EXECUTIVE SUMMARY

9th European Conference on Rare Diseases and Orphan Products

Rare Diseases 360°
Collaborative strategies to leave no one behind

10-12 May 2018 Vienna
Messe Wien Exhibition & Congress Center

Organised by:
Co-organised by:
With co-funding from:

#ECRDVienna
IN PARTNERSHIP WITH

ASSOCIATE PARTNERS

- BlackSwan Foundation
- Childhood Cancer International
- CORD (Canadian Organization for Rare Disorders)
- CORD (Chinese Organization for Rare Disorders)
- EAHP (European Association of Hospital Pharmacists)
- ECPC (European Cancer Patient Coalition)
- EORTC (European Organisation for Research and Treatment of Cancer)
- EPF (European Patients’ Forum)
- E-Rare
- GÖG (Gesundheit Österreich)
- IFSW (International Federation of Social Workers)
- IRDiRC (International Rare Diseases Research Consortium)
- ISPOR (International Society for Pharmacoeconomics and Outcomes Research)
- Medical University of Innsbruck
- NORD (National Organisation for Rare Disorders)
- PHARMIG (Association of the Austrian Pharmaceutical Industry)
- Rare Diseases International
- Russian Patients’ Union
- Social Platform
- SWAN Europe
- ZSI (Zentrum für Soziale Innovation)
WHAT IS THE EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS (ECRD)?

- The European Conference on Rare Diseases & Orphan Products is the unique platform/forum across all rare diseases, across all European countries, bringing together all stakeholders - patients’ representatives, academics, healthcare professionals, industry, payers, regulators and policy makers.
- It is a biennial event, providing the state-of-the-art of the rare disease environment, monitoring and benchmarking initiatives. It covers research, development of new treatments, health care, social care, information, public health and support at European, national and regional levels.
- It is synergistic with national and regional conferences, enhancing efforts of all stakeholders. There is no competition between them, but rather our efforts are complementary, fully respecting initiatives of all.

KEY FACTS AND FIGURES

850+ attendees
58 countries represented
Over 120 expert chairs, speakers and panellists
6 parallel themes over 30 sessions
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PARTICIPANTS

300 connections from 41 countries to live videostream
200+ posters
#ECRD2018 used 4680 times

OBJECTIVES

- Disseminate the most up-to-date information related to the rare disease environment to all relevant stakeholders
- Demonstrate the importance of EU actions in the field of rare diseases and review progress made to date
- Elaborate strategies and mechanisms for developing further exchange of information between stakeholders
- Exchange knowledge and best practices on all relevant issues related to the rare disease environment

KEY POINTS OF SUCCESS

- The contribution of the entire rare disease community (patients’ representatives, academics, healthcare professionals, industry, payers, regulators and policy makers) representing all rare diseases, across borders
- The conference’s comprehensive range of topic areas and the viewpoint of rare disease patients as equal experts in the areas of research, diagnosis, product pathways to patients, the digital patient, quality of life, sustainability and economical perspectives in rare diseases and international policy
- Knowledge sharing of experience from different countries including Argentina, Austria, Belgium, Canada, Colombia, Croatia, France, Germany, Greece, Japan, Ireland, Italy, Lithuania, Netherlands, Norway, Saudi Arabia, Spain, Sweden, Switzerland, UK and USA
- The identification of the main priorities for future rare disease policy by participants on-site and those connecting remotely by live vote. The clear winner was patient-reported outcomes and real world evidence, followed by the systematic collection of findings from diagnostics and care
- Next steps will include the adoption of a European Joint Programme Co-Fund for Rare Diseases in 2019 to help structure rare disease research, and the possible adoption of the current legislative proposal for European cooperation on health technology assessment, a policy that will positively impact the delivery of rare disease treatments
- EURORDIS calls for new EU rare disease policy framework at ECRD 2018 Vienna: Specifically, EURORDIS is calling for a strong drive from the EU institutions and in particular, a reaffirmed leadership from the European Commission in the area of rare diseases to find solutions at a supranational level to help improve equity of access to health and to achieve universal health coverage and the health-related UN Sustainable Development Goals

Follow all of EURORDIS’ activities throughout the year, by signing up to our monthly newsletter available in English, French, German, Spanish, Italian, Portuguese and Russian: eurordis.org/enews
COMMITTEES

PROGRAMME COMMITTEE CO-CHAIRS

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WHAT ATTENDEES SAID:

“Really strong programme and lots of networking opportunities.”

“The vast range of themes was highly appreciated and having the social issue integrated among the themes, that usually meet the interest of the broader audience, was an added value. To be repeated.”
KEY TAKEAWAYS FROM THE POLICY OPENING SESSION

- On the opening day of ECRD, Terkel Andersen (1), President of EURORDIS-Rare Diseases Europe, called for a new EU policy framework and strong drive from the EU institutions in the area of rare diseases.

- Magdalena Daccord (2), Associate Director, Head of Operations EMEA – DIA called for patients to be at the epicenter of the drug development life cycle.

- Ana Rath (3), Director, Orphanet, called for us to address the local and global level at the same time and set out that we need a place at the table at the WHO, UN and EU.

- Rainer Riedl (4), President of Pro Rare Austria, the national rare disease alliance in Austria and host of ECRD 2018, spoke of the importance of patients’ dedication in achieving what is needed, and cited the need for networking at all levels with all stakeholders.

- Magdalena Arrouas (5), Acting Director for Public Health and Medical Affairs in the Austrian Ministry of Health, announced that considering the increasing awareness of the added value of collaboration in different fields like horizon scanning, HTA reports and joint negotiations, a topic of high priority on the Agenda of the Austrian Presidency of the Council will be the future European cooperation on HTA.

- Vytenis Andriukaitis (6), European Commissioner for Health and Food Safety, cited the success of the European Reference Networks as a prime example of how common efforts can lead to EU success stories, while Martin Seychell (7), Deputy Director-General, Directorate-General for Health and Food Safety, European Commission, confirmed that the EU Health Programme will be incorporated into the EU Social Fund Plus, and that the EU sees health as a fundamental cornerstone of social priorities.

- Maria Prähofer (8), gave an inspiring account of her experience of living with MPS IVA, Morbus Morquio, and called for patients to get involved in research and explained how a home treatment has helped to improve her quality of life.

- On the international front, Dr Rüdiger Krech (9), Director, Universal Health Coverage and Health Systems Office of the Assistant Director-General, World Health Organization, strongly suggested we go global to pool resources, while Daniela Bas (10), Director, Division for Social Policy and Development Department of Economic and Social Affairs, United Nations, called for an integrated social approach, beyond only a medical approach, due to the often disabling character of rare diseases.

Videos of opening and plenary sessions, speaker presentations and posters are available via: rare-diseases.eu

WHAT ATTENDEES SAID:

“Congratulations on providing an extremely interesting and comprehensive programme. I wish we had brought more delegates to the meeting.”

“Interactive sessions, such as the genome editing workshop, worked really well and stayed in my mind after.”
Theme 1 explored how we can exploit current achievements in genomics, how to prepare for new developments on the horizon and how to ensure no patients are left behind in order to shape the research and diagnostic landscape.

Participants examined the potential challenges associated with new technology enabling self-diagnosis, and considered how we support those patients for whom, despite all these innovations, their condition is likely to remain undiagnosed.

Sessions considered recent, innovative schemes of co-design and funding and how to carry out research that profits all stakeholders as well as how to attract investment. Ethicists, researchers and patients were invited to scrutinise the impact of recent developments in genome editing and discuss the skills, experience and tools required to enable patients to truly be considered and respected as equal partners in research and development.

The main conclusions that came through in this Theme suggest that:

- More efforts should be invested in, not only the training and empowerment of patients as experts, but also in promoting awareness and training of healthcare professionals to consider patients as equal partners and respect the fact that they possess a high level of knowledge.

- The landscape is rapidly changing and this is providing new opportunities for research, for diagnosis, for treatment, for funding and for many ways of building new partnerships.

- Collaboration between stakeholders is key. For effective collaboration, stakeholders need to be very clear about the frameworks within which they are working as well as what their goals, expectations and motivations are.

- Because of all the new opportunities in the research and diagnosis landscape, there are also many new risks, challenges and questions that remain unanswered. It is therefore essential to maintain an open dialogue and keep the patient at the centre of these efforts.
Theme 2 addressed how regulators, health technology assessment (HTA) bodies, and patients are asked to work together to prepare the arrival of breakthrough medicinal products. Sessions explored new tools and initiatives on both the regulatory and HTA side, trying to highlight where synergies are effective or still needed and how patients can improve the quality of their involvement, all along the technologies’ lifecycle.

The main conclusions from this theme were:

- With regard to orphan medicinal products’ pipeline development, the trends continue at the same pace: 1,986 orphan drug designations as of April 2018, translating into 160 authorised indications (Violeta Stoyanova, COMP member, Netherlands). Interestingly, the Gianno Benzi Foundation analysed how many of the designated orphan medicinal products were abandoned after their designation: of the first 788 designated products, 185 or 23.5% were abandoned - either due to lack of a clinical trial, or to negative results leading to a premature interruption.

- The role of the Committee for Orphan Medicinal Products at the EMA to indicate whether a new orphan product represents a significant benefit over existing treatments is increasingly important. The experience of the COMP and some guidance on the concept of significant benefit were published in 2017.

- Participants discussed whether horizon scanning can help detect which of the many products in development are more likely to come to the market with a high potential to affect public health. Kristina Larsson (EMA) described how the EMA examines the horizon: Systematically; 3-10 years before marketing authorisation application (MAA), and periodically; 3-20 years before MAA.

- Repurposed drugs are also among the potential candidates for orphan drugs. In the absence of more systematic collection of real-world and/or off-label use of clinical data, and the lack of a suitable economic model of incentives, it is rather difficult to apply drug repurposing as a treatment option in rare diseases (Diego Ardigo, Chiesi Pharmaceuticals).

Horizon: Systematically; 3-10 years before marketing authorisation application (MAA), and periodically; 3-20 years before MAA.
Looking at the PRIME programme and its experience at two years (Zahra Hanaizi, EMA): 177 requests received, 36 PRIME products granted, of which 30 in rare diseases (84%) - three products that benefited from the PRIME programme are being currently evaluated for a marketing authorisation. The feedback from industry (Steven Hall Pfizer) is positive: EMA’s recognition of the product when collaborating or seeking additional investment for further development / clinical studies is important.

Patient involvement it is key throughout the life-cycle of health technologies, and a mentoring programme can certainly help. In 2017, patients were involved in 925 activities at the European Medicines Agency, which held its first public hearing in September 2017. The CHMP will now invite more systematically patients to their oral explanation with the applicants (Nathalie Bere, EMA). Rob Camp (EURORDIS) explained the new EUROCAT programme for Community Advisory Boards, which provides a platform for interactions between developers of health technologies and patients’ representatives.

For the assessment of pharmaceuticals, Anne Willemsen (EUnetHTA) was pleased to confirm the EUnetHTA JA3 reports are now produced in parallel with the EMA evaluation and the HTA reports on pharmaceuticals were published two months after the European Marketing Authorisation.

The patient community is supporting the proposal for a long-term European Cooperation on HTA as proposed by the European Commission (Flora Giorgio, DG Sante), and calls the European Parliament and Council for the adoption of the proposed Regulation.
THEME 3
THE DIGITAL PATIENT

THEME LEADERS:
Julian Isla, Data and Artificial Intelligence Resource Manager, Microsoft and Dravet Syndrome European Federation (DSEF), Spain
Justina Januševičienė, Executive for the development of health care technologies and innovations, Lithuanian University of Health Sciences, Lithuania & Former Director, Healthcare resources and innovation management department, Ministry of Health, Lithuania

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Virginie Hivert, Therapeutic Development Director, EURORDIS-Rare Diseases Europe

While other industries are fully immersed in the digital era, the health industry is struggling to undergo a real digital transformation. The obstacles to create digital assets and relationships in the field of health range from unbalanced physician-patient relationships to clinical institutions focused on transactions and non-continuous care.

Theme 3 explored: how technology can help patients with rare diseases; how the regulatory world is evolving; current initiatives in Europe that aim to embrace this digital transformation; and real examples from patient organisations already starting this journey. We saw how some patient organisations are using a conversational interface to capture information, how new technology like artificial intelligence is being used for diagnosis, how the different European Reference Networks (ERNs) are connecting information, and how all this work has to be done for a real use under good processes and development.

Resounding messages that came out of discussions in from Theme 3 sessions were:

• New technology will create exciting opportunities but also new risks, as information will be more accessible to hackers and medical systems will be more exposed to cyberattacks. Accurate information and awareness are elements crucial to understand in order to mitigate the risks while we are evolving into a new era of medicine.

• Sharing data is needed to generate knowledge and for precision medicine. Data donation came through as an important topic in different discussions. We need to define new ways to share data and ensure patient data ownership. Data can be shared very easily. This is a big advantage but also a big risk. However, there is broad consensus among rare disease patients that the benefits of data sharing are always higher than the risks. ERNs offer an opportunity to securely connect and share health data to achieve better outcomes for patients.

• The GDPR is a real opportunity for patients to take back ownership of their data. The GDPR is not only a technical solution or a regulation, it is a process. There are new roles such as data managers that all institutions and all companies have to implement to be compliant with the GDPR. Patients will have more control on their own data and will be able to correct it and even delete it in some cases.
Rare diseases pose serious health, social and everyday challenges, which are often highly debilitating, and significantly affect the autonomy and the fundamental human rights of people living with a rare disease and their carers.

However, people living with a rare disease and their carers should be recognised and esteemed as people, not as a diagnosis. They should have the possibility to live a life with fulfilling personal relationships, to be able to contribute meaningfully to the lives of others and to society. Freedom to decide on their own lives, autonomy, security and dignity are important factors of what we call “quality of life”.

This theme revisited the concept of quality of life and explored the ways in which it can contribute to decision making and to shaping the provision of treatments and care.

• What really matters when talking about Quality of Life? Happiness, dignity, normality, autonomy, inclusion, health were the main words suggested by the audience.

• The societal aspects of Quality of Life have to be taken more into account, considering long term Quality of Life and considering potential side effects of treatments and care. Meaningful patient involvement in Health Technology Assessment does make a crucial difference;

• On the one hand, information about the impact of treatments and other interventions on quality of life is not always available. On the other hand, institutionally, interest in the use of quality of life evidence, such as
patient reported outcomes, is increasing to measure real life impact.

• When measuring quality of life, it is often best to use mixed methods and the content should be informed by patients and carers, ensuring that what is being measured relates to the outcomes that they wish to attain. For this reason, in some Quality of Life topics, it is necessary to develop a new survey instrument which will be more helpful than a validated Quality of Life questionnaire;

• If standardised Quality of Life measurement instruments are used, they should be tested first to ensure that they fit the question and the community being investigated. This is not a given, especially with rare diseases.

• A great number of people living with a rare disease are living with disability, facing several limitations in Activities of Daily Living. These disabilities are not always visible and may vary between acute and stable periods. A 360° and integrated human rights, health and social approach is crucial to give visibility to the disabilities faced by people living with a rare disease and to break down existing barriers, ensuring that they access the services and support that they need;

• Achieving integrated care for rare diseases requires collaboration, communication, knowledge, case managements and maybe a miracle – as suggested by participants;

• There are many good examples of delivering quality of life from across Europe on how to integrate and support patients’ cross-care needs, but also social needs, access to social services, navigating the many bureaucracies that people have to contend with. There does not seem to be one best way, but overall, the time that is invested in reducing patients’ and carers’ burden of organising care and treatment, finding information, explaining their disease to social services and so on, seems to be very well invested time indeed.
Theme 5 looked at economical aspects in rare diseases from different stakeholder perspectives, evaluated existing collaborative approaches and discussed options to further develop an environment conducive to innovation and to faster access for patients to care and cures.

The sessions in this theme explored ambitions to refine a shared understanding on how to improve access to rare disease therapies and how to ensure a sustainable medicines development business model for all stakeholders involved.

Participants shared findings on the economic and financial impact of rare diseases on healthcare systems and societies, including testimonials and case studies from patients, as well as the example of the potential that European Reference Networks (ERNs) have to improve the understanding of challenges and provisions of solutions.

There was a very positive attitude and a ‘can-do’ mind-set that ran through all of these workshops, and the different themes that were addressed ranged from the burden of disease to its measurement, to how much do we know about the cost of the illness, the impact on families, what kind of evidence gap exists and what can be done.

The theme looked both into the impact of the current policies on access to rare disease therapies as well as into innovative concepts and collaborative approaches which are being piloted throughout Europe.

- Alternative frameworks as to what constitutes value of an orphan medicine that can justify prices are currently being discussed in the academic field. The possibility of a paradigm-shift from cost per patient to focusing on the role of budget impact adding societal perspectives is not yet mature enough, but will certainly be important in the years to come.

- One general observation was that we should be careful about new paradigms in the field of rare diseases development and trying to change everything. What has happened so far has not been a failure. On the contrary, great progress has been made. The challenge is to find complimentary and additional approaches, not to replace the existing ones.

- The future lies in collaboration, and there is a huge opportunity and potential that has been untapped in the collaboration between the field of economics on the one hand, and the rare disease community on the other. There is a lot to be done; there is a lot that can be done. Economics that is well understood and well implemented can be a friend of the rare disease community.
THEME LEADERS:
Durhane Wong-Rieger, President & CEO Canadian Organization for Rare Disorders, Chair of Rare Diseases International, Founder of the Asia Pacific RD Alliance (APARDO), Canada
Professor Hugh Dawkins, Director, Office of Population Health Genomics, Health Department of Western Australia, Australia

EURORDIS SUPPORT:
Paloma Tejada, Director, Rare Diseases International, EURORDIS-Rare Diseases Europe
Clara Hervas, Public Affairs Junior Manager, EURORDIS-Rare Diseases Europe

At the heart of theme 6 was the objective of having a world where all people living with a rare disease receive equitable treatment and support, and all advances in rare disease research, policies and services benefit all those affected, regardless of location. This was discussed from both top-down and bottom-up perspectives, and in all areas, from policy and research to products and practical solutions. Participants examined the ways in which they can leverage global policies and agencies to explicitly support rare diseases, whilst also recognising diversity and ensuring equality so that all persons living with a rare disease have access to the best possible care in their local environment. There was a focus not only on research and innovations in areas such as diagnosis, but also on the real-world application of this and the impact on patients and their families.

The main conclusions of this theme were:
• **We are certainly moving towards global equity** even if we have not yet reached it.
• Rare Diseases International and the NGO Committee for Rare Diseases will be essential for driving forward international collaboration and leveraging global entities to achieve recognition of rare diseases as a global health priority and public health issue.
• Regional advances in Latin America, Asia, Middle East and Australia will feed into global policy.

• **Eco-systems of knowledge must be created**, from both research-orientated, scientific databases and registries, to ones that are being used in clinical care.
• To achieve our goals globally it is **essential to invest regionally** and we have already seen the success of this in the locally designed rare disease centres of excellence.
• **Individuals and their power to inspire must remain at the core of what we do**, coming together over the huge commonalities of rare diseases.