



[Press release]

## Announcement: Winners of the EURORDIS Black Pearl Awards 2018

*Stars of the rare disease community recognised for their efforts through the EURORDIS Black Pearl Awards.*

**#EURORDISAwards2018**

**15 January 2018, Paris** – The recipients of the EURORDIS Black Pearl Awards 2018 have been announced as:

EURORDIS Young Patient Advocate: [Sammy Basso, Associazione Italiana Progeria Sammy Basso, Italy](#)

EURORDIS European Rare Disease Leadership Award:

[Bruno Sepodes, Chair, Committee of Orphan Medicinal Products, European Medicines Agency, Portugal](#)

EURORDIS Policy Maker Award: [Elena Gentile, Member of the European Parliament, Italy](#)

EURORDIS Scientific Award: [Professor Michele De Luca, University of Modena and Reggio Emilia, Italy & Dr Tobias Hirsch, BG University Hospital Bergmannsheil, Germany](#)

EURORDIS Volunteer Awards:

[Chris Sotirelis, United Kingdom](#)

[Helene and Mikk Cederroth, Wilhelm Foundation, Sweden](#)

EURORDIS Patient Organisation Award: [European Pulmonary Hypertension Association](#)

EURORDIS Company Award for Innovation: [Novartis](#)

EURORDIS Company Award for Patient Engagement:

[The PARADIGM Project \(the European Federation of Pharmaceutical Industries and Associations-EFPIA, Bayer, UCB and MSD\)](#)

EURORDIS Visual and Audio Media Award: [Special Books by Special Kids, Christopher Ulmer, USA](#)

EURORDIS Written Media Award:

['On peut changer le monde en vendant des crêpes et des ballons', Serge Braun, AFM-Téléthon, France](#)

EURORDIS Lifetime Achievement Award: [Alastair Kent OBE, United Kingdom](#)



The EURORDIS Black Pearl Awards are presented to the stars of the rare disease community - patient advocates, organisations, policy makers, scientists, companies and media - for their major achievements and their outstanding commitment to the rare disease cause. Nominations are submitted by the general public, shortlisted by a nominations committee and then selected by the EURORDIS Board of Directors.

There were **over 350 nominations, representing 37 different countries** worldwide.

The EURORDIS Black Pearl Awards Ceremony takes place in Brussels on 20 February to celebrate [Rare Disease Day](#), the global awareness-raising campaign for rare diseases.

To attend the event in person [find out more about ticket prices](#) or to watch the ceremony online, join the livestream at 20:15 CET via [blackpearl.eurordis.org/live](http://blackpearl.eurordis.org/live).



## The EURORDIS Photo Award

The [EURORDIS Photo Award](#), the remaining EURORDIS Black Pearl Award, is an opportunity to visually express the reality of living with a rare disease on a daily basis and to share your rare disease story.

Anyone around the world can [submit a photo](#) before 31 January 2018. World-renowned National Geographic photojournalist [Marcus Bleasdale](#) will select a shortlist of 5 photos that will be opened to a public vote. **Online voting will open on 13 February** and the winner will be announced live at the EURORDIS Black Pearl Awards Ceremony on 20 February (the three finalists with the most votes will each receive a [prize](#)).

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## EURORDIS-Rare Diseases Europe

EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of over 700 rare disease patient organisations from more than 60 countries that work together to improve the lives of the 30 million people living with a rare disease in Europe.

By connecting patients, families and patient groups, as well as by bringing together all stakeholders and mobilising the rare disease community, EURORDIS strengthens the patient voice and shapes research, policies and patient services. Follow [@eurordis](#) or see the [EURORDIS Facebook page](#). For more information, visit [eurordis.org](#).

## Rare diseases

The European Union considers a disease as rare when it affects less than 1 in 2,000 citizens. Over 6,000 different rare diseases have been identified to date, affecting an estimated 30 million people in Europe and 300 million worldwide. Due to the low prevalence of each disease, medical expertise is rare, knowledge is scarce, care offering inadequate and research limited. Despite their great overall number, rare disease patients are the orphans of health systems, often denied diagnosis, treatment and the benefits of research.

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## Full details of recipients of the EURORDIS Black Pearl Awards 2018:

### EURORDIS Young Patient Advocate: **Sammy Basso, Associazione Italiana Progeria Sammy Basso, Italy**

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 create his own advocacy group, l'Associazione Italiana Progeria Sammy Basso, which has been instrumental in informing the general public about, as well as to promote the need for, Progeria research.

Together with Bologna's Institute for Molecular 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 has plans to become a researcher and contribute actively to the study of Progeria, for which he has already been instrumental in raising awareness.

### EURORDIS European Rare Disease Leadership Award: **Bruno Sepodes, Chair, Committee of Orphan Medicinal Products, European Medicines Agency, Portugal**

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. He became Chair of the COMP in 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 collaboration with 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.

### EURORDIS Policy Maker Award: **Elena Gentile, Member of the European Parliament, Italy**

Elena Gentile is an Italian politician who has supported the rare disease cause since she began practicing 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; and has been determined 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 has supported 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.

### **EURORDIS Scientific Award: Professor Michele De Luca, University of Modena and Reggio Emilia, Italy & Dr Tobias Hirsch, BG University Hospital Bergmannsheil, Germany**

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.

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 the 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 policy-making 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®.

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 Volunteer Award: **Chris Sotirelis, United Kingdom**

Chris Sotirelis is a patient with beta thalassaemia major and has been a tireless patient advocate for EURORDIS and the Rare Disease community at national and international level for many years. As a previous UK Thalassaemia Society Trustee, 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 being the UKTS 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 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. Closely 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.

### EURORDIS Volunteer Award: **Helene and Mikk Cederroth, Wilhelm Foundation, Sweden**

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' concept, 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.

### EURORDIS Patient Organisation Award: **European Pulmonary Hypertension Association**

The recipients of the Patient Organisation Award, PHA Europe, are dedicated to improving the lives of patients living with pulmonary hypertension (PH) in Europe by working with its members to enhance awareness of PH, promote optimal standards of care for people living with the disease, ensure the availability of all approved treatments and encourage 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.

### EURORDIS 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.

### EURORDIS Company Award for Patient Engagement: **The PARADIGM Project (the European Federation of Pharmaceutical Industries and Associations-EFPIA, 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. It is indeed timely to provide a framework that allows structured, meaningful, sustainable and ethical patient engagement throughout the development of medicinal products. On the longer term, this framework will strengthen both the understanding of stakeholders and the systems-readiness towards patient engagement across the diverse range of stakeholders, and to ensure maximum synergies with other initiatives focusing on the patient's voice in the life cycle of medicines.

Furthermore, the development of an inventive and workable sustainability roadmap to optimise patient engagement in key decision-making points 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 in the lifecycle of medicines.

## EURORDIS Visual & Audio Media Award: **Special Books by Special Kids, Christopher Ulmer, USA**

The recipient of the Visual & Audio Media 2018 is Christopher Ulmer, who, in 2016, 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.

## EURORDIS Written Media Award: **'On peut changer le monde en vendant des crêpes et des ballons', Serge Braun, AFM-Téléthon, France**

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 has 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 focuses on 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.

## EURORDIS Lifetime Achievement Award: **Alastair Kent OBE, United Kingdom**

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.