

#ECRDVienna

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National Center
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Translational Sciences



OFFICE OF ORPHAN PRODUCTS DEVELOPMENT



EUROPEAN SOCIETY OF HUMAN GENETICS

pro rare austria
allianz für seltene erkrankungen



European Hospital and
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▶ Rare Diseases
International



▶ CORD (Chinese
Organization for Rare
Disorders)



▶ IRDiRC (International
Rare Diseases
Research Consortium)



▶ Russian Patients'
Union



▶ EAHP (European
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▶ ISPOR (International
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▶ ECPC (European
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▶ SWAN Europe



▶ EORTC (European
Organisation for
Research and
Treatment of Cancer)



▶ Medical University of
Innsbruck



▶ ZSI (Zentrum für
Soziale Innovation)

KEY FACTS AND FIGURES

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.



PARTICIPANTS

850+ attendees	300 connections from
58 countries represented	41 countries to live videostream
Over 120 expert chairs, speakers and panellists	200+ posters
6 parallel themes over 30 sessions	#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 to **improve equity of access to health** and to **achieve universal health coverage and the health-related UN Sustainable Development Goals**

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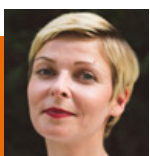
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RARE DISEASES 360°

COLLABORATIVE STRATEGIES TO LEAVE NO ONE BEHIND

THE EUROPEAN CONFERENCE ON RARE DISEASES AND ORPHAN PRODUCTS 2018 VIENNA

Organised by EURORDIS-Rare Diseases Europe, and co-organised by Orphanet and DIA, the 9th edition of the European Conference on Rare Diseases & Orphan Products took place in Vienna from 10 – 12 May 2018.

ECRD 2018 Vienna brought together **over 850 participants from 58 countries around the world** (more than ever before!), representing various stakeholder groups including **300+ patients** and **100+ healthcare industry professionals**, as well as payers, regulators, policy makers, researchers, healthcare professionals and academics.

Over **300 people connected from 41 countries** during the live online transmission of the plenary and opening sessions, while over 200 posters were displayed and 120 speakers gave high-quality talks over the two-day event.



The overarching theme for ECRD 2018 “**Rare Diseases 360° – collaborative strategies to leave no one behind**” reinforces the unique quality of this event for the rare disease community in Europe, which brings together and facilitates effective policy discussions between all rare disease stakeholders. It also encompasses the comprehensive range of topics covered at the conference and the viewpoint of **rare disease patients as equal experts, representing all rare diseases, across borders.**

THE CONFERENCE FOCUSED ON 6 THEMES:

THEME 1

STRUCTURING THE RESEARCH
& DIAGNOSIS LANDSCAPE

THEME 2

BREAKTHROUGH MEDICINES ON THE HORIZON:
REGULATORS, HEALTH TECHNOLOGY ASSESSORS (HTA)
AND PATIENTS WORKING TOGETHER

THEME 3

THE DIGITAL PATIENT

THEME 4

QUALITY OF LIFE:
MAKING WHAT MATTERS, MATTER

THEME 5

ECONOMICAL PERSPECTIVES IN RARE DISEASES

THEME 6

GLOBAL RARE EQUITY:
ARE WE THERE YET?



SETTING THE CONTEXT AT ECRD 2018 VIENNA

The rare disease community, including all rare disease patient organisations and their partners, can be deservedly proud of its achievements over the last 20 years. But as we look forward to the next decades there is still much to achieve. Advancements in science and medicine - such as transformative treatments, game-changing digital technologies, innovation in the organisation of health and social care - and increased empowerment of patients, will provide many opportunities.

We need to accelerate the momentum we have built over the last 20 years to ensure no single person living with a rare disease is left behind. ECRD Vienna 2018 was the opportunity to discuss and reach solutions on how we can look to the future to improve the lives of the estimated 30 million people living with a rare disease in Europe and 300 million worldwide.

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.”

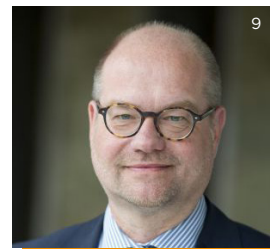
KEY TAKEAWAYS

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 **EMA – 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 SUMMARIES

THEME 1

STRUCTURING THE RESEARCH & DIAGNOSIS LANDSCAPE

THEME LEADERS:

Daria Julkowska, Scientific Coordinator,
E-Rare, France

Lauren Roberts, Director of Support, Genetic
Alliance UK, National Coordinator, Swan UK, UK

EURORDIS SUPPORT:

Virginie Bros-Facer, Scientific Director,
EURORDIS-Rare Diseases Europe

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 SUMMARIES

THEME 2

BREAKTHROUGH MEDICINES ON THE HORIZON: REGULATORS, HEALTH TECHNOLOGY ASSESSOR (HTA) EXPERTS AND PