Dr Lapointe holds a PhD in bioethics and she has worked for several years with geneticists at Necker Hospital and the ERN ITHACA with the French rare diseases network AnDDI-Rares. She developed an expertise in the genomic area, collaborating with human and social research teams around programmes linked to incidental findings and patient consent. She was also involved as a board member in the Inserm Ethics Committee (IEC) with a constant dialogue between the scientific and medical research community and society.

In 2016, alongside Ana Rath from Orphanet, she drafted the third national plan for rare diseases. Her domain was "information, training and e-health". The third plan, launched the 4th of July 2018, is the result of the concerted efforts of all the stakeholders committed to promoting an ambitious health and research policy designed to help patients with rare diseases and their families. In October 2018, she joined the French Ministry of Health as project manager of the rare diseases mission and with her team she works closely with the Ministry of Research to undertake the third national plan with its 55 actions.

This award celebrates her determination and engagement, as well as her active involvement in advocating for rare diseases.

2022

Award selection made in Partnership with **Orphanet**

SCIENTIFIC AWARD

Prof. Franz Schaefer

The Scientific Award is awarded to Prof. Franz Schaefer for his scientific excellence, outstanding dedication to the rare disease community and the positive impact he has made in rare disease research and patient communities on an international level.

Prof. Schaefer is Professor of Pediatrics and Chief of the Pediatric Nephrology Division at Heidelberg University Hospital. He received his M.D. in 1986 at Würzburg University Medical School. He performed research scholarships at the Institute of Child Health, London, the University of Virginia and Stanford University and he has also served on the boards and councils of numerous medical societies. He is the current President-Elect of the International Pediatric Nephrology Association.

Prof. Schaefer has a special interest in rare kidney disease research. In 2009 he established the eRare-funded PodoNet Project for Research in hereditary and immune mediated steroid resistant nephrotic syndrome. The PodoNet Registry has become the world's largest database for this group of rare glomerulopathies and has led to the identification of new genetic disease entities and prognostic biomarkers. Since 2021 Prof. Schaefer has contributed his expertise in clinical data management by contributing to the development of a rare disease data ecosystem as Pillar co-lead in the European Joint Programme for Rare Diseases (EJP RD) and since 2021 by leading the data integration workpackage of ERICA, the ERNs' research coordination programme. Prof. Schaefer's publication record encompasses 650 scientific articles and book chapters.

We would like to acknowledge his leadership and outstanding work in the ERN Coordinators Group and in several of the cross-ERNs Working Groups, as well as his prominent role in rare disease Registries and his innovative approach in the use of health outcome measures to drive improvements in care and his work within the European Joint Programme on Rare Diseases. It is thanks to his scientific leadership, research and collaborative spirit that key achievements have been made in the field of rare kidney diseases, paediatric nephrology and hypertension. His dedication, engagement, collaboration with scientists, clinicians and patients cannot be overstated, and we believe him to be a truly deserving recipient of this award.

2022

YOUNG PATIENT ADVOCATE AWARD

Danielle Drachmann

Danielle Drachmann founded Ketotic Hypoglycemia International (KHI) - the world's largest patient organisation for patients with idiopathic (unexplained) ketotic hypoglycemia - after failing to get a diagnosis, despite extensive clinical and genetic investigations, on her two children's dangerously low blood glucose (sugar) and high ketone levels. This international patient association works in close collaboration with leading medical experts from all over the world and has initiated patient-driven research projects later disseminated in scientific publications. The organisation is also very active on its social media platforms, where it strives to unite the families in an online community, so they can support, guide and help each other navigate the life living in a ketotic hypoglycemia rollercoaster.

Drachmann also recently entered the European Health Parliament and the European Medicines Agency as a Patient Representative for patients with rare hypoglycemia diseases. She is working at the Center for Research with Patients and Relatives at Odense University Hospital, Denmark, while also sitting in the Research Committee at H.C Andersen's Children's Hospital, Odense University Hospital, Denmark, as a patient and relative representative.

Danielle's outstanding resilience in the face of the medical mystery, idiopathic ketotic hypoglycemia (IKH), combined with her perseverance in initiating patient-driven research with leading medical experts from different fields of medicine, has been an extraordinary example of how we envision the future of rare disease research:
Passionate, patient-driven and co-created.

2022

SOCIAL MEDIA AWARD

Milica and Noa

Milica (Serbia) is the mother of a little hero and an amazing girl called Noa. Noa was born prematurely at 26 weeks, weighing only 700g. Due to the complications after her birth, she had 3 brain bleeds, and they developed into an extreme Hydrocephalus that left 80% of her brain damaged. Up to now, Noa has had 18 brain surgeries, and she had some other diagnoses due to hydrocephalus, like severe epilepsy and cerebral palsy, and about 10 more diagnoses. But Noa never let her rare condition dictate her life. She is now 9 years old, almost walking, she still has to learn to talk but she knows gestures and understands a lot now, even though the diagnoses meant she had a chance of survival of less than 1%, she is making more out of her 1% than most would have thought possible.

Through social media (mainly Instagram) Milica tries to educate and bring people closer to the life of a child with special needs. There is no room for negativity on their social media: you can only find beautiful and inspirational things like therapies, cute stories and Noa's everyday life. Noa's story provides a source of inspiration for anyone reading it via Milica who's "just her voice until she finds her own and continues to inspire with her own words".

Learn more about Milica and Noa here

2022

MEDIA AWARD

Ewenflix

The EURORDIS Media Award is awarded to the EwenLife on-demand video platform for offering all people living with a rare disease and their families free and accessible online tools to break through the sense of isolation frequently going hand in hand with rare diseases.

The platform hosts videos where patients, parents and caregivers talk about how rare diseases affect their everyday life - and share some personal tips or advice. What is also noteworthy about the project is the abundant sense of optimism which is prevalent in the interviews, seminars/web series and filmed testimonies which the platform hosts.

The award also recognises the needs of people living with a rare disease and brings them to the attention of a large audience as well as simultaneously forging bonds of solidarity among the rare disease community.

2022

COMPANY AWARD FOR PATIENT ENGAGEMENT

Spinal Muscular Atrophy (SMA)-NBS Alliance

The EURORDIS Company Award for Patient Engagement recognises the collaborative effort of the companies involved in the European Alliance for Newborn Screening in SMA to ensure that newborn screening programmes in all European countries diagnose all newborn children with SMA. This close collaboration reinforces the demand of the rare disease community for a harmonized approach to newborn screening in Europe.

The achievements of the Alliance, which is succeeding in creating much-needed awareness and momentum towards an accelerated implementation of newborn screening for SMA in an ever-growing number of European countries, merits acknowledgement. The Award also recognises how the Alliance has set a model example which has the potential to encourage similar initiatives across other rare diseases.

EURORDIS truly appreciates the companies' mutual engagement with patient advocates and multiple stakeholders around a single cause, which further reflects their commitment for and solidarity with the rare disease community.

SMA NBS Alliance

2022

COMPANY AWARD FOR HEALTH TECHNOLOGY

Aparito

The EURORDIS Company Award for Health Technology recognises companies developing technologies with significant positive impact on the daily lives of people living with a rare disease.

EURORDIS commends Aparito as a company at the forefront of innovative technologies that report patient experiences accurately. By placing patient needs at the heart of the solution, Aparito's expertise facilitates medicines development and contributes to addressing complex regulatory challenges.

Aparito's collaborative approach, working across rare diseases and alongside patient organisations, clinicians as well as sponsors, also deserves recognition. With the ever-increasing pace of developments in science and technology, Aparito's remarkable journey in the field of remote patient monitoring is testament to the potential of digital solutions to profoundly impact the delivery of healthcare for people living with a rare disease.

aparito

2022

COMPANY AWARD FOR INNOVATION

Lysogene

The EURORDIS Company Award for Innovation acknowledges companies undertaking ground-breaking activities to advance rare disease research and medicines development. EURORDIS recognises Lysogene as a pioneering gene therapy company which has established itself among major players driving European-led innovation, in just over ten years. We are particularly encouraged by Lysogene's promising advances towards delivering novel treatments in neuro degenerative and neuro developmental disease areas with high unmet medical needs.

Lysogene's continued readiness to advocate for early diagnosis and engage patients and caretakers as equal decision makers in the product development lifecycle also merits acknowledgement. The award also celebrates the remarkable personal journey of the company founder, Karen Pignet-Aiach, as a mother, patient advocate and entrepreneur determined to change the landscape for severe rare diseases affecting children's central nervous systems.

2022

EURORDIS PHOTO AWARD

1st by Catarina Costa

Smiling underwater

Angelman Syndrome

Portugal

2nd by Lucía Lamat

Dalmatian(s)

Congenital Melancoytic Nevus

Spain

3rd by Emiliano Cribari

Federico

Trisomy 9 Mosaic

Italy

2021

EURORDIS BLACK PEARL AWARDS

2021

MEMBERS AWARD

AKU Society

The EURORDIS Members Award is awarded to the AKU Society for their 17 years of outstanding advocacy and research in the rare disease field.

The AKU Society is a UK-based patient group that helps people with the ultra-rare genetic disease Alkaptonuria (AKU or Black Bone Disease) and that supports research into treatments and a cure. Patients suffer from severe joint deterioration, heart problems and a host of other issues as life progresses, leaving most of them very disabled.

The AKU Society was founded in 2003 by AKU patient Bob Gregory and his doctor Professor Ranganath from the Royal Liverpool and Broadgreen University Hospitals. They were soon joined by Nick Sireau, whose two sons were born with the disease in 2000 and 2003. When he realised there was little to no support for AKU patients, Nick left his job to join the AKU Society and dedicate his life to one goal: cure his sons' AKU.

AKU
Alkaptonuria Society

2021

MEMBERS AWARD

AKU Society

Over the next 17 years with Nick's unique leadership and drive, the AKU Society expanded upon the existing knowledge of AKU, sponsoring ground-breaking research and raising money for innovative patient support. The AKU Society became aware of a drug called nitisinone that was thought to stop the symptoms of the disease in their tracks. This culminated in 2012 with the National Health Service funding the first ever dedicated centre for AKU. The National Alkaptonuria Centre, based in Liverpool, not only offers yearly health checks and advice on how to live with AKU but also provides nitisinone off-label to all patients in England and Scotland.

In 2012 the AKU Society was also awarded funding from the European Commission to found a consortium made up of hospitals, pharmaceutical companies and universities from across Europe, called DevelopAKUre. When the trials ended in 2019 they had shown that nitisinone was safe for use in treating AKU and reduced the harmful homogentisic acid by 99.8%, with a statistically positive impact on the clinical symptoms of the disease. In 2020, the European Commission formally licensed the drug for use in AKU across Europe.

Thanks to the work of the AKU Society, no one born with AKU in Europe will ever face a future of pain and disability again.

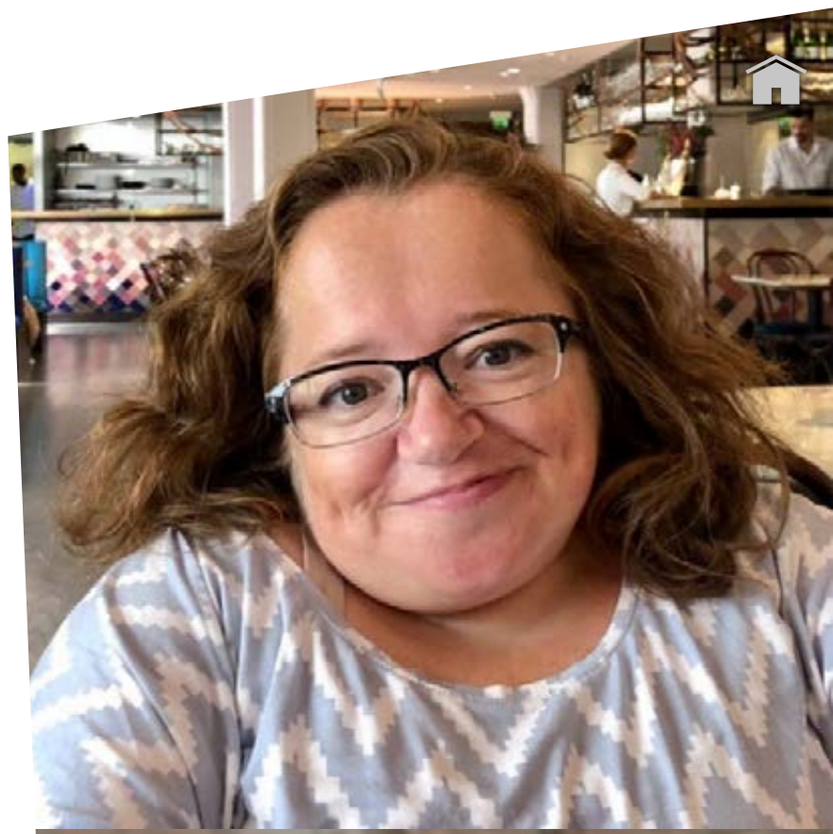
2021

VOLUNTEER AWARD

Ingunn Westerheim, Rebecca T Skarberg

The EURORDIS Volunteer Award recognises the outstanding contributions and dedication that Ingunn Westerheim and Rebecca Tvedt Skarberg have shown in the rare disease community and to EURORDIS as volunteers. It acknowledges their commitment to rare diseases with the Osteogenesis Imperfecta Federation Europe (OIFE), several EURORDIS activities, the European Joint Programme, Rare Disease Day, Rare Disease Week, Bond ERN and many others.

This award also recognises their work as EURORDIS volunteers and all their numerous achievements in the field. Their determination to travel all over the world to participate at meetings to raise awareness of rare diseases further proves their commitment to the cause.



2021

LIFETIME ACHIEVEMENT AWARD

Milan Macek

The EURORDIS Lifetime Achievement Award recognises the lifelong dedication Milan Macek has shown in addressing the needs of people living with a rare disease in the Czech Republic and in Europe. It acknowledges his commitment to rare diseases within EUCERD, Rare 2030, the European Society of Human Genetics, the CF Network, Orphanet, and more. Perhaps most notable is chief advisor of the Czech EU Presidency under which the EU Council recommendation on an action in the field of rare diseases was adopted in June 2009 – a soft legislation that has served as the basis for the tremendous progress in the field over the last decades.

This award also recognises his current engagement in the Rare 2030 Research Advisory Board and his key role in leading progress for the Rare Disease Community in the Czech Republic, particularly his efforts to raise awareness on the importance of keeping rare diseases a European public health priority with colleagues in the new Czech EU Council Presidency team. Milan Macek's lasting support to EURORDIS activities, notably the ECRDs, is highly appreciated.

2021

POLICY MAKER AWARD

Kateřina Konečná Member of the European Parliament

The EURORDIS Policy Maker Award recognises Kateřina Konečná's dedication into making real differences in the rare disease community. In her role as a member of the European Parliament she has shown incredible leadership and excellence in championing the rare disease cause in Europe.

As a Member of the European Parliament (Czech Republic), Kateřina Konečná is a member of the ENVI Committee where she focuses on access to healthcare and a number of public health issues often addressing inequalities across EU countries. In this context, she has been a champion for the rare disease community bringing awareness on patient engagement in HTA or on increased funding for ERNs.

In the previous term, she was actively engaged in activities of the network of Parliamentary Advocates for Rare Diseases, and in the current term, she is a signatory of the Pledge4RD. She is also engaged in the European Parliament Working Group on innovation, access to medicines and poverty-related diseases, and the Interest Group on Patient Access to Healthcare, both of which she co-chairs. At national level, Ms. Konečná also works closely with the rare disease patient alliance in her home country and the national centre for rare diseases.

2021

EUROPEAN RARE DISEASE LEADERSHIP AWARD

The EURORDIS European Rare Disease Leadership Award 2021 recognises the outstanding leadership and dedication shown to the rare disease community by Victoria Hedley, Dr. Enrique Terol and Dr. Birutė Tumienė. Their work has been crucial for the development of the European Reference Networks and has greatly contributed to stronger European cooperation in the rare disease field.

Victoria Hedley

Victoria has been an active participant in the generation of rare disease policies and policy-related outputs since 2012. She currently leads the knowledge-based activities of the Rare 2030 foresight project and previously led the policy side of RD-ACTION, the EU Joint Action for Rare Diseases. She has experience and understanding of a wide range of topics under the Rare Disease umbrella and belonged to the coordination team for the EUCERD Joint Action for RD. In these capacities she contributed to the development of EU-level Recommendations around topics such as Cross-Border Genetic Testing; the Incorporation of Rare Diseases into Social Services and Policies; Patient Registration and data collection; National Plans and Strategies; and, most prominently, European Reference Networks. She led RD-ACTION support for the conceptualisation and implementation of European Reference Networks (ERNs), and designed key workshops to assist the ERNs in addressing shared policy challenges.

For the past 5 years she has led the resource on the 'State of the Art of Rare Diseases Activities in Europe'. On the research side, Victoria co-leads the data-related activities on conect4children and is a seed member of the GO-FAIR Implementation Network for Rare Diseases. She is now co-lead of the new Newcastle Centre for Rare Diseases, which she brought to fruition in 2020, and which is now consolidating the broad range of disease-focused and methodological expertise in the Newcastle region, to optimise patient-centred research and innovation.

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2021

EUROPEAN RARE DISEASE LEADERSHIP AWARD

Dr. Enrique Terol

Enrique Terol MD, specialized in Family and Community Medicine, MSc and PhD in Public Health. His professional experience in Spain includes the clinical practice as specialist in Family and Community Medicine, implementation of programmes of public health, managerial positions as CEO or Medical Director of Primary and Specialised Healthcare institutions and healthcare systems planning, organisation and evaluation.

Enrique Terol works as Policy Officer in DG SANTE unit B3 of the European Commission and is in charge of the implementation of the European Reference Networks (ERNs) under the framework of the Directive of Cross-border Health care since 2011. He was Deputy General Director of Quality and Health Planning of the Ministry of Health of Spain between 2004 and 2008 and was in charge of the development of the Spanish Strategies on Rare Diseases, Cancer, Diabetes, Mental Health and Patient Safety. He was also involved in the design and implementation of the Spanish System of Centres of Reference for rare and complex diseases.

EUROPEAN RARE DISEASE LEADERSHIP AWARD

Dr. Birutė Tumienė

Dr. Birutė Tumienė is a clinical geneticist by background.
She graduated Faculty of Medicine in Vilnius University, where she also had her residency training in clinical genetics and PhD studies in rare genetic epilepsies; currently in her Alma Mater she gives lectures on genetics and rare diseases. Besides, she is the Head of Unit for Genetic counselling in the Centre for Medical Genetics, and a Coordinator of International Affairs in the Coordination Center for Rare Diseases at Vilnius University Hospital Santaros Klinikos.

Her international rare diseases-related activities are as intensive, with a track record of 14 years. Currently, she is a National Coordinator of Orphanet Lithuania, Operating Group member and Pillar 3 co-leader in European Joint Program on Rare Diseases and an Advisor to the WHO Special Envoy for European Region. Besides, she is Panel of Experts member in the foresight study Rare2030, Advisory Committee member in the European Rare dIsease research Coordination and support Action ERICA and Lithuanian Representative in the European Reference Network Board of Member States (ERN BoMS).

In the ERN BoMS, she participates in the WG on Research and chairs a Working Group on ERN integration into national systems.
Her activities also extend to professional/scientific organizations: she is a Board member of European Society of Human Genetics, one of the founders of Baltic Society of Inherited Metabolic Diseases.

Through all these activities, Dr. Tumienė fosters manifold special interests in rare diseases, that combine perspectives of a professional, lecturer, policy-maker and an advocate of rare disease patients.
She comes from a small newcomer EU13 country; hence, her particular concerns about equity, right to health and accessible, quality care across Europe, and beliefs in common European values and collaboration as a way to solve pan-European rare disease problems.

2021

SCIENTIFIC AWARD

Prof. Alain Hovnanian

The EURORDIS Scientific Award recognises Prof. Alain Hovnanian's scientific excellence, dedication and collaborative approach to working with the rare disease patient community, as well as the positive impact he has made on rare disease research, particularly with Epidermolysis Bullosa and Pachyonychia Congenita.

Alain Hovnanian is a physician-scientist board certified in Genetics and in Dermatology at Necker hospital in Paris. From 1993 to 2000, he joined the Wellcome Trust Centre for Human Genetics at the University of Oxford.
He developed positional cloning approaches and successfully identified the genetic bases of several orphan skin diseases. Since 2009, he is professor of Genetics at Necker hospital for sick children in Paris.

He runs a translational research activity focused on genetic skin diseases, has bi-weekly consultations and runs an INSERM research laboratory dedicated to the diagnosis and treatment of genetic skin diseases. His laboratory ("Genetic skin diseases: from mechanisms to treatments") is part of the new Imagine Institute for genetic diseases in Paris. His laboratory aims at bringing innovative and targeted treatments to people suffering from severe genetic skin diseases.

For years, Dr Hovnanian has worked to find a cure to RDEB, After identifying the gene responsible for the disease in 1993, he also identified the genetic basis of Darier disease, Hailey-Hailey disease, Netherton syndrome (NS) and a subset of Olmsted syndrome. These breakthroughs were essential to better understand the pathogenesis of these diseases and to develop specific treatments.
Through the development of innovative medicinal products or repurposed medicine, Dr Hovnanian research has significantly contributed to bring new treatments for several orphan skin diseases.

2021

YOUNG PATIENT ADVOCATE AWARD

Rachele Somaschini

Rachele was born in Milan, Italy, in 1994. Soon, she was diagnosed with Cystic Fibrosis, threatening the likelihood to reach adulthood.

Today, if you google Rachele Somaschini you get plenty of results regarding her career in motorsports. She managed to become a National Rally Champion and she is known as a relentless advocate for Cystic Fibrosis, via her fundraising project #CorrerePerUnRespiro (Italian for "Racing for a breath").

The success of the project and her collaboration with different patient organisations has brought the needs of people living with Cystic Fibrosis to a wider audience. Thanks to #CorrerePerUnRespiro Rachele has raised over 200 000 euros for the Italian Cystic Fibrosis Research Foundation (Fondazione Ricerca Fibrosi Cistica). In addition to fundraising, the other main goal of the project is to increase awareness about the condition, by means of an unfiltered narrative that only a young person living with the disease is able to provide, through daily struggles with therapies, hospitals, limitations and precautions in her routine.

Always backed up by her family, Rachele sticks to her treatment and therapies with impeccable responsibility, doing all she can to preserve her physical conditions and yet living life to the fullest.
It was thanks to her father Luca that she learnt to love speed and racing. Successfully sharing her life with Cystic Fibrosis, from food choices to her travel bags full of medicament and tools for therapies, Rachele counts on thousands of loyal followers participating with her in the most important races of all: beat Cystic Fibrosis thanks to scientific research.

2021

VISUAL & AUDIO MEDIA AWARD

Noémie Desquiotz-Sunnen

Noémie Sunnen was born in Luxembourg. She studied classical and lyrical canto at the Music Conservatory of Luxembourg and at the Conservatoire National Régional de Nancy. She obtained several prix supérieurs and other prizes. She has performed several operatic roles in Luxembourg, including Queen of the Night in the Magic Flute by W.A.Mozart. She has also appeared at the opera company in Trier, Germany.

For several years now she has performed as a soprano soloist at a number of concerts and recitals together with various orchestras and choirs. Her repertoire ranges from baroque and classical music to the romantic music of the 19th century, as well as contemporary music of the twentieth and twenty-first centuries, including Lied, oratorios and opera.

Noémie is suffering from motor-neurone disease ALS (Amyotrophic lateral sclerosis), but she still continues singing and has organised several concerts to raise awareness about the disease and gather funds for research.

2021

WRITTEN MEDIA AWARD

Ana Ilic - Poetry Collection

Ana Ilić is a poet, a passionate advocate for social inclusion and a public activist. Ana is from Serbia and lives with Friedreich's Ataxia, which she challenges everyday through words. She uses poetry as a portal for channelling her thoughts allowing her audience to join her in her journey as a person living with a rare disease. The unique and raw nature of her poetry has allowed greater and meaningful public understanding of the true realities experienced by the rare disease community.

Ana graduated at the department of philosophy at the University of Nis in 2019 and also studied at the IT academy. She is the author of three notable poetry books and award-winning poems included in several poetry anthologies. Her poems have been published in professional magazines, translated into English and Polish, read on television and radio shows with the participation of famous Serbian public figures, published at about 30 literary events throughout Serbia, at the Belgrade Book Fair and at the largest book fair in the world, in Frankfurt. With the support of the diaspora, the first book of poems was promoted in Los Angeles and Toronto. Ana has participated in more than 100 interviews over the past few years and is a true representative of the Serbian community for rare diseases.

Every day, Friedreich's ataxia occupies a small part of her body, making it increasingly difficult for her to live normally. Although in a wheelchair, with serious cardiomyopathy, insulin life, skeletal deformities, she amazes us with her incredible energy, love for art and philosophy, constant desire for new knowledge and search to find answers to the most important life questions. Today, Ana works as a graphic designer in Nis, where she lives and creates, and is a student of the fifth generation of the Academy of Democracy, a prestigious CRTA, a civil society organization dedicated to the development of democratic culture and civic activism.

2021

COMPANY AWARD FOR PATIENT ENGAGEMENT

Takeda

The EURORDIS Company Award for Patient Engagement recognises Takeda's commitment to collaborative and constructive engagement with the rare disease patient community.

EURORDIS particularly appreciates the Company's long-standing and consistent support, helping to amplify rare disease patients' voice across diseases and borders.

A company striving to change the landscape of rare diseases, Takeda has been exploring innovative solutions to stimulate multi-disciplinary, patient-focused collaboration on critical issues affecting rare disease patients in Europe and beyond – from advocating for timely and accurate diagnosis and equitable patient access to rare disease therapies, through to encouraging multi-stakeholder dialogue on real-world evidence. Among the first companies signing the EURORDIS Charter for Clinical Trials in Rare Diseases in 2010, we also acknowledge Takeda's approach to integrating patient insights into medicines' development during their life-cycles.

Takeda's over eighty-year legacy in rare diseases is yet another reflection of the Company's dedication to improving the lives of people with rare and complex conditions.

2021

COMPANY AWARD FOR HEALTH AND TECHNOLOGY

Epihunter

The EURORDIS Company Award for Health Technology recognises companies developing technologies with significant positive impact on the daily lives of people living with a rare disease. We commend Epihunter for creating an affordable and accessible digital solution that helps people with absence epilepsy to live more confident, fuller lives.

We are encouraged by the strength of the technology to empower people to manage their condition better and have meaningful interactions with those around them. Epihunter's innovative approach also contributes to raising much-needed awareness on a silent and invisible condition that is all too often misunderstood. It is testament to the potential of technology to help people living with rare and complex diseases overcome feelings of isolation.

Through detecting, signaling and recording absence seizures, Epihunter's device provides clinicians and researchers complete and accurate data, which is key for improving care and advancing knowledge of absence epilepsy.

2021

COMPANY AWARD FOR INNOVATION

Orchard Therapeutics

The EURORDIS Company Award for Innovation recognises companies undertaking ground-breaking activities to advance rare disease research and treatment development.

EURORDIS would like to acknowledge Orchard Therapeutics' commitment to addressing areas of great unmet need for patients and the rare disease community through the development of one-time, potentially curative treatments.

We commend the company for their leadership in harnessing the potential of hematopoietic stem cell gene therapy to change the course of severe inherited disorders. This Award particularly recognises Orchard Therapeutics for developing and bringing to Europe gene therapy for early onset metachromatic leukodystrophy (MLD), which addresses the urgent need of young patients and their families to treat a disease for which no approved therapeutic option previously existed. EURORDIS also appreciates Orchard Therapeutics' continued readiness to facilitate and advocate for early diagnosis in underserved disease areas.

Orchard therapeutics™

2021

EURORDIS PHOTO AWARD

1st by Pavol Kulkovský

'Together Forever'

Proximal Spinal Musclar Atrophy Type 2

Slovakia

2nd by Khim Bahadur Lamichhane

'OI Can'

Osteogenesis Imperfecta

Nepal

3rd by Díana Júlíusdóttir

'Living With A Rare Disease'

Alternating Hemiplegia of Childhood

Iceland

2020

EURORDIS BLACK PEARL AWARDS

2020

MEMBERS AWARD

ALAN Maladies Rares

The EURORDIS Members Award recognises ALAN's outstanding work in providing empowering information and support to patients living with a rare disease and their families, thereby giving patients greater autonomy and significantly improving their quality of life. ALAN's specialised team provide socio-therapeutic support and psychological counselling throughout the different stages of a patient's experience living with a rare disease, all free of charge. ALAN's support helps patients living with a rare disease to feel empowered and to thrive, (re)constructing and redirecting their lives after diagnosis.

The patient organisation ALAN was founded in 1998 initially to support and inform Luxembourg-based individuals affected by neuromuscular diseases. In 2005, they expanded activities to serve people living with other rare diseases.

In addition to providing socio-therapeutic support for patients, ALAN organises numerous public outreach activities to raise awareness for rare diseases among the general public, politicians, authorities and institutions. They are actively involved in implementing the National Plan for Rare Diseases, developed under their expertise in 2018 in collaboration with the Ministry of Health and other stakeholders. The aim of the plan is to ensure equal access to diagnostics, therapy, care and research for all people living with a rare disease.

ALAN Maladies Rares Luxembourg is an exemplary EURORDIS Member for the holistic approach used to support patients in so many aspects of their lives – from access to healthcare information to school, work and family life and social inclusion. ALAN is a shining example among EURORDIS Members and a truly deserving recipient of this award.

2020

VOLUNTEER AWARD

Claudia Crocione

Claudia Crocione is a long-standing patient advocate and Managing Director of HHT Europe. The EURORDIS Volunteer Award 2020 recognises Claudia Crocione's exceptional work as a patient advocate for the rare disease community, as well as her outstanding contributions to EURORDIS on a volunteer basis for many years.

The EURORDIS Volunteer Award recognises Claudia's personal commitment to the HHT (Hereditary Haemorrhagic Telangiectasia) community, both in Italy and across Europe.

She is a very active advocate in her work representing HHT patients and families in the VASCERN European Patient Advocacy Group (ePAG), the Council of European Rare Disease Federations, and is currently helping to establish the HHT CAB (Community Advisory Board). In addition, we would like to recognise her voluntary contributions to numerous other EURORDIS activities such as the EURORDIS Membership Meeting Budapest and our webinar series.

Claudia's commitment, resilience and motivation to make a positive difference to the lives of people living with a rare disease is reflected in all of her activities, and she is a truly deserving recipient of the Volunteer Award 2020.

2020

POLICY MAKER AWARD

Dr. Vytenis Andriukaitis

The EURORDIS Policy Maker Award recognises Dr. Vytenis Andriukaitis' outstanding work and support of the rare disease community in his role as European Commissioner for Health and Food Safety 2014-2019.

During his tenure, Dr. Andriukaitis supported significant developments to European policy, which have had a positive impact on outcomes for people living with rare diseases. He was a key driving force in the development and the launch of the European Reference Networks in 2017, which promise huge transformative potential for healthcare delivery in the years to come.

EURORDIS and the wider rare disease community also greatly appreciated his support for the proposal for European Cooperation on Health Technology Assessment (HTA). This very important European legislation will ensure a more transparent system for therapies, thereby reducing inequalities through an evidence-based decision-making process.

Above all, throughout Dr. Andriukaitis' time as Commissioner for Health, he was a tenacious and courageous proponent of the added value of European action in public health. Dr. Andriukaitis is a strong defender of meaningful patient engagement in all areas that have an impact on their lives, from healthcare to research, and a passionate supporter of the rare disease community. The former Commissioner is a truly deserving recipient of this award and we are delighted to celebrate these achievements as his mandate comes to an end.

EUROPEAN RARE DISEASE LEADERSHIP AWARD

Dr. Daria Julkowska

The EURORDIS European Rare Disease Leadership Award 2020 recognises the exemplary leadership and dedication Dr. Daria Julkowska has shown to the rare disease community and the positive impact her work has had on rare disease research and partnerships, both in Europe and beyond.

We would like to acknowledge Daria's outstanding commitment to various rare disease programmes; E-Rare, the ERA-Net for research programmes on rare diseases, where she became coordinator of the programme; IRDiRC as Chair of the Funders Constituent Committee and then Coordinator of the Scientfic Secretariat; and most notably as coordinator of the European Joint Programme on Rare Diseases (EJP), which brings together research and funding stakeholders from across Europe.

We would like to acknowledge Dr. Julkowska's determination in establishing the EJP and overcoming numerous challenges in bringing this large collaborative consortium together. It is thanks to her leadership that this ambitious yet game-changing Rare Disease research programme was successfully evaluated and funded, bringing together over 130 institutions in over 30 countries. The importance of the EJP in the field of rare disease research cannot be overstated, and we are delighted to celebrate Daria's commitment to rare disease leadership tonight.

2020

SCIENTIFIC AWARD

Prof. Annemieke Aartsma-Rus

The EURORDIS Scientific Award 2020 is given to Professor Annemieke Aartsma-Rus, Professor of Translational Genetics at Leiden University Medical Centre, the Netherlands, and Visiting Professor, Newcastle University, UK.

This Award recognizes her exceptional achievements in the field of Duchenne Muscular Dystrophy (DMD) over the past 20 years, during which time she has published over 150 articles. She is the most influential DMD researcher in the past ten years in the world, according to Expertscape, and has been recognised as a member of the junior section of the Dutch Royal Academy of Sciences, marking her as one of the top 50 scientists in the Netherlands under 45.

By sharing her expertise on numerous EU-funded projects, Annemieke has demonstrated her total commitment to the rare disease community and to scientific collaboration at a European level, working on projects including TREAT-NMD, Bio-NMD, SCOPE-DMD, BIO-IMAGE and NEUROMICS. At the global level, she also plays a crucial role in contributing to the work of IRDiRC, in particular to its Therapies Scientific Committee.

As well as being a distinguished world leader in the field of academic DMD research, Annemieke goes above and beyond, donating her time and talents to patients and their families to explain and translate difficult concepts, making them comprehensible to those living with DMD. With her colleagues, she made a video explaining antisense oligonucleotide therapy development, to ensure patients and families grasp the concept and its potential impact. Annemieke has also played a significant role in the EURORDIS Open Academy as a faculty member, especially encouraging the attendance of researchers to the Summer School on medicines development, allowing them to learn more about the issues which matter to patients.

Industry revere her knowledge and judgement, as she educates and urges them to always consider patients at the heart of the conversation. It is with humour and extraordinary kindness that she delivers her intellectual capability to a wide audience. Professor Annemieke Aartsma-Rus is a truly deserving recipient of this Black Pearl Award.

2020

YOUNG PATIENT ADVOCATE AWARD

Jana Popova

The EURORDIS Young Patient Advocate Award 2020 is awarded to Jana Popova for her exceptional advocacy work for people living with a rare disease. A PhD student in Media and Digital Communications and freelance journalist, she uses her skills and expertise in media and communications to raise awareness of the issues faced by the SMA (Spinal Muscular Atrophy) community, as well as rare diseases as a whole. She is the author of numerous articles about social policies, health systems, education, as well as different aspects of the daily life of people with neuromuscular disorders in Bulgarian as well as in English. She believes that people with chronic diseases – whether neuromuscular disorders, rare diseases, or other conditions – should raise their voices together to advocate for a better quality of life.

She first became involved in patient advocacy aged 15 as a volunteer for the Bulgarian Association for Neuromuscular Diseases, for whom she still volunteers now, 13 years later. Since 2017 she has been a member of the European Alliance of Neuromuscular Disorders Associations (EAMDA), where Jana has been instrumental in developing cooperation between different European patients' organisations for neuromuscular disorders. Since 2018 Jana has also been involved in the EPF (European Patients' Forum) Youth Group, advocating for young patients' rights and needs on the highest level.
In addition, Jana has participated at the EURORDIS Summer and Winter Schools, and the EPF's Summer University for Young Patient Advocates as a trainer, helping to educate and empower other patient advocates to make their voices heard.

This award recognises Jana's boundless commitment to amplifying the voice of patients with neuromuscular diseases and other young patients, showing that together, patient advocacy can have a huge impact.

2020

VISUAL & AUDIO MEDIA AWARD

‘Our Curse’ Tomasz Śliwiński & Magda Hueckel

The EURORDIS Visual & Audio Media Award recognises Tomasz Śliwiński & Magda Hueckel’s important and impactful documentary film, ‘Our Curse’, telling the story of the first six months of their son Leo’s life with Congenital Central Hypoventilation Syndrome (CCHS), also known as Ondine’s Syndrome. CCHS is a rare congenital disease of the central nervous system and patients do not have a breathing reflex, meaning they are artificially ventilated at least during the night.

The documentary has had a huge impact in raising awareness and improving the situation for patients like Leo living with CCHS. As a result of the discussion raised by this powerful and emotional film, donations have increased across the world for research into CCHS, new medical devices have already come to market, and the families of patients like Leo feel much less alone in their experiences. The CCHS Foundation has been established in the USA, and Tomasz and Magda are its Regional Coordinators in Poland, with the principal aim of raising money to support CCHS research. Additionally, together with Anna Palusińska, Tomasz and Magda have started the Polish CCHS Foundation “Lift the Curse”. By documenting their personal experiences as parents of a child with a life-threatening rare disease, Tomasz and Magda have provided an authentic and genuine insight into what life can entail for rare disease patients and their families.

‘Our Curse’ has deservedly received global critical acclaim, winning a long list of film festival awards, and being nominated for Best Documentary (Short Subject) at the 87th Academy Awards. This Black Pearl Award recognises Tomasz and Magda’s powerful talent and ability to highlight the needs of people living with a rare disease to a wider audience, and for that we would like to thank them on behalf of our rare disease community.

2020

WRITTEN MEDIA AWARD

‘Diagnosis’ Dr. Lisa Sanders

Dr. Lisa Sanders is an American physician, medical author and journalist, and assistant professor of internal medicine and education at Yale School of Medicine. In 2002, she began writing a column for the New York Times magazine called ‘Diagnosis’, covering complex and rare medical mystery cases, which has also inspired a Netflix documentary series released in 2019.

Her prestigious and popular column has brought the needs and experiences of people living with a rare disease to the attention of a much wider global audience. The cases tackled in ‘Diagnosis’ give an invaluable platform to the rare disease community and share powerful stories with the broader public, who might not otherwise encounter the world of people living with a rare disease and their struggles to reach a diagnosis.

The stories told in ‘Diagnosis’ demonstrate the immense power of collaboration across the global rare disease community, to share research, expertise and therapies to help patients across borders. Such collaboration often helps patients to realise they are not alone, however rare their condition may be. Dr. Sanders’ column has made an important contribution to raising awareness about rare diseases across the USA and beyond, and we are delighted to honour her work with this Black Pearl Award.

2020

HOLISTIC CARE AWARD

EB Haus

This year, in 2020, we are delighted to introduce a new Black Pearl Award, which recognises the need in the decade to come for a 360°, holistic and innovative approach to patient care, based on each person's individual experience of living with a rare disease.

The inaugural Holistic Care Award recognises the outstanding, holistic facilities of the EB Haus, a project of DEBRA Austria, and master-minded by founder and CEO Dr. Rainer Riedl. It took several years, tens of millions of euros in donations, and sincere personal dedication to reach the EB House's opening in Salzburg in 2005.

The EB House is the world's first special clinic for "butterfly children", who live with Epidermolysis Bullosa. The centre's four inter-disciplinary units demonstrate the benefits of combining patient-facing support with clinical training and high-quality research under one roof. Thanks to its prestige and reputation as a world-leading facility, the EB House has welcomed butterfly children not only from Austria, but all over Europe, and has also inspired the creation of similar centres which combine research with clinical care and training facilities in other countries.

The EB House's recognition as the first Austrian Centre of Expertise for rare diseases, and as a member of the European Reference Network (ERN) for rare skin diseases, are a testament to the success of this world-class centre.

The EB House Austria is an excellent example of a centre operating with a holistic approach to improving the lives of patients living with a rare disease; from treating patients with the latest therapies, to educating clinicians and researching potential treatments for the years to come.

The EB Haus is a shining example of the benefits of holistic care and we therefore believe it to be a truly deserving recipient of this first ever EURORDIS Holistic Care Award, particularly in the year of the EB Haus' 15th anniversary, and DEBRA Austria's 25th year.

2020

COMPANY AWARD FOR PATIENT ENGAGEMENT

Boehringer Ingelheim

The EURORDIS Company Award for Patient Engagement 2020 recognises Boehringer Ingelheim's longstanding commitment to collaborative and comprehensive engagement with rare disease patients and patient organisations, including the scleroderma, idiopathic pulmonary fibrosis and cystic fibrosis communities.

In the area of Cystic Fibrosis, Boehringer Ingelheim collaborates with EURORDIS and the patient community to help structure early and meaningful patient input in clinical research. Over the last two years the company has participated in a Cystic Fibrosis Community Advisory Board, which serves as a platform for transparent and effective collaboration with the patient community to promote dialogue, exchange information and foster partnerships.

In addition, we note the Company's holistic approach to patient support which addresses patients' medical needs as well as barriers to access, and the social context of people living with a rare disease.

Boehringer Ingelheim's 'More Than Scleroderma' initiative is indicative of this focus. Launched in 2017, the campaign and accompanying website aim to provide patients, caregivers and healthcare professionals with medically accurate information about living with this rare disease and available social support networks.

Since 2004 the Company has been part of the EURORDIS Round Table of Companies demonstrating an openness to exchange on critical issues impacting rare disease patients. In numerous aspects of their work, Boehringer Ingelheim prioritise patient engagement to develop therapies to best serve the community's needs, and for this they are the deserving recipient of the Black Pearl Company Award for Patient Engagement 2020.

2020

COMPANY AWARD FOR HEALTH TECHNOLOGY

TOBEA

The EURORDIS Company Award for Health Technology recognises companies developing technologies with significant positive impact on the daily lives of people living with a rare disease. TOBEA (Thinking Outside the Box Engineering Applications) is an engineering start-up firm based in Athens and established in 2012.

In 2020 this Award is given to TOBEA's unique SEATRAC device which facilitates access to outdoor water activities for people with limited mobility. Where the SEATRAC is available, a wheelchair user can enjoy a swim in the sea unassisted, using the specially designed chair on tracks and a waterproof remote control. In 2019 the team installed SEATRACs on beaches in Greece, Turkey, Cyprus and Southern Italy.

An individual's mobility can often be limited by living with a rare disease, and this innovative technological solution promotes quality of life, well-being and the equal right to health for people with limited mobility. In addition, the solar powered SEATRAC is an environmentally conscious innovation, which is sensitive to its carbon footprint and leaves the landscape intact if removed.

2020

COMPANY AWARD FOR INNOVATION

Healx

The EURORDIS Company Award for Innovation commends companies undertaking ground-breaking activities to advance rare disease research and treatment development.

With this in mind, EURORDIS recognises Healx's promising alternative model for drug discovery which integrates Artificial Intelligence (AI) and machine learning to identify treatments and possible cures for rare diseases.

The Company's Healnet platform integrates AI, pharmacological expertise and patient perspectives to identify potential therapies for rare diseases without a known treatment.
The model aims to inverse traditional pharmaceutical drug discovery processes. Instead of identifying a disease area then developing a drug to match that target, Healx matches treatments to the patient's gene expression, and works with other stakeholders to repurpose and modify existing treatments to better cater to patients' needs.

Amongst the Company's promising projects engaging the patient community are collaborations with FRAXA, the Fragile-X Syndrome patient group and the Barth Syndrome Foundation to employ Healx's technology and share expertise to accelerate therapy discovery for these two rare diseases.

The award places a spotlight on Healx's significant investment in multistakeholder collaborations to develop potentially life-changing medicines for people living with a rare disease, as well as the Company's drive to promote alternative frameworks to address rare diseases.

2020

EURORDIS PHOTO AWARD

1st by Cheryl Holbert Millard
for The Progeria Research Foundation

'Zein and his Mom'

Hutchinson-Gilford Progeria Syndrome

Egypt

2nd by Meghan Garriott

'Infusion Day'

Severe Combined Immune Deficiency

USA

3rd by Kelly Wilkins

'Pure Joy'

Homocystinuria, Osteoporosis, Scoliosis

USA

2019

EURORDIS BLACK PEARL AWARDS

2019

MEMBERS AWARD

ALS Liga

Originally a small local self-help group founded in 1995, ALS Liga Belgium is now a professionalised patient organisation for, and on behalf of, all people living with Amyotrophic Lateral Sclerosis (ALS) in Belgium.

The EURORDIS Members Award recognises their great work in providing services and support that have significantly enhanced the quality of life of people living with ALS, not only across Europe but also internationally.

This patient organisation has played an important role in ensuring the stimulation and financing of much-needed ALS research through the establishment of the fund 'A cure for ALS', within which all donations are devoted entirely to scientific research on the disease.

ALS Liga Belgium are dedicated to providing expert care and direct patient support, for example through their partnership with the specialised Belgian care centre Middelpunt, the patient organisation offer tailored care, adapted shelter places and therapies for both patients and families.

The patient organisation works to defend the rights of people living with ALS at governments, agencies and institutions, lobbying at national and international levels to achieve better ALS-patients' access to clinical trials, to optimise social legislation, and to continue to provide a better quality of life for those living with ALS.

As well as offering psychosocial and administrative support, ALS Liga Belgium also have a service which provides sophisticated aid goods for both mobility and communication, free of charge.

They have already initiated several international projects, both within and outside Europe, and cooperate actively within an International Alliance. The ALS League plays an active role in research communication with third parties and continues to advocate priority support for projects focused on rare diseases.

2019

VOLUNTEER AWARD

Richard West

Richard West is a patient from the United Kingdom living with Behçet's syndrome. Following his diagnosis in 1995 and after having experienced the many challenges of his condition, Richard has dedicated himself to helping others with Behçet's Syndrome, as well as those living with other rare diseases experiencing similar challenges. The EURORDIS Volunteer Award recognises this admirable commitment of 20 years as a dedicated and humble advocate of rare disease issues on behalf of the rare disease community.

This award also recognises his role as moderator of the Rare Connect Behçet Community, the creation of the International Behçet Society, as well as his long standing support as one of our representatives at the Patient and Consumer Working Party at the European Medicines Agency. As well as his advocacy and awareness-raising work on behalf of Behçet's Syndrome, Richard has been an active member of both the EURORDIS Therapeutic Action Group (TAG) and Drug Information, Transparency and Access (DITA) Task Force, and has been particularly involved in taking important actions on the off-label use of medicines in rare diseases.

Richard is currently working on a project about the care and treatment available to people with mental health distress in the UK, including those affected by rare diseases.

2019

VOLUNTEER AWARD

Russell Wheeler

Russell Wheeler is a trustee and patient advocate for the Leber's Hereditary Optic Neuropathy Society (LHON Society). Russell started volunteering within the patient community 6 years ago after his son lost his sight in a matter of a few months due to the rare genetic condition, LHON, for which there was no effective treatment. As he learned that the rest of his family were also at risk of losing their sight due to its maternal inheritance, and after attending the EURORDIS Summer School in 2012, Russell was driven to become involved in patient advocacy.

In addition to Russell's work for the LHON Society, The EURORDIS Volunteer Award serves to recognise his exceptional work as a patient advocate for all rare eye conditions as a patient board member of the European Reference Network ERN-EYE, and for all rare diseases as one of our EURORDIS volunteers.

Equally, Russell's active involvement at the International Society for Pharmacoeconomics and Outcomes Research (ISPOR) and participation in the Patient Focused Medicines Development (PFMD) project show his dedication and support to the cause.

Since 2015, Russell has been a fellow of The European Patients Academy (EUPATI) and is a member of the EURORDIS Drug Information, Transparency and Access (DITA) Task Force as well as the European Patient Advocacy Groups (ePAG) Steering Committee.

His commitment to the rare disease community is reflected in all of his activities, using his skills to help facilitate better science and research and in turn making a difference to the lives of those living with a rare disease, which is why he is being awarded one of the EURORDIS Volunteer Awards this year.

2019

LIFETIME ACHIEVEMENT AWARD

Michael Griffith

Michael Griffith is the co-founder of Fighting Blindness, a charity set up in 1983 dedicated to funding research into retinal blindness. Diagnosed with retinitis pigmentosa (RP) as a young adult, Michael has lived through the gradual loss of his sight, now having less than 10% vision remaining. At the time of diagnosis, there was a lack of information on this condition, which affected not only himself, but many members of his extended family. Michael was motivated to make a difference and decided to invest in research through Fighting Blindness.

Under his leadership, Fighting Blindness raised funds, and commissioned genetic scientists in Trinity College Dublin to start a project investigating the cause of RP. This group went on to be the first in the world to discover the first gene responsible for this condition. Since then, Fighting Blindness has invested nearly €20 million into Irish research, has had significant breakthroughs including potential treatments, and is one of Ireland's leading medical research charities.

After having three of his five daughters diagnosed with epidermolysis bullosa (EB), Michael went on to establish the charity Debra Ireland and the Medical Research Charities Group, and through these he has been a central figure in making a true and lasting difference for people living with EB, and other rare diseases, through his collegiate approach to advancing and funding medical research.

Michael has been a key player in providing platforms that represent the patient voice, improve patient access and collaboration in research and in turn, influence the development of patient-centred health policy thanks to his innovative work with both Rare Disease Ireland and the Irish Platform for Patient Organisations, Science and Industry (IPPOSI).

Michael also founded the Genetic and Rare Disorders Organisation (GRDO), a group acting as a national alliance for voluntary groups representing the views and concerns of people affected by genetic or other rare diseases.

The EURORDIS Lifetime Achievement Award recognises his exceptional work and vast achievements as a leader and driver for change in the field of rare diseases. The ripple effect of Michael's impact has been phenomenal, with his passion, drive and energy continuing to inspire others.

2019

POLICY MAKER AWARD

Edmund Jessop

Edmund Jessop trained in public health and has worked for the National Health Service in England for the past 30 years. He is also a Fellow of the UK Faculty of Public Health and has taught courses on public health, including health economics and health technology assessment for over 15 years. Since 2002, Edmund has been medical adviser to the National Commissioning Group (NCG) which plans, funds and monitors services for patients with extremely rare diseases. Through the NCG, Edmund has been involved in the design of managed access agreements for orphan drugs and the decision making on other very specialised technologies.

The EURORDIS Policy Maker Award recognises his outstanding work and support of the rare disease community through his dedication to patient advocacy and to the improvement of rare disease policy. The value of his contribution is in having tackled rare diseases as a public health issue.

As public health lead of the NCG and a member of the team for the reform of highly specialised care in the UK, Edmund's role was of huge value in protecting the rare disease services at a time of important reform with the establishment of NHS England.

His approach on reform for these services has resulted in significant improvement for care for rare diseases in the UK, with significant life increase and improvement for the 71 services concerned.

The Award also acknowledges his active involvement at a European level. Edmund was a member of the Rare Diseases Task Force in the UK, and was later nominated as representative of the UK at the EU Committee of Experts on Rare Disease (EUCERD). He has acted as the UK representative of the Commission Expert Group on Rare Diseases (CEGRD), has actively supported EUROPLAN for the last ten years, and has championed the development of European Reference Networks in contributing to their original design and in bringing his expertise from the UK. Edmund has chaired sessions at the European Conference on Rare Diseases & Orphan Products (ECRD) and has also taught at the EURORDIS Summer School for many years.

Edmund is very committed to visiting the Centres of Excellence of the 71 nationally highly specialised healthcare services, which is invaluable to maintain their focus on the needs of patients, and is further testament to his motivation to improve the lives of people living with a rare disease.

2019

RARE DISEASE LEADERSHIP AWARD

Till Voigtländer

Till Voigtländer is an associate professor in the field of neurobiology at the Medical University of Vienna. The EURORDIS European Rare Disease Leadership Award recognises the outstanding leadership and commitment Till has shown for the rare disease community, and the positive impact he has made on rare disease policy both in Austria and on an international level.

Dealing almost exclusively with very rare diseases in his diagnostic lab, he became Orphanet country coordinator for Austria in 2004. In 2008, he organised a petition for a National Plan for rare diseases that was presented to the Austrian Minister of Health. In response to this petition, Till was officially assigned head of a committee that was mandated with the elaboration of the outlines of the National Plan.

Through his organisation of various rare disease congresses and events (including the first Rare Disease Day walk in Austria), Till was able to mobilise and bring together the leaders of individual patient organisations, which lead to the foundation and constitution of the National Alliance for Rare Diseases, Pro Rare Austria, in 2011.

In the same year, the National Coordination Centre for Rare Diseases (CCRD) in Austria was founded on behalf of the Ministry of Health, again with Till as a head and initiator. The CCRD's main task was to finalise the rare disease National Plan, which was eventually adopted in 2015. Since then, the CCRD under the leadership of Till has been focusing on structuring and shaping the clinical landscape of rare diseases in Austria by working on the designation and quality control of national centres for rare diseases.

The Ministry of Health also nominated him as Austrian representative in the European Union Committee of Experts on Rare Diseases (EUCERD) (2010-2013), the Commission Expert Group on Rare Diseases (CEGRD) (2014-2016), as well as the Cross-border Healthcare Directive Expert Group (2012-2013). The latter set the base for him to be nominated for the Board of Member States on European Reference Networks (ERN), where he was elected co-chair and has since then become a leading figure in the organisation and shaping of the ERN structure. Till also functions as an important communicator for the rare disease cause through his countless talks and presentations on the topic both in Austria and abroad.

2019

SCIENTIFIC AWARD

Professor Philip Van Damme

Professor Philip Van Damme is professor of Neurology at the University of Leuven, Belgium. After training as a medical doctor, Philip started neurology training in 1999. He obtained his PhD in 2004 with a study of the pathophysiology of Amyotrophic Lateral Sclerosis (ALS), and after graduating as a neurologist, became a member of staff at the University Hospital in Leuven in 2006.

He has continued working in the field of ALS, combining the diagnosis and care for patients with ALS, with clinical studies and fundamental research on this disease.

Since 2013, Philip has been Director of the Neuromuscular Reference Centre (NMRC) at the University Hospital in Leuven, which coordinates the multidisciplinary care of ALS patients.

He also is a principal investigator in the laboratory for Neurobiology (part of the Neuroscience Department at the University of Leuven and the VIB Centre for Brain & Disease Research). His current research focuses on genetic modifiers of ALS and on disease pathways in pluripotent stem cell models derived from ALS patients.

Philip has collaborated with the patient organisation ALS Liga Belgium intensively since the start of his career, setting up fundraising campaigns for research and contributing to creating awareness. He regularly plans projects with the patient organisation and through this collaboration; Philip and ALS Liga Belgium are aiming to reach their mutual objectives of improving care for patients with ALS, better understanding the disease causes, and identifying new options for treatment.

2019

YOUNG PATIENT ADVOCATE AWARD

Laëtitia Ouillade

Laëtitia Ouillade, who was born in France in 1993, is a patient advocate dedicated to raising awareness about Spinal muscular atrophy (SMA). Laëtitia was diagnosed with SMA Type 2 in 1995 and since a very young age, she has been helping to inform the general public about this disease. Throughout her school years, she would often give talks and testimonies to raise awareness among the pupils on the question of handicap and the reality of living with SMA.

Laëtitia's dedication is reflected further in her TV and radio appearances, which have been instrumental in bringing awareness of the needs of people living with a rare disease to a wider audience. As well as giving live interviews on the TV studio set for the AFM-Téléthon, she has also organised two events to benefit the Telethon.

At 17, Laëtitia gained her high school diploma in sciences with distinction and began her studies at the University Institute of Technology. Since receiving her engineering diploma of the National School of Cognitive Science in 2016, she has been working full-time at ATOS Bordeaux as a research engineer.

Also in 2016, she joined the SMA GI (Interest Group) of AFM-Téléthon and a year later, she became the leader of the Group. Within the SMA Interest Group, she manages the social networks and scientific information, boosting the SMA meetings in different regions and setting up telephone helplines with the regional offices of AFM.

Laëtitia has given much of her time and energy to the rare disease cause and last year, she trained as a patient expert during the EURORDIS Summer School 2018 and in her role as a patient representative in a Working Group of the International Rare Diseases Research Consortium (IRDIRC). In the projects of the GI, Laëtitia continues her exceptional advocacy work for people living with a rare disease and is planning a national meeting, as well as help to promote the access of new treatments for SMA Type 3 through an interview with the Ministry of Health later this year.

2019

VISUAL & AUDIO MEDIA AWARD

Anne-Dauphine Julliand

The EURORDIS Visual & Audio Media Award recognises Anne-Dauphine Julliand for her inspiring feature-length documentary film, Et Les Mistrals Gagnants, which was released in 2017.

Anne-Dauphine was born in 1973 in Paris. After studying journalism, she took her first steps as a writer in the daily press, and later in real estate press.
She married in 2000 and had four children:
Thaïs, Gaspard, Arthur and Azylis.

In 2006, her daughter Thais was diagnosed with metachromatic leukodystrophy, a rare form of lysosomal disease and a year later, Thais tragically died of this disease. The family soon discovered that her younger sister Azylis was also carrying this disease.

In 2011, Anne-Dauphine Julliand published Two Little Steps on the Wet Sand, which tells the story about the life, illness and death of Thaïs, then A Special Day in 2013 where she recounts her life as a mother facing the serious and rare illness of her two daughters.

Four years later, Anne-Dauphine directed the documentary Et Les Mistrals Gagnants. It raises awareness of various rare diseases through following the lives of five children each living with a different rare disease. By documenting these stories, and allowing these children to speak for themselves, Anne-Dauphine has helped portray an unfiltered and genuine insight into what it means for each of these individuals to live with a rare disease.

The documentary was broadcast in Belgium, Japan, Spain, Germany and Lebanon, bringing the needs of people living with a rare disease to a wider audience, as well as finding an engaging way to reach an audience that may not already be part of the rare disease community.

WRITTEN MEDIA AWARD

Bojana Mirosavljević

The awardee of the Written Media Award 2019 is Bojana Mirosavljević for her specialised journal for rare diseases, 'Word for Life' (Serbian: 'Reč za život').

From her personal experience of having a daughter Zoya with Batten disease, and then the huge grief of losing her to the disease in 2013, Bojana invested all her efforts in building a better environment for other patients and parents, including this magazine.

Distributed bi-monthly in both printed and electronic form, 'Word for Life' is the first and only journal dedicated to rare diseases in the Balkan region and has brought the needs of people living with a rare disease to the attention of a wider audience in Serbia and beyond.

The journal brings together important aspects of the rare disease cause, sharing with its readers powerful and supportive patient stories; knowledge and experience from rare disease experts; updates on rare disease legislation; and rare disease news from all over the world. This award recognises the journal's important contribution to raising awareness about rare diseases through its reach not only to families, but also to medical professionals, and other relevant institutions across the Balkans.

2019

COMPANY AWARD FOR PATIENT ENGAGEMENT

Project HERCULES

The EURORDIS Company Award for Patient Engagement recognises the collaborative effort of all the Companies involved in Project HERCULES to support access to new treatments for Duchenne Muscular Dystrophy (DMD). Following its establishment in November 2017 by the patient organisation Duchenne UK, seven companies expressed an interest in working together on Project HERCULES: Pfizer, PTC Therapeutics, Roche, Sarepta Therapeutics, Solid Biosciences, Summit Therapeutics, and Wave Life Sciences.

Project HERCULES provides a unique, international platform, bringing together patient organisations, industry, academia, charities and experts. It has allowed companies to engage with patients and caregivers, further the knowledge of DMD, and address key issues to better understand patient needs.

Through this all-important engagement, Project HERCULES aims to develop tools and a high-quality evidence base for DMD to support the Health Technology Assessment (HTA) process, and in turn enable more transparent and consistent reimbursement decisions for new DMD treatments.

This Award celebrates how Project HERCULES has set a model example of multi-stakeholder collaboration and has the potential to encourage similar initiatives across other rare diseases.

2019

COMPANY AWARD FOR HEALTH TECHNOLOGY

Air Liquide

Air Liquide Medical Systems is the subsidiary of Air Liquide Healthcare that is dedicated to Medical Devices. The EURORDIS Company Award for Health Technology recognises Air Liquide Medical Systems' longstanding commitment and international reach in the development of life-changing respiratory devices and services. These have benefitted many people around the world who live with a rare disease and have respiratory deficiencies.

Air Liquide is particularly attentive to technological advances. Their continuous innovation proposes technological solutions, brings to market easy-to-use medical products and equipment, and provides essential services that protect and improve the lives of patients not only in hospitals and clinics, but also at home. Through the company's periodic reviews and modifications on the performance, safety and reliability of their medical devices, they are able to make life-changing improvements for patients.

The Award also recognises how, as well as partnering with several international research Centers of Excellence, Air Liquide develops each of their products in collaboration with healthcare professionals in order to meet patients' needs and promote advances within the healthcare environment.

2019

COMPANY AWARD FOR INNOVATION

Chiesi

Chiesi Farmaceutici S.p.A. is an international company based in Parma, Italy, with a strong focus on research, development, production and the commercialisation of innovative medicines in the Respiratory, Neonatology, Rare Disease and Special Care Therapeutic Areas.

Chiesi has worked in the development and access of more traditional and highly innovative drugs, and they aim to be a point of reference for patients affected by health conditions that are difficult to treat, making rare diseases one of their priorities.

The EURORDIS Company Award for Innovation recognises Chiesi's strong commitment to rare diseases, including the Company's significant investment in Research and Development for rare diseases and its involvement at European and global levels, notably through its important contribution to the work of the International Rare Diseases Research Consortium (IRDiRC).

The Award also recognises the Company's support to policy development, as well as its strong pipeline for bringing to market a wide number of treatments to address many rare diseases, including products for diseases which are often neglected with extremely low prevalence.

EURORDIS applauds the innovation that is the hallmark of the rare disease community.
Continued innovation through effective collaboration is needed now for the millions of people worldwide who are living with a rare disease, for which Chiesi is an excellent example.

2019

EURORDIS PHOTO AWARD

1st by K.Deniz Kalayci

'Deep'

CFC Syndrome- Cardiofaciocutaneous

Turkey

2nd by Audrey Guyon

'Eleanore'

Rett syndrome

France

3rd by Cindy Symons

'Family'

Tuberous Sclerosis Complex

Belgium

2018

EURORDIS BLACK PEARL AWARDS

2018

VOLUNTEER AWARD

Chris Sotirelis

Chris Sotirelis is a patient with beta thalassaemia major. He has been a tireless patient advocate for EURORDIS and the rare disease community at national and international level for many years. He was involved in the first ever thalassaemia clinical outcomes patient registry until 2001. His expertise includes the setting up of the National Haemoglobinopathies Register (NHR), and previously being the UK Thalassaemia Society representative on the NHR commissioning group. Since then he has been directly involved in the development of surveys to assess patient quality of life.

More recently, he has been leading the creation of a PROM (patient reported outcome measure) aimed at being integrated within the NHR. Its aim is to elicit areas of inequity and on how patients experience the impact of their treatment. His earlier work within the Sickle Cell and Thalassaemia Screening Programme Steering Group Committee has allowed him to give a patient perspective and help develop the ethics underlying screening for a genetic condition like thalassaemia, as well as on issues of "informed consent" and "informed choice."

As one of the European Medicines Agency's (EMA) experts, affiliated to EURORDIS, he has strived to increase engagement in patient-critical areas within the wider EMA regulatory framework, and has been consulted on many Health Technology Assessment (HTA) Parallel Scientific Advice sessions.

He has been an invited speaker to many conferences and workshops on Access and Reimbursement and, notably, has presented the "Patient perspective on HTAs for Personalised Medicine" during the plenary session debate of the HTAi conference in Bilbao in 2012. He is very engaged as the lead representative of his patient community in NICE Health Technology Assessments (Single and Multiple Appraisals) and in drafting national commissioning policies for standard specification of care and orphan medicinal products for thalassaemia patients.

2018

VOLUNTEER AWARD

Helene and Mikk Cederroth

Helene and Mikk Cederroth are two passionate and dedicated patient advocates who have contributed remarkably to the undiagnosed rare disease cause. Together they are the founders of the Wilhelm Foundation, which helps approximately 3 in 10,000 children who suffer from often fatal undiagnosed brain diseases. Helene and Mikk tragically lost three children due to undiagnosed rare brain conditions and made the courageous decision to dedicate their lives to the undiagnosed rare disease cause. By helping unite experts in different fields, Helene and Mikk have been instrumental in increasing the chances of people getting a diagnosis.

Throughout their lives they have both provided help, support and love, to patients and their families, particularly through Wilhelm Foundation's 'silver lining' initiative, whereby they help provide gifts or experiences to bring joy to families affected by brain conditions. In September 2014, they co-organized the First International Congress for Undiagnosed Diseases, and soon after launched the Undiagnosed Diseases Network International together with the NIH and other partners. The Second International Congress for Undiagnosed Diseases congress was in Budapest 2015, the third in Vienna 2016 February, the fourth in Tokyo in November 2016 and the fifth in Stockholm their home town in August 2017.

The purpose of these congresses is to encourage and develop collaborations that will significantly improve diagnosis of unsolved patients for all undiagnosed conditions, not just neurological ones. Their admirable fight to turn a loss into something so positive and constructive for the rare disease community is why they are being awarded one of the EURORDIS Volunteer Awards this year.

2018

LIFETIME ACHIEVEMENT AWARD

Alistair Kent

Alastair Kent is an expert in his field who has changed the face of genetic research in his long and dedicated career, providing patient support for children, adults and families living with different forms of genetic disorders.

A central figure in the UK, Europe and worldwide, he has influenced and advised patient organisations, the charitable sector and indeed government through the Department of Health. Campaigns led by Alastair have significantly influenced legislation to the benefit of patients and have received the gratitude of lawmakers.

Since 2013 Alastair has sat on the NHS England Rare Diseases Advisory Group, the Genomics England Ethics Advisory Committee, the Scottish Medicines Consortium Task & Finish Group on Improving Access to Medicines for Patients with Rare Diseases and the Department of Health's Rare Diseases stakeholder forum, which he has chaired from 2014. Since 2004 he has sat on the Public Population Projects in Genetics (P3G) Ethics Committee and the UK Genetics Testing Network Steering Committee for the Department of Health since 2003. He has also sat on: the Royal College of Physicians (Now Joint Committee of the Royal College of Physicians, RCPath and British Society for Human Genetics) Clinical Genetics Committee (since 1997); the Association of British Insurers Genetics Advisory Committee (since 1996); and the European Alliance of Genetic Support Groups (since 1993) of which he became president in 1995.

Prior to becoming the director of Genetic Alliance UK, Alastair was director of Action for Blind People between 1989 and 1993, as well as being Director of Education, Employment and Residential Services at the Royal National Institute for Deaf People between 1986 and 1989. From 1982 until 1986 he was Principal of Barnstead Place at Queen Elizabeth's Foundation for the Disabled, before which he worked as a County Careers Officer specialising in special needs for North Yorkshire County Council between 1981-82. From 1977-1981 he was a Specialist Careers Officer for Cambridge County Council and before this was a careers officer for Norfolk County Council from 1973 until 1977. From 2007-2013 Kent was a member of the ethics committee for the '1000 Genomes' International Project. Between 2008 and 2011 he was on the EMEA committee for advanced therapies. He has been recognised by his country for his numerous services to healthcare with an OBE, and thus we hope that this Lifetime Achievement Award from EURORDIS is a fitting recognition from fellow representatives and practitioners of the rare disease community.

2018

POLICY MAKER AWARD

Elena Gentile

Elena Gentile is an Italian politician who has supported the rare disease cause since she began practising as a paediatrician in Italy and throughout her political mandate. After gaining a degree in medicine and working as a paediatrician in Cerignola Hospital, Elena Gentile began her political career and, from 1985, she spent five years as Councillor for Health, Environment and Social Services in Cerignola, Italy and was elected Mayor of the city in 1991. As a respected political figure, she has used her position as a Member of the European Parliament since 2014 to help give a voice to Idiopathic pulmonary fibrosis (IPF) patient groups by supporting the first European IPF Patient Charter in EU Parliament and by mobilising MEPs around a written declaration on IPF. She has demonstrated her determination to collaborate with member states to enable access to EMA approved orphan products for IPF patients. Elena Gentile further exhibited her dedication to the improvement of rare disease policy through her support of the Parliamentary Advocates for Rare Diseases, a EURORDIS initiative which launched in October 2017. Her ongoing collaboration with UNIAMO, the Italian National Alliance for Rare Diseases, has resulted in the organisation of the photographic exhibition 'Rare Lives' at the European Parliament; and she is supporting the high level meeting on the European Reference Networks (ERNs) organised by the Rare Bone Diseases Network (ERN BOND) at the European Parliament on the occasion of Rare Disease Day 2018.

2018

RARE DISEASE LEADERSHIP AWARD

Professor Bruno Sepodes,

The Rare Disease Leadership Award is being deservedly presented to Professor Bruno Sepodes, an exceptional leader with international influence.

Currently Professor of Pharmacology and Pharmacotherapy at the Faculty of Pharmacy of the University of Lisbon, he develops his research in Pharmacology and Translational Medicine.

Simultaneously, he is completing his final year as Chair of the Committee for Orphan Medicinal Products (COMP), and is a member of the Committee for Human Medicinal Products (CHMP) and of the Committee for Advanced Therapies (CAT) at the European Medicines Agency. His collaboration with the European Medicines Agency started as a member of the COMP in 2008 and followed as a member of the Patients' and Consumers Working Party during 2012. Bruno has exhibited his expertise further as an expert for the National Medicines Authority (INFARMED) and for the Veterinary General Directorate (DGV). Concerning the involvement in research projects, international collaborations include the William Harvey Research Institute (UK) and other relevant research institutes.

A true leader in his field, Bruno has authored and co-authored over 70 scientific publications in international journals, and more than 100 scientific communications (on pharmacology, toxicology and therapeutics), presented to national and international scientific meetings. The EURORDIS European Rare Disease Leadership Award recognises Bruno's long-standing collaboration with the European Medicines Agency and indeed EURORDIS as well as his outstanding leadership and commitment to the importance of including patient advocates as equal stakeholders in all discussions and decisions.

2018

EURORDIS SCIENTIFIC AWARD 2018

Professor Michele De Luca and Doctor Tobias Hirsch

The recipients of the EURORDIS Scientific Award 2018 are Professor Michele De Luca and Doctor Tobias Hirsch, as well as their respective teams at the Centre for Regenerative Medicine "Stefano Ferrari" (CMR) at the University of Modena and Reggio Emilia (Italy) and the Severe Burn Injury Centre of the University Hospital of the Ruhr University, in Bochum (Germany) for their collaboration in developing a life-changing gene therapy.

This therapy, which enables the human epidermis to regenerate through the use of transgenic stem cells, recently saved the life of a young boy named Hassan, affected by the rare disease junctional epidermolysis bullosa (EB). There are many forms of EB, most of which result in blistering and lesions of the skin and mucosal membranes. Due to the lack of available treatments, this disease significantly reduces the patient's quality of life and can also be life-threatening.

Doctor Tobias Hirsch's scientific career includes a past as a resident in Plastic Surgery at the University of Heidelberg. Between 2007 and 2010, he was a Postdoctoral Research Fellow at the Laboratory of Molecular Oncology and Wound Healing, at the BG University Hospital Bergmannsheil, Ruhr University Bochum, Germany. Additionally, between 2005 and 2007 he was a Postdoctoral Research Fellow and Member of Faculty in the Division of Plastic Surgery, at the Harvard Medical School, Boston, MA, USA.

EURORDIS SCIENTIFIC AWARD 2018 CONTINUED

After trying established therapies without success, Dr Tobias Hirsch's medical team from Bochum, Germany decided to adopt an experimental approach. They took a sample of Hassan's skin which was unaffected by the bacterial infection and sent it to Modena, where Prof De Luca's team cultured in the lab a large amount of transgenic epidermis. This new skin derived from genetically modified stem cells was then transplanted onto the wound surfaces. Hassan is the first patient worldwide to have been treated successfully in the entire body by this gene therapy that was developed after many years of research by Prof Michele de Luca and his team, an exemplary example of cross-border scientific collaboration. Phase I/II clinical trials are now being carried out also on other types of epidermolysis bullosa. In the future it could be extended to treating other genetic skin conditions. Professor Michele De Luca is the Director, co-Founder and Scientific Director of Holostem Terapie Avanzate S.r.l. and the author of over 120 peer-reviewed publications. Through these ventures, he has exhibited his dedication to innovation in healthcare. In 2017, he was awarded The Niche's Stem Cell Person of the Year Award. In 2014, Michele was one of the winners of the ISSCR Public Service Award for his involvement in public debate and policymaking in Italy and their championing of rigorous scientific and medical standards and stringent regulatory oversight in the introduction of new stem cell treatments into the clinic.

Prof De Luca and Prof. Graziella Pellegrini at CRM are also leading other innovative stem cell and regenerative medicine work including the development of Europe's first stem cell-based ATMP (Advanced Therapy Medicinal Product), in the form of a new adult stem cell-based therapy for vision loss called Holoclar®.

The research team behind the scientific breakthrough

2018

YOUNG PATIENT ADVOCATE AWARD

Sammy Basso

Sammy Basso, who was born in Schio, Italy in 1995, is a patient advocate dedicated to raising awareness about Hutchinson-Gilford progeria. Sammy is the eldest of approximately one hundred people in the world living with progeria, and currently studies natural sciences, focusing on biology.

Whilst only 9 years old, he helped to created his own advocacy group, l'Associazione Italiana Progeria Sammy Basso, which has been instrumental in informing the general public and promoting the need for, progeria research.

Together with Bologna's Institute for Molecuar Genetics – National Research Council (IGM-CNR), l'Associazione Italiana Progeria Sammy Basso created the Italian Network for Laminopathies, a group of Clinical and Research Centers performing clinical and molecular diagnosis or biomedical research in the field of laminopathies.
As the driving force behind his advocacy group, he has been unstoppable in fulfilling his dreams to explore the world despite the apparent limitations of his condition, shown in his Nat Geo People Documentary, Il viaggio di Sammy, which documented his trip to the USA along Route 66, and his book of the same name.

Sammy plans to become a researcher and contribute actively to the study of progeria, for which he has already been instrumental in raising awareness.

PATIENT ORGANISATION AWARD

Pulmonary Hypertension Association Europe

Pulmonary Hypertension Association Europe is dedicated to improving the lives of patients living with pulmonary hypertension (PH) in Europe by working with its members to enhance awareness of PH, promoting optimal standards of care for people living with the disease, ensuring the availability of all approved treatments and encouraging research for new medicines and therapies. Founded in Vienna in 2003, the organisation has grown to a level where it now includes 29 patient associations from 33 countries in Europe. Their call to action, which was presented in the European Parliament in 2012, is to improve access to expert care, improve awareness and screening, encourage clinical research and innovation, empower patient groups and ensure the availability of psychosocial support.

The European Parliament event on Organ Donation and Transplant in October 2016, initiated by PHA Europe, is an example of how far the organisation has succeeded in reaching out to the decision makers at EU level, thanks to close collaboration with the European patient federations for diseases where organ transplants are relevant. Four representatives from PHA Europe are also members of the ePAG (European Patient Advocacy Group) network.

PHA Europe recently conducted their 'white spots' programme. PHA identified countries in which there are no PH patient associations – 'white spots' – and consequently, in European countries with more than one million citizens, only two countries remain with no patient associations. PHA continued to grow with their fellowship programme, which aims to improve communication between member associations.

The Annual PH European Conference (APHEC) gives member associations opportunities for capacity building as well as for information and education. The 2016 APHEC featured three international PH medical opinion leaders as speakers, a cardiologist, a pulmonologist and a paediatrician, who provided the attendees with the latest information on treatment strategies, surgery and research.

VISUAL & AUDIO MEDIA AWARD

Christopher Ulmer

The recipient of the Visual & Audio Media Award 2018 is Christopher Ulmer, who, founded Special Books by Special Kids, a video project that seeks to normalise the diversity of the human condition. After achieving a BA in Communications and a Masters Degree in Teaching, Christopher Ulmer became a teacher for children with disabilities ranging from brain disorders to autism, and was touched by the connection that he made with these incredible children who wanted to be understood in the world.

He decided to create a book series where his students explained life from their perspective. Originally denied by 50 publishers, Christopher turned to Facebook as a tool to publish his interviews via video and, after 6 months, the page had over 150,000 followers. Spurred on by the support of the rare disease community, Christopher began to interview patients outside of the classroom, and soon decided to do his video interviews full-time in the hope of bridging the gap between individuals with a diagnosis and the general public.

Since the creation of Special Books by Special Kids, the Facebook page has 1.6 million likes, as well as 200,000 Youtube subscribers and 245,000 Instagram followers. By connecting societies around the world, he has helped build a global dialogue around rare diseases and has built a media movement that supports acceptance regardless of diagnosis. This award recognizes how Christopher has helped to portray an unfiltered and genuine insight into what it means for individuals to live with a rare disease, and how he has, importantly, found an engaging way to reach those not directly part of the rare disease community.

2018

WRITTEN MEDIA AWARD

Dr Serge Braun

The awardee of the Written Media Award 2018 is Serge Braun for his book, 'On peut changer le monde, en vendant des crêpes et des ballons.' Dr. Braun worked for over a decade on neuromuscular diseases, working in university research, then in the private sector, where he conducted a gene therapy program dedicated to Duchenne muscular dystrophy, which led to the first worldwide clinical trial of gene transfer for a myopathy. He went on to work in cancer immunotherapy and HIV, whilst being vice-president of Alsace BioValley and co-founder of Neurofit, a company specialized in neurosciences.

From 2005, he joined the Association française contre les myopathies (AFM-Téléthon) and drove its scientific policy with one goal: the development of innovative therapies for rare diseases. The book for which he wins this award talks about both his experience as Scientific Director of the AFM-Téléthon as well as the cause of rare diseases and new developments in genetics, biology, and orphan drugs to cure these diseases.

Not only does the book didactically address scientific issues, it is through individual stories and anecdotes that Serge describes how the community has transformed the landscape of genetics, biology and orphan drugs. Through the testimony of families, researchers, doctors and volunteers, Serge gradually reveals the vision of ordinary people doing extraordinary things. Aimed at non-scientists as well as specialists, the book is both accessible and engaging, with profits going to the AFM-Téléthon.

2018

COMPANY AWARD FOR PATIENT ENGAGEMENT

European Federation of Pharmaceutical Industries and Associations, Bayer, UCB and MSD

The EURORDIS Company Award for Patient Engagement recognises the achievements of the European Federation of Pharmaceutical Industries and Associations, Bayer, UCB and MSD, in particular recognising their leadership as a group in championing the development of patient engagement activities within the Innovative Medicines Initiative, particularly through the PARADIGM project. It is timely to provide a framework that allows structured, meaningful, sustainable and ethical patient engagement throughout the development of medicinal products. In the longer term, this framework will strengthen both the understanding of stakeholders and system-readiness towards patient engagement across the diverse range of stakeholders, and ensure synergies with other initiatives focusing on the patient's voice in the life cycle of medicines.

The development of an inventive and workable sustainability roadmap to optimise patient engagement across medicines' R&D, demonstrates the inherent link between patient education, patient engagement and truly valuable innovation.

This Award celebrates commitment to meaningful engagement of patient representatives throughout the lifecycle of medicines.

2018

COMPANY AWARD FOR INNOVATION

Novartis

The EURORDIS Company Award for Innovation recognises Novartis for its longstanding track record in developing medicines for rare diseases. Through effective collaborations with the scientific, medical and patient communities, Novartis has a promising pipeline to address many rare diseases, including rare cancers. In 2017, Novartis received the first FDA approval for a gene therapy to treat cancer in children and young adults.

EURORDIS applauds the innovation that is the hallmark of the rare disease community. There are over 6000 rare diseases, an estimated 30 million people living with a rare disease in Europe and 300 million worldwide. The fact remains that few treatments are available for the majority of these diseases; many have no appropriate treatment or go undiagnosed. Continued innovation through effective collaboration is needed now for the millions of people worldwide who are living with a rare disease, for which Novartis is an excellent example.

2018

EURORDIS PHOTO AWARD

1st by Nicole Traubenek

'Our way'

Esophageal Atresia (EA)

Germany

2nd by Conny Wenk

'Philipp & Moritz'

Osteogenesis Imperfecta

Germany

3rd by Nicola Miller

'Unsung Heroes'

Xeroderma Pigmentosum

UK

2017

EURORDIS BLACK PEARL AWARDS

2017

VOLUNTEER AWARD

Elizabeth Vroom

Elizabeth received a degree in Dental Medicine (DMD) from the University of Groningen, before specialising in Maxillofacial Orthopedics and Orthodontics at Radboud University Nijmegen. She uses her expertise to help Duchenne Muscular Dystrophy (DMD) patients with orthodontic problems.

A mother to a son with DMD, she 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) and serves as Chair.

She has spent over 20 years dedicating her efforts to raising awareness, to raising funds, to educating and to working to see that patients receive optimal care, as well as helping to set up patient organisations around the world.

She has served on a number of advisory boards for patient care, research, ethics, development of new medicines and regulatory issues in the Netherlands as well as on pan-European ones. She has also been a member of EURORDIS' RD-Connect Joint Patient Advisory Council and currently chairs the TREAT-NMD Project Ethics Council.

Elizabeth shared her expertise at EURORDIS' 2015 and 2016 Summer Schools. EURORDIS' Summer School aims to empower patient representatives in the areas of clinical trials and EU regulatory affairs.

Elizabeth established the World Duchenne Awareness day. All those who joined to celebrate this day in 2016: 88 advocacy groups in 40 countries, have highlighted its success. This Award serves to recognise all that Elizabeth has accomplished in supporting so many rare disease organisations and her longstanding commitment to the rare disease community.

2017

LIFETIME ACHIEVEMENT AWARD

Anders Olauson

Anders Olauson has devoted the past 30 years of his life to the rare disease cause, notably rare conditions affecting the lives of children and their families. Through personal experience, Mr Olauson comments: "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."

Taking action to provide a platform for people living with a rare disease, Anders helped found the Ågrenska Centre in 1989, a national competence centre based in Sweden providing support for children, teenagers and adults affected by rare diseases. Here, he continues to serve as Chairman and established the Ågrenska Academy, a virtual centre for information, research and the dissemination of knowledge of rare diseases. His work also involves contact with legislative bodies on both a national and regional level, healthcare professionals, education and labour unions and other key players in the field of rare diseases.

Anders brought his enthusiasm and experience to his role as the President of EURORDIS-Rare Diseases Europe in 2000, as well as adherence as a member of the EURORDIS Board of Directors from 1999 to 2010. EURORDIS appointed Anders as its representative at the European Patients' Forum (EPF); elected President from 2005 to 2015, Anders is now the Honorary President of EPF. EPF is active in the field of European public health and health advocacy representing the patient voice on an EU level.

Since 2006, Anders has been a member of the Advisory Group for Health Research within the Directorate-General for Research and Innovation of the European Commission. The Swedish Government also appointed Anders as a member of the Advisory Council at the National Board of Health and Welfare in 2008, serving until 2014.

Anders' continued efforts have led to his involvement with the NGO Committee for Rare Diseases, initiated by Ågrenska and EURORDIS. This committee's objectives are to increase the visibility of rare diseases on a global level, as well as to make rare diseases a priority in public health, research, medical and social care policies.

Anders' vision and goals for the rare disease community are reflective of his long-term dedication to rare diseases. EURORDIS is delighted to award Anders with this Lifetime Achievement Award as it serves to honour all that Anders has done for the rare disease community.

2017

POLICY MAKER AWARD

Frédérique Ries

Frédérique Ries, Member of the European Parliament since 1999, has been a pioneer in, and a committed advocate for, the implementation of patient-centric health policies that focus on Union added value, of which rare diseases constitute the prime example.

As a member of the Committee on the Environment, Public Health and Food Safety within the European Parliament, Ries' support and groundwork were instrumental in achieving the adoption of the Regulation on Advanced Therapies in 2007. Increasingly campaigning for the rights of patients with rare diseases, she then collaborated with EURORDIS in 2008 to organise the first Public Hearing on Rare Diseases at the European Parliament for Rare Disease Day. In 2015, she presented a written declaration on improving treatment for rare diseases along with other Members of the European Parliament.

Her most recent work, presenting a proposal for a Pilot Project on a Foresight Study on Rare Diseases (Rare 2030) that promotes a continuous bottom-up research approach with participation from all stakeholders including patients, is proof of Ries' dedication towards tackling rare diseases in the European Union. For all of this, she is the unequivocal winner of the EURORDIS Policy Maker Award 2017.

2017

SCIENTIFIC AWARD

Lucia Monaco

Dr Lucia Monaco graduated in chemistry in 1979 from the University of Pavia and received her training in biochemistry at the University of Iowa in Iowa City, USA and in molecular biology at the European Molecular Biology Laboratory in Heidelberg, Germany.

In her role as Chief Scientific Officer at the Fondazione Telethon, she has made a significant impact in the field of rare genetic diseases in Italy and abroad, in particular through her strong commitment to the IRDiRC initiative.

Dr Monaco has also shown strategic vision in developing Fondazione Telethon's Rare Disease Programme and linked it with both academic and commercial partners, as well as patient organisations across Europe. She has maintained a significant presence at scientific and patient centred meetings including (IRDiRC), ICORD, and EURORDIS-Rare Diseases Europe.

Moreover, she has actively supported and shaped key research infrastructure developments in the field of rare diseases, particularly related to biobanking and data sharing via EuroBioBank and RD-Connect. Her personal enthusiasm, guidance and dedication inspires many scientists and clinicians to become involved in state-of-the-art research for rare diseases including the successful development of gene therapies. The EURORDIS Scientific Award recognises the major impact Dr Monaco's work has had on rare diseases.

2017

MEDIA AWARD

Aldo Soligno

Aldo Soligno is an emerging talent in Italian documentary photography. In October 2014, he 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. It investigates the needs, hopes, difficulties, but above all, the joys and daily achievements of those living a 'rare life'.

This project was carried out through home visits to 28 families in 7 European countries thanks to the collaboration with UNIAMO, the Italian Federation of Rare Disease associations. Through Aldo's work, he has inspired other projects, such as #MaketoCare, that in turn, have placed a spotlight on the rare disease community. The project was published in six major European magazines and was the object of several talks and presentations. It was also broadcast on the Italian national television channel RAI.

Aldo receives the Media Award for his dedication and continued efforts to increase the visibility of people living with a rare disease.

2017

COMPANY AWARD

GlaxoSmithKline

GSK is one of the world's largest pharmaceutical companies, developing pharmaceuticals, vaccines and consumer healthcare products. Beginning in 2010, the Company has dedicated rare diseases as a core therapeutic area. In 2016, years of collaboration between GSK, Fondazione Telethon and Ospedale San Raffaele brought to market an innovative gene therapy to treat patients with the rare disease Adenosine deaminase severe combined immunodeficiency syndrome (ADA-SCID). This advancement shows the results of partnership between patient organisations, clinicians, large and small companies to develop and make available treatments to meet unmet needs of rare disease patients.

GSK has been an active member of the EURORDIS Round Table of Companies (ERTC) since 2008 and has supported EURORDIS Membership Meetings, RareConnect, and the launching of the Rare Barometer Programme.

2017

PATIENT ORGANISATION AWARD

The Dravet Syndrome European Federation

EURORDIS is especially pleased to present the 2017 EURORDIS Patient Organisation Award to Dravet – the Dravet Syndrome European Federation. Dravet was founded in 2014 by eight patient organisations in seven countries, who have worked together to raise awareness and to fight the impact of Dravet Syndrome, otherwise known as Severe Myoclonic Epilepsy of Infancy (SMEI), on patients and their families.

The organisation now counts thirteen members and aims to help people in countries without local support and associations for Dravet Syndrome. Among many other actions, Dravet raises awareness about this rare disease, looks for solutions to improve the quality of life of patients and helps countries to create their own association.

Dravet continues to stimulate research and development into treatments by specialist physicians and researchers for the 1 in 20 000 individuals affected by this Rare Disease.

This incredibly well deserved Award serves to recognise the efforts and successes that have resulted from the hard work and determination of those at Dravet.

2016

EURORDIS BLACK PEARL AWARDS

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.

2016

LIFETIME ACHIEVEMENT AWARD

Renza Barbon Galuppi

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.

Firmly believing in the integration of disabled people in society, she became a scuba diving instructor for disabled people and President of the Parents Association for Rehabilitation through equestrian sports. Her daughter Laura won a medal at the Beijing Paralympics' Games in this sport!

In the past 10 years, as President of UNIAMO, 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.

Her vision of a rare disease community, translated into a project, brought for the first time, all the national key stakeholders, including patients' representatives, around the same table to share perspectives and objectives to improve the quality of care in order to overcome the inequalities linked to the regionalised health system. EURORDIS is pleased to award Renza with this Lifetime Achievement Award as it serves to honour all that Renza has done for the rare disease community.

2016

POLICY MAKER AWARD

Christian-Silviu Busoi

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 by training and a former lecturer in Public Health and Health Management at the Victor Babes University of Medicine and Pharmacy, Busoi has translated his expertise into concrete parliamentary action at the European Level. As a member of the ENVI Committee (on the Environment, Public Health and Food Safety) within the European Parliament, he has used this platform to champion patients' rights in each country, launching the public "Patients' rights" campaign, with particular attention to cross-border health care. Busoi has also advocated strongly for rare disease clinical trials, supporting a multi-centre collaboration at EU level to encourage partnerships between all rare disease stakeholders.

In specifically holding parliamentary events in support of rare cancers and rare diseases, such as on patient registries, and co-hosting the Rare Disease Day policy event to improve access to therapies for rare diseases, he has shown devotion and passion in addressing the needs of rare disease patients across Europe, making him a truly deserving winner of the EURORDIS Policy Maker Award 2016.

2016

EUROPEAN RARE DISEASE LEADERSHIP AWARD

European Commission

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. The three awardees have been instrumental in the development of European Union policy on rare diseases, consistently going above and beyond the call of duty to find solutions that have a positive impact for rare disease patients.

Antoni Montserrat Moliner became the main Policy Officer on Cancer and Rare Diseases at the European Commission almost a decade ago. His role was pivotal in the development and adoption of the Commission Communication in 2008 and the Council Recommendation on Rare Diseases in 2009, which shaped the European Union rare disease policy landscape. Montserrat has championed patient engagement, particularly in his involvement in EUROPLAN and National Plans. Appointed Senior Expert on Cancer and Rare Diseases within the Directorate of Public Health of the European Commission in 2015, 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). His background has shone through his understanding of the format and issues that are important to all rare disease stakeholders. 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. He has demonstrated leadership through his day-to-day, hands-on approach in supporting the whole community through the Commission Expert Group on Rare Diseases.

Michael Hübel, Head of the Unit of Programme Management and Diseases at DG-SANTE within the European Commission, has demonstrated strong and visionary leadership within this role. He 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. Hübel has remained devoted to rare diseases and respected in his leadership of the unit, offering constant direction and vision in his work.

2016

SCIENTIFIC AWARD

Prof. Dr. Peter N. Robinson

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. HPO is currently widely used in both research and clinical setting, and became a standard in describing human phenotypes, so contributing to make data interoperable and able to be shared for a better knowledge and recognition of rare diseases. 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).

The EURORDIS scientific award recognises the major impact Peter Robinson's work has for rare diseases. He has concentrated his diverse background and skills (he is mathematician, paediatrician, geneticist and bioinformatician) to improve the understanding and the diagnosis of inherited diseases.

He is a paradigmatic collaboration personality, sharing his vast and deep knowledge and achievements with other groups across the world, and allowing for the development of outstanding initiatives for the benefit of patients.

2016

MEDIA AWARD

France Télévisions - AFM-Téléthon

The EURORDIS Media Award recognises the long-standing support, for the past 30 years, of France Télévisions in broadcasting live the French Telethon organized in partnership with the AFM-Téléthon. Millions of people have taken part and donated to the AFM-Téléthon cause and thanks to this it has been 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, all leaders in research and development of biotherapies for rare genetic diseases. 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.

2016

COMPANY AWARD

Actelion

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, including PAH as well as other rare disorders.

Actelion has engaged with and supported patient organisations since its founding. This has especially helped raise awareness and understanding of PAH, including the importance of psycho-social support in disease management. 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.

2016

PATIENT ORGANISATION AWARD

UNIQUE

EURORDIS is especially pleased to present the organisation 'UNIQUE' as the 2016 Awardee of the Patient Organisation Award.

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.

2016

EURORDIS PHOTO AWARD

1st Public Vote by Erika Weis

'Feel free'

Pastic tetraparesis and high myopia

Romania

2nd Public Vote

by Alexander Zhdanovich

'Life is a swing.
I choose to fly up high'

Cri du chat syndrome

Belarus

3rd Public Vote by Alexandra Garikova

'Rare ballet for child
with aniridia'

Aniridia

Egypt

2015

EURORDIS BLACK PEARL AWARDS

2015

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. Rosa first entered the world of rare diseases in 1996 when 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.

Convinced that nothing could be achieved but at European level, 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. She has been a source of inspiration to many and she continues to be a faithful and long-standing supporter of EURORDIS.

2015

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. Pharmaceutical companies believed that larger markets for medicines represented more profitable drugs.

Ms Meyers recounts the moment she realised that, although she was fighting because of one little child, who happened to be her son, she couldn't possibly be the only family with this problem. To find out, she called up various support groups for other rare diseases asking 'Are you having this problem too?' and most of them said yes.

This realisation led her to found the National Organization for Rare Disorders (NORD) in the USA. 32 years later and NORD is an incredible 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. Although retired now, Ms Meyers continues to be an inspiration to rare disease patient advocates and was involved in the discussions around some of the earliest drafts of what later became the European Orphan Drug Regulation.

2015

POLICY MAKER AWARD

Glenis Willmott

Glenis Willmott, Labour Member of the European Parliament for the East Midlands in the UK since 2006 and three-time 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, and the reason she is so deserving of the EURORDIS Policy Maker Award, 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 and demonstrate Ms Willmott's devotion to improving the lives of people living with a rare disease.

EUROPEAN RARE DISEASE LEADERSHIP AWARD

Professor Josep Torrent-Farnell

Professor Josep Torrent-Farnell is a qualified Pharmacist and a specialist in Internal Medicine with a degree in Medicine and Surgery from the University of Barcelona as well as postgraduate courses in Pharmacology and Toxicology, Public Health and European Institutions and a doctorate in Clinical Pharmacology. He 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. During this time Josep established a high standard of scientific evaluation for designation and provided much scientific advice and assistance. 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.

2015

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.

Professor Bushby is actively involved in many European projects including being a founding co-ordinator of the TREAT-NMD Network of Excellence whose objective is to ensure that the most promising new therapies reach patients affected by neuromuscular diseases as quickly as possible.

Kate has played a leading role in the European and national rare disease policy area, acting as vice chair on the European Union Committee of Experts on Rare Diseases (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 along with Volker Straub and Hanns Lochmüller 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.

The EURORDIS Scientific Award recognises her outstanding research achievements into inherited neuromuscular diseases and her commitment to patients. Her impressive body of work has seen her become a leader in the rare disease community at the forefront of developments in the field of translational medicine. Moreover, Kate has continuously demonstrated her true commitment to patients at many levels. This award serves to recognise her long-standing contributions that have shaped rare neuromuscular disease research and will continue to fuel the future of rare disease research as a whole.

2015

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.

Peter continues to become increasingly active in the rare disease community, demonstrating commitment and passion. 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.

2015

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.

Pfizer has been an active participant in the EURORDIS Round Table of Companies (ERTC) since 2007, contributing to thoughtful dialogue with ERTC member companies, EURORDIS and other rare disease stakeholders to work towards accelerated development and availability of rare disease treatments and care in Europe. Pfizer has also joined other companies in fostering the empowerment of rare disease patient organisations through support of EURORDIS' capacity and community-building actions, such as the EURORDIS Membership Meetings. The Company continues to encourage partnerships with patient communities and advocacy organisations as well as nurture active dialogue with healthcare stakeholders and regulatory bodies.

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. Through their actions, companies should work to cooperate with and support the actions of patient advocacy groups.

It is for these reasons Pfizer is a highly deserving recipient of the EURORDIS Company Award 2015 and it is our pleasure to acknowledge the contributions of the Company and its employees in the area of rare

2015

PATIENT ORGANISATION AWARD

Children with SMA - Vitaliy Matyushenko

EURORDIS is especially pleased to present the Foundation "Children with SMA" as the 2015 Awardee of the Patient Organisation Award. "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.

SMA is a motor neuron disease characterised by the progressive degeneration of nerve cells in the spinal cord and brainstem, leading to muscle weakness, muscle atrophy, and respiratory complications. 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.

2015

EURORDIS PHOTO AWARD

1st Public Vote

by Alexander Zhdanovich

'Cantabrian warrior wounded in battle'

Osteogenesis Imperfecta

Spain

Instagram Winner

by Mary Ann Lana

"Eli and Lucy the horse'

Fanconi Anemia

America

Expert's Choice

'Easy Rider'

Spinale Muskelatrophie

Germany

2014

EURORDIS BLACK PEARL AWARDS

2014

VOLUNTEER AWARD

Lise Murphy

Lise Murphy is a true soldier of the rare disease movement in Europe. As an individual affected by the rare disease Marfan syndrome, she has had the experience this inherited disease across three generations, with her father and son also affected. Lise Murphy has used her experience and patient expertise generously for the benefit of all the rare disease community.

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.

EURORDIS would like to thank Lise Murphy for her tireless dedication as a volunteer, with her active involvement and continued commitment to the Therapeutic Action Group (TAG) and as co-chair of the Drug Information Transparency and Access (DITA) Taskforce.

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. Achievements within this position have included presenting the EMA model of working with patients before the Heads of Medical Agencies and at the EFPIA Think Tank in Brussels.

Lise Murphy 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. Her relentless dedication to EURORDIS and the rare disease community as a whole makes her a truly deserving recipient of the EURORDIS Volunteer Award 2014.

2014

LIFETIME ACHIEVEMENT AWARD

Marlene Haffner MD, MPH

For over 30 years, Dr Marlene Haffner has had an immeasurable impact upon the development of orphan drug therapies. She has dedicated most of her professional life to facilitating the development of therapies for the diagnosis, treatment and prevention of rare diseases.

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. In this role she was responsible for the administration of the US Orphan Drug Act, the first act of this type in the world. Her influence in this role impacted far beyond the USA, as she applied her valuable knowledge and experience to assist the development of similar orphan drug programmes in Japan, Australia, and the EU, amongst other countries and regions.

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.

Her success can easily be put into numbers: 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.

Dr Haffner graduated from George Washington School of Medicine and completed further training at the Columbia University School of Medicine and the Albert Einstein College of Medicine, New York City, before undertaking a Master's degree in Public Health from the Johns Hopkins Bloomberg School of Public Health in Baltimore. 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. Without such tenacity and ingenuity, many of the orphan drug programmes around the world would not exist today.

2014

POLICY MAKER AWARD

Antonyia Parvanova

Dr Antonyia Parvanova, Member of the European Parliament since 2009, has shown an outstanding commitment to rare diseases in her overarching objectives to improve Public Health across the European Union's Member States.

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.

Dr Parvanova has publically voiced the importance of equality in access to health services, to provide affordable, high quality and safe medical care for all European citizens.

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. As Rapporteur of the EU Directive on Medicinal Products for Human Use: transparency of measures regulating the prices, Dr Parvanova has shown substantial commitment to improving rare disease patients' lives.

Dr Parvanova studied Medicine and Health Management in Varna, Public Health at Maastricht (the Netherlands) and health policy in England.
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.

EURORDIS has benefited from the support of Dr Parvanova, who has participated as a speaker at several EURORDIS events. This award recognises her unique dedication in addressing the needs of people living with rare diseases as a Member of the European Parliament, and in her support of crucial amendments in several EU pieces of legislation that have a positive impact on the rare disease community at large.

POLICY MAKER AWARD

Paola Testori Coggi, Guido Rasi, Luca Pani

In 2014 EURORDIS has decided to jointly award three leaders for the European Leadership award to recognise their leadership and support for rare diseases and their capacity for far-reaching, innovative policies.

Our intention in awarding three Italians is to pay tribute to their pioneering European spirit and continuation of the work that other renowned Italians began, such as Altiero Spinelli and Alcide De Gasperi in the construction of the European Union as well as more recently Mario Monti.

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.

2014

POLICY MAKER AWARD CONTINUED

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.

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.

2014

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. Between 1984 and 1997, he headed the Institute for Human Genetics at the University of Nijmegen, the Netherlands, and since 1987, he has been board-certified as a Clinical Geneticist.

Dr Ropers has made many contributions to the molecular elucidation of monogenic disorders by positional cloning, with a focus on eye diseases, deafness and particularly mental retardation (MR).

His department forms part of the European MRX Consortium which plays a central role in researching the molecular causes of X-linked mental retardation, 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.

Dr Ropers has also set out to study autosomal recessive forms of MR and related disorders in a systematic manner, employing whole genome SNP typing for homozygosity mapping in large consanguineous families in developing countries. 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. Dr Ropers has served on numerous editorial boards and is member of the Royal Netherlands Academy of Arts and Sciences and of the Berlin-Brandenburg Academy of Sciences.

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.

2014

MEDIA AWARD

Rick Guidotti

Rick Guidotti is "changing the world, one picture at a time".

A graduate of New York's School of Visual Arts, Rick Guidotti led a successful career as a fashion photographer, working between New York, Paris and Milan for high profile clients such as the fashion house Yves Saint Laurent and magazines Elle and Harpers Bizarre. 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.

His work has been recognised internationally, raising public awareness for the beauty of difference. He has given confidence, self-belief and humanity to people frequently defined by their diagnosis. 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.

2014

COMPANY AWARD

Sobi

Sobi, established in 2001, is an international healthcare company dedicated to rare diseases, focusing on developing innovative treatments across four key therapeutic areas: haemophilia, inflammation/autoimmune diseases, inherited metabolic diseases and oncology.

The two EU authorised treatments, with a third that is provided on special license, is supported by Sobi's many other products in development. Sobi is particularly sensitive to the need for targeted treatments that are accessible to paediatric populations.

As a pharmaceutical company Sobi is dedicated to working with all stakeholders to develop successful treatments for rare disease patients. Their strong involvement in the unique initiative, Alkaptonuria DevelopAKure consortium, is exemplary of this.

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).

Sobi supports networking and capacity-building for rare disease patient advocates via the EURORDIS Membership Meeting. This award recognizing the excellence and consistency of Sobi's work in the rare disease community also marks 10 years of membership in the EURORDIS Round Table of Companies (ERTC).

2014

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.

The company's encouragement of staff to volunteer at therapeutic recreation summer camps serving children with serious illness is a further example of Orphan Europe's commitment to the rare disease community.

Orphan Europe is one of the industry representatives within the Commission Expert Group on Rare Diseases (replacing the EUCERD). A member of the EURORDIS Round Table of Companies (ERTC), Orphan Europe also consistently supports networking and capacity-building for rare disease patient advocates via the EURORDIS Membership Meeting.

2014

PATIENT ORGANISATION AWARD

Allianz Chronischer Seltener Erkrankungen

Allianz Chronischer Seltener Erkrankungen (ACHSE), the German National Alliance for Rare Diseases was founded in 2004. The umbrella organisation represents 120 rare disease patient organisations in Germany.

ACHSE 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. This is especially true in recognising the outstanding achievement of ACHSE in its contribution to the progress of the German National Plan for Rare Diseases, (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 promotes research into rare diseases by empowering its member organisations, through networking activities, and by coordinating the selection process of its scientific board, which proposes the awardee of the annual Eva Louise Köhler Research Award. This award offers € 50 000 towards funding rare disease research projects.

EURORDIS are grateful for ACHSE's cooperation in the Council of National Alliances, which brings together national experiences and strategies to produce a strong European framework. ACHSE is exemplar of a valuable and effective platform for rare diseases, making them most deserving recipients of the EURORDIS Patient Organisation Award 2014.

2014

EURORDIS PHOTO AWARD

Honorable mention

by Svetlana Gombats

'We be of one blood, ye and I'

Cri du Chat syndrome

Belarus

Expert's Choice

by Borodina Kseniya

'I was born and that is all that needs to be happy!'

Williams syndrome

Russia

Favourite Prize

by Jeanette Dunne

'The world through her eyes'

Diploid-triploid mosaic syndrome

Ireland

1st Public Vote

by Marina Klaric

'My hero'

Niemann-Pick disease type C

Croatia

2013

EURORDIS BLACK PEARL AWARDS

2013

VOLUNTEER AWARD

Lesley Greene

Lesley Greene is a true pioneer of the rare disease movement in Europe. In 1980, upon the diagnosis of her first-born 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. Twelve years later, 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.

Her experience as a mother of a daughter affected by a rare metabolic disease and her background as a teacher have enabled her to speak at many conferences for patients, regulators and industry, and to serve on many committees over the past 30 years. Her commitment to the rare disease movement has never faltered and she is truly deserving of this EURORDIS Volunteer Award.

2013

LIFETIME ACHIEVEMENT AWARD

Eva Luise Köhler

In 2004, Horst Köhler was elected Federal President of Germany (Mandate: 1 July 2004 – 31 May 2010). 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 German National Alliance for Chronic Rare Diseases: ACHSE. She is also the Chair of the Board of Trustees of the Eva Luise and Horst Köhler Foundation for people with rare diseases, created in March 2006.

Specifically, the Eva Luise and Horst Köhler Foundation supports research in the field of rare diseases. Its primary aim is to make sure that people with rare diseases receive an accurate diagnosis as early as possible so that they can be treated competently and effectively. 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.

"With the Foundation, we want to initiate and promote research projects improving the diagnosis, treatment, research and support programmes."

Eva Luise Bohnet was born on 2 January 1947, in Ludwigsburg, Germany. After graduating in 1966, she studied German and History at the Pädagogische Hochschule in Ludwigsburg. In 1975, she took the second qualification examination for teaching at primary and secondary schools in Herrenberg. In 1969, she married Horst Köhler, a senior research fellow at the Tübingen Institute for applied economic research. Her daughter and son were born in 1973 and 1977 respectively. Between 1969 and 1977, Eva Luise Köhler worked as a teacher at a specialised school for children and young people with learning disabilities before taking a position as a primary school teacher in Bonn.

Ms Köhler's daughter is affected by the rare disease Retinitis pigmentosa, a degenerative eye disease that causes severe vision impairment and often blindness. Ms Köhler's daughter has become blind due to this disease.

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. Mrs Köhler's achievements in this field have inspired other First Ladies in various other European countries including Mrs Sandra Elisabeth Roelofs, First Lady of Georgia and Mrs May Panou Papoulia, First Lady of Greece.

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.

Despite fierce opposition and often very sensitive political situations, 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.

These major legislative steps, her unique dedication and her relentless commitment to addressing the needs of people living with rare diseases, have created a favorable environment for therapy development in Europe, and positively impacted the lives of rare disease patients and their families, thus making her the ideal recipient for the EURORDIS Policy Maker Award.

2013

EUROPEAN RARE DISEASE LEADERSHIP AWARD

Dr Ruxandra Draghia-Akli

Dr Ruxandra Draghia-Akli (MD, PhD) is Director of the Health Directorate at the Research & Innovation DG of the European Commission.

Dr Draghia-Akli 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 EU has taken the lead in creating an unprecedented international effort – IRDiRC – in April 2011, to foster international collaboration in rare disease research, a highly challenging area of medical research that has the potential to benefit tremendously from the recent advances in genomics, proteomics and other omics technologies.

