2016 EURORDIS Awards for Excellence in the Field of Rare Diseases

23 February 2016, Brussels – Her Royal Highness Princess Astrid of Belgium today presents the 2016 EURORDIS Awards at the EURORDIS Awards and *Black Pearl* Evening in Brussels.

The evening, held to mark the occasion of Rare Disease Day 2016 (29 February), is held by EURORDIS, the European Organisation for Rare Diseases.

The EURORDIS Awards recognise outstanding patient advocates and organisations, volunteers, scientists, companies, media and policy makers who have contributed to reducing the impact of rare diseases on people's lives.

Also in attendance of the event is Rare Disease Day Ambassador Sean Hepburn Ferrer, the eldest son of the late Audrey Hepburn, who passed away from a rare cancer.

Yann Le Cam, Chief Executive Officer, EURORDIS, commented, "The rare disease patient organisations, researchers, patient advocates, volunteers, policy makers, companies and leaders that win the EURORDIS Awards show exceptional commitment to supporting people affected by, or people that are living with someone affected by, a rare disease. The EURORDIS Awards are an important opportunity every year to take stock and recognise the hard work of the various individuals and organisations that help reduce the impact of rare diseases, and encourage others to do the same."

Nominations for the EURORDIS Awards 2016 were submitted by the general public with the EURORDIS Board of Directors making the final decision. The award winners are listed below (see end of press release to read full profiles of each winner and details of their commitment to the field of rare diseases). Photos of the award winners are available on the EURORDIS Flickr page.

Policy Maker Award
Cristian-Silviu Buşoi, Member of the European Parliament, Romania

Volunteer Award
Tsveta Schyns-Liharska, Belgium/Bulgaria

Media Award
France Télévisions - AFM-Téléthon, France

Patient Organisation Award
UNIQUE - The Rare Chromosome Disorder Support Group, UK

Scientific Award
Prof. Dr. Peter N. Robinson, Institute for Medical Genetics, Universitätsklinikum Charité, Germany

European Rare Disease Leadership Award
Joint winners: Antoni Montserrat Moliner, Jarek Waligóra and Michael Hübel, Directorate General of Health and Food Safety (DG-SANTE) within the European Commission, Belgium

Company Award
Actelion Pharmaceuticals Ltd., Switzerland

Lifetime Achievement Award
Renza Barbon Galuppi, Italy

###

About the EURORDIS Awards & *Black Pearl* Evening

The EURORDIS Awards and *Black Pearl* Evening serves: to recognise the individuals and organisations that together are improving the lives of people affected by rare diseases; to raise awareness and increase support for the rare disease cause throughout Europe; and to generate funding for EURORDIS programmes. For more information, please visit www.blackpearl.eurordis.org.

About Rare Disease Day

Rare Disease Day was launched by EURORDIS and its Council of National Alliances in 2008. Held on the last day of February each year, a rare day, it seeks to raise awareness of the impact that rare diseases have on the lives of patients and those who care for them. What began as a European event quickly became international in scope, with participants from more countries joining each year. See the Rare Disease Day 2016 video.

Since Rare Disease Day began, thousands of events have been held throughout the world, reaching hundreds of thousands of people. The political momentum resulting from the Day has also served advocacy purposes, contributing to the advancement of EU policies on rare diseases and the creation of national plans for rare diseases in a number of EU Member States and now in other countries. For more information visit RareDiseaseDay.org.

About Rare Diseases

The European Union considers a disease as rare when it affects fewer than 1 in 2,000 citizens. Over 6000 different rare diseases have been identified to date, affecting over 60 million people in Europe and the USA alone. 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.

About EURORDIS

EURORDIS, the European Organisation for Rare Diseases, is a non-governmental patient-driven alliance of patient organisations representing over 700 rare disease patient organisations in 63 countries. EURORDIS represents the voice of an estimated 30 million people living with a rare disease in Europe. Follow @eurordis or see the EURORDIS Facebook page. For more information visit www.eurordis.org.

Press contacts

Eva Bearryman
Junior Communications Manager
EURORDIS
Tel: +33 (0)1 56 53 52 61
eva.bearryman@eurordis.org

Lara Chappell
Communications Director
EURORDIS
Tel: +33 (0)1 56 53 52 60
lara.chappell@eurordis.org

Full details of award winners

Policy Maker Award – Cristian-Silviu Buşoi MEP, Romania

Cristian-Silviu Buşoi, 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, Buşoi 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. Buşoi 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.

Volunteer Award - Tsveta Schyns-Liharska, Belgium/Bulgaria

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

Media Award - France Télévisions - AFM-Téléthon, France

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.

Patient Organisation Award - UNIQUE - The Rare Chromosome Disorder Support Group, UK

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.

Scientific Award - Prof. Dr. Peter N. Robinson Institute for Medical Genetics, Universitätsklinikum Charité, Germany

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.

European Rare Disease Leadership Award - Joint winners: Antoni Montserrat Moliner, Jarek Waligóra and Michael Hübel, Directorate General of Health and Food Safety (DG-SANTE) within the European Commission, Belgium

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.

Company Award - Actelion, Switzerland

Established in 1997, Actelion focuses on the discovery, development and commercialization of innovative drugs for diseases with significant unmet medical needs. The Company has, in particular, made a difference for pulmonary arterial hypertension (PAH) in Europe and globally. Actelion's pipeline reflects continued commitment to address unmet medical needs, 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.

Lifetime Achievement Award - Renza Barbon Galuppi, Italy

Renza Barbon Galluppi is believed to be a "wonder woman" for her ability and strength to be where she is needed at the right moment, even though it means attending conferences, meetings and workshops in three different cities in two days. It has been calculated that in the past few years she has spent an average of 15 hours a day in activities related to Rare Diseases advocacy and to answering individual patients or Patient Organisations and organizing projects to train Rare Disease patients' representatives on key topics. It all started with the diagnosis of a typical type of hyperphenylalaninemia given to two of her three children. The delay in the delivery of the diagnosis to her eldest daughter and its consequences led her first to start the collaboration with the Patient Organisation involved in metabolic diseases but shortly after with UNIAMO, the Italian Federation of Rare Diseases, to contribute to addressing all the transversal needs. 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.