IRDiRC now brings together the EU itself with France, Germany, Spain, UK, USA, Canada, Australia, leading pharma companies and pioneering biotechs, EURORDIS, NORD and more. The ambitious goal of this International Consortium is to develop 200 new therapies for rare diseases and the means to diagnose most rare diseases by the year 2020.

For her role in these initiatives and for her support of development of orphan medicinal products to treat rare diseases, Dr Ruxandra Draghia-Akli is the recipient of the EURORDIS European Rare Disease Leadership Award.

2013

SCIENTIFIC AWARD

Ségolène Aymé

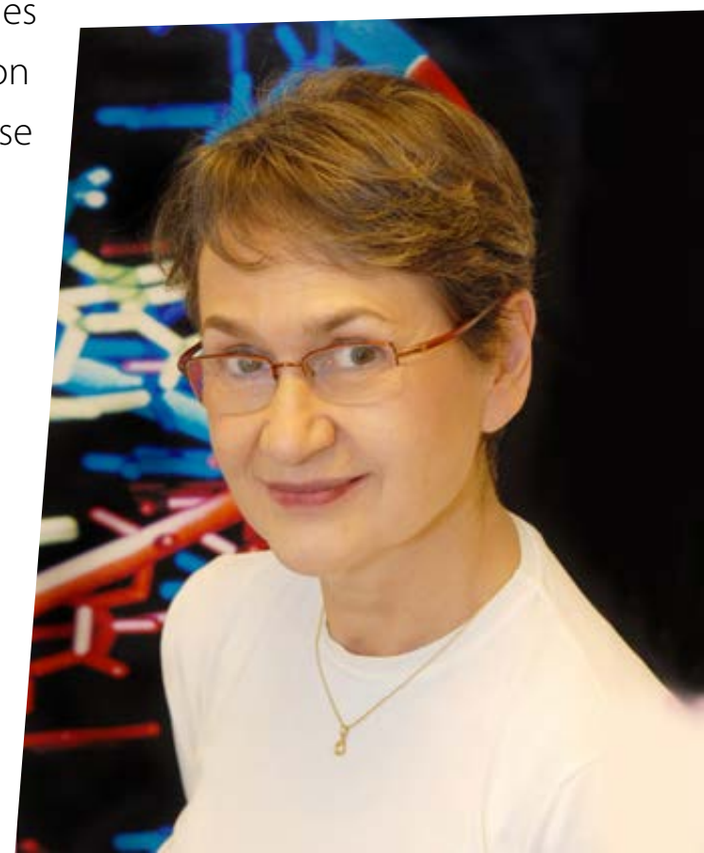
Dr Ségolène Aymé is a medical geneticist and Emeritus Research Director at the French National Institute of Health and Medical Research (INSERM). She developed Orphanet, the world's leading reference portal for expert validated rare disease and orphan drug information.

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. Orphanet is an exemplary international collaboration.

Dr Ségolène Aymé also serves as Chair of the European Committee of Experts on Rare Diseases (EUCERD), heads the Scientific Secretariat of the International Rare Disease Research Consortium (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 designed to improve the situation for rare disease patients and those who care for them.

Her unflagging energy, intelligence and dedication are an inspiration for the rare disease community and make Dr Ségolène Aymé truly deserving of this EURORDIS Scientific Award.



2013

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 London. He was the Financial Times' Moscow correspondent and then bureau chief from 1998 to 2004, and previously served as Paris correspondent, financial correspondent, general reporter and corporate reporter.
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.

He is author, most recently, of Inside Putin's Russia and The French Exception. Mr Jack has written articles for medical journals including the British Medical Journal and the Lancet.
He has written specialist reports on the French insurance industry, audit committees, networking and work shadowing; as well as chapters in books on Russia, ethics, and financial reporting.

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.

2013

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. Moreover, the Company's 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. This service provides patients and healthcare professionals with a dedicated, central point of contact to assist in navigating the challenges of reimbursement, providing information about co-pay assistance, and engages in local patient access and compassionate programmes.

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. Celgene is also a longstanding sponsor of EURORDIS Membership Meetings and contributes to the EURORDIS Round Table of Companies at the highest level, both in terms of membership level and quality participation.

Celgene is being presented with a EURORDIS Company Award in recognition of the Company's steadfast commitment to generating treatments for rare diseases.

2013

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. This alliance was established under GSK's Centre of Excellence for External Drug Discovery, which seeks to collaborate with companies at the leading edge of highly innovative and transformative science.

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).

EURORDIS is presenting Prosensa with this EURORDIS Company award in recognition of its innovation and promise for the future.

2013

COMPANY AWARD

Genzyme

Genzyme is a pioneer in researching, developing and marketing medicinal products for patients living with diseases. Founded in 1981 to develop a treatment for Gaucher disease, Genzyme launched the world's first enzyme replacement therapy in 1991. Now part of Sanofi, Genzyme, a Sanofi Company continues to offer hope to people living with genetic diseases, endocrine and cardiovascular diseases.

Genzyme has taken seriously its responsibility to increase patients' access to the life-saving therapies it has developed. 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. Over time, similar programs have been developed to distribute new therapies for other diseases.

Genzyme supports numerous patient organizations worldwide. 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.

EURORDIS is presenting Genzyme, a Sanofi Company with a EURORDIS Company Award in recognition of Genzyme's, pioneering achievements to the benefit of rare disease patients and actions to ensure patients' access to life-saving products.

2013

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.

Kay created Alström Syndrome UK in 1998 with three key aims:

- to help people with Alström Syndrome to provide support for their families, their care givers and the professionals who are working with them,
- to raise awareness amongst both the public and medical professions of Alström Syndrome,
- to raise funds for research into Alström Syndrome.

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.

2013
EURORDIS PHOTO AWARD

1st by Eleni Maggouta

'Don't let disability rule your life'

Friedreich ataxia

Greece

2nd by Laurent Reininger

'Unique in the world but not for celebrity '

Mitochondrial Cardiomyopathy

France

3rd by Marian VÃ¡rka

'Shaggy but lucky'

Dystrophic epidermolysis bullosa

Slovakia

2012

EURORDIS BLACK PEARL AWARDS

2012

VOLUNTEER AWARD

Michele Lipucci Di Paola

Michele Lipucci Di Paola, PhD is a long standing dedicated volunteer of EURORDIS. He is former Vice President of the EURORDIS Board of Directors and was a member of the Board between 1997-2005.

As a result of the diagnosis of a family member with Thalassemia, he and his organisation AVLT (Associazione Veneta Lotta alla Talassemia) worked in cooperation with other international thalassemia associations to create and provide support to an international network of researchers and clinicians dedicated to identifying new clinical protocols and innovative therapeutic approaches such as gene therapy for this disease.

The result of this activity was support to national and international research projects.

Michele holds a PhD in plant biology and is Associate Professor at the University of Pisa, in addition to serving as patient representative member on the EMA

Committee for Advanced Therapies (CAT). Dr. Lipucci Di Paola exemplifies the international movement to improve conditions for people living with rare diseases. His numerous affiliations and achievements have made and will continue to make an important difference at both the national and international levels.

2012

POLICY MAKER AWARD

Androulla Vassiliou

Currently European Commissioner for Education, Culture, Multilingualism and Youth, Androulla Vassiliou, JD, served as Commissioner for Health and Consumer Policy from February 2008 to end of 2009. During this time, she was instrumental in making significant strides for European citizens living with a rare disease. Of particular importance were her political support that led to the launch of the Public Consultation, Rare Diseases: Europe's Challenge, and the consequent adoption of the Commission Communication on Rare Diseases in November 2008, as well as the Council Recommendation on Rare Diseases in June 2009.

These two documents have established an overall and comprehensive policy framework, with an EU-wide and integrated strategy between EU and national levels on rare diseases related issues such as information, patient empowerment, research, diagnosis, treatment and care for rare disease patients throughout Europe.

EUROPEAN RARE DISEASE ACHIEVEMENT

Kerstin Westermark,

Kerstin Westermark, MD, PhD, will complete her second term as Chairperson of the Committee for Orphan Medicinal Products (COMP) in 2012 at the European Medicines Agency - EMA.

She has been a member of the COMP, first as Swedish delegate from 2000-2006 and then as Chairperson from 2006. She was the first woman to be elected chair of an EMA scientific committee.

During her mandate as COMP Chairperson, she has established close collaboration with the FDA and has encouraged interactions with other international agencies. Kerstin has enhanced close collaboration with the rare disease community, patient advocates and EURORDIS.

Kerstin currently heads a national centre for patients with Wilson Disease. She is a senior expert at the Swedish Medical Products Agency in Uppsala, where she has also served as Head of the Department of Clinical Trials. Kerstin is an Adjunct Professor of Medicine at the Uppsala University Faculty of Medicine.

2012

SCIENTIFIC AWARD

Professor Alain Fisher Hôpital Necker - Enfants Malades, France

and Professor Maria Grazia Roncarolo The San Raffaele Telethon Institute for Gene Therapy, Italy

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.

Severe Combined Immune Deficiencies (SCID) represent a collection of rare diseases caused by gene defects that prohibit the development and/or function of disease-fighting immune cells. Beginning in the first few months of life, these rare disorders are life-threatening and often lethal due to susceptibility to infections as well as complications due to the inflammatory and autoimmune responses. A bone marrow stem cell transplant from a compatible donor is one treatment option for these patients, but since there is a considerable lack of suitable bone marrow donors, there was a high need for alternative treatments.

Two European teams of researchers and clinicians have achieved the successful development of the first gene therapy for two of these rare diseases. This represents a commendable example of outstanding scientific research and collaboration that has resulted in a tremendously positive impact on the life of SCID patients and 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.

2012

SCIENTIFIC AWARD CONTINUED

At the Necker Hospital in Paris, the longstanding focus of the INSERM team has been to characterise the genetic and molecular bases of inherited immunodeficiencies, identifying the causes of approximately 25 of them (T-cells deficiency, IgG and IgA deficiency, familial lymphohistiocytosis and related diseases, autoimmune lymphoproliferative syndrome, etc). Their work has contributed to a better understanding of how the human immune system functions as well as enabling the development of diagnostic tests and, most importantly, to lay down the basis for the gene therapy of the X-linked severe combined immunodeficiency (XL-SCID). This has now been successfully applied for close to 13 years and the results have led to a gradual extension of the application of this therapy for other inherited diseases of the hematopoietic system.

At the San Raffaele Hospital in Milan, the team of the Telethon Institute for Gene Therapy (TIGET) has focused on the ADA-SCID which is a form of SCID due to a mutation in the adenosine deaminase (ADA) gene, required for the generation of immune competent cells. ADA-SCID affects approximately 350 newborns worldwide every year. The team has established a novel gene therapy which is based on transferring a healthy ADA gene into the patient's own hematopoietic stem cells. These cells are reintroduced in patients who are pre-treated in a way that allows optimal outgrowth of the healthy stem cells and the generation of a competent immune system. It has been shown that this therapy is safe and results in a complete cure providing effective protection against severe infections. The TIGET team is presently developing gene therapy for another immunodeficiency (Wiskott-Aldrich syndrome) and for a severe metabolic disease (Metachromatic leukodystrophy).

2012

MEDIA AWARD

BBC

The BBC has been covering rare diseases and helping to raise awareness about the challenges faced by rare disease patients for over 3 decades. Indeed, the BBC's Songs of Praise in 1982 helped EURORDIS Past President, Lesley Greene, to find other 'orphan parents' and specialists for her daughter's rare metabolic disease and subsequently establish a patient group. The BBC also commissioned a programme called Diagnosis in 2011 featuring several rare diseases.

In addition, and on many occasions, a rare disease is featured on the popular medical dramas Casualty and Holby City. Indeed, Rosie Marcel who plays "nasty Jak" in Holby City recently revealed she has Behçets Syndrome. Furthermore, through its grants programme, BBC Children In Need regularly supports many projects linked to rare diseases which would otherwise struggle to obtain funding. The visibility of such broadcasts and the BBC Children In Need appeal provide huge publicity, support and networking opportunities which are invaluable to a community which struggles because of its "unpopular" image.

Over the years, the work of the BBC has put a face to rare diseases and highlighted important issues for people living with rare diseases, such as delayed or inaccurate diagnosis, difficulty accessing care, financial burden and tremendous feeling of isolation.

2012

COMPANY AWARD

Shire

Shire established a Human Genetics Therapies (HGT) unit two decades ago to develop novel products for patients diagnosed with Rare Diseases. Since then, the Company has developed treatments for several rare conditions such as Hunter Syndrome and Gaucher Disease and put products on the market in more than 50 countries around the globe.

Most recently, Shire concluded the trials and regulatory submission for approval of a treatment for Hereditary Angioedema.

Shire has shown steadfast support for Rare Disease patient organisations, is an Emerald Member of the EURORDIS Round Table of Companies and contributor of grant funding for projects and events such as the EURORDIS Membership Meetings.

2012

COMPANY AWARD

CSL Behring

CSL Behring has a more than 90-year heritage of innovation, achievement and commitment to address diseases that are rare.

Today, with one of the broadest product portfolios for rare diseases, CSL Behring has received approvals for 15 rare disease products in the last seven years. In particular in 2011, European authorities approved a treatment of Hereditary Angioedema, and the European Commission granted marketing authorisation - valid for all Member States - for CSL Behring's treatment of primary immunodeficiency diseases and secondary immunodeficiencies.

CSL Behring has a strong record of positive collaboration with patient organisations, including EURORDIS, where they are a long-standing Emerald member of the EURORDIS Round Table of Companies and contributor of grant funding for projects such as the European Patient's Preferred Rare Diseases Policy Scenarios Project-POLKA.

CSL Behring

Biotherapies for Life™

2012

COMPANY AWARD

Sigma Tau Pharmaceuticals, Inc

The Sigma-Tau Group was founded by Dr. Claudio Cavazza in 1957 and the United States operations, Sigma-Tau Pharmaceuticals, was created in 1980 and became an early leader in developing medicines for rare diseases. By 1984 it was one of the first companies to receive an Orphan Drug Designation in the United States.

Sigma-Tau now provides seven innovative medicines for patients with rare diseases and has several more in late stage development. Sadly, Dr. Cavazza passed away last year but his passion to improve patient health lives on in the company's development and commercialisation strategies. Sigma-Tau is a founding member of the National Organization for Rare Disorders (NORD) Corporate Council. Sigma-Tau also supports EURORDIS and its key projects, including the publication of The Voice of 12,000 Patients and the Rhapsody and POLKA projects. Sigma-Tau is a Ruby member of the EURORDIS Round Table of Companies.

2012

PATIENT ORGANISATION AWARD

Association Française des Myopathies

The French Muscular Dystrophy Association (AFM) federates patients with neuromuscular diseases and their parents. Thanks in great part to donations from France's annual Telethon (€90 million in 2010), the AFM has become a major player in biomedical research into rare diseases in France and worldwide.
It is currently funding 36 clinical trials on 31 different genetic diseases affecting the eyes, blood, brain, immune system and muscles, to name but a few.

The French Muscular Dystrophy Association has boosted many research projects and, thanks to previous Telethons, has also created its own research tools, alone or in partnership with public institutions.

Today, the AFM relies on 4 "laboratories of excellence" which are leaders in their fields: Généthon and the Atlantic Institute of gene therapy for gene therapy; I-Stem Institute for stem cells; the Institute of Myology for the research and the treatment of muscular diseases. To move faster towards therapeutic successes, the AFM strengthens the coordination, the complementarity and the interactions between these laboratories by gathering them under one common banner: the Institute of Biotherapies. With the Généthon bio-production centre, the AFM stands out through its unique ability to produce and test its own gene-based medicines.

EURORDIS

Black Pearl

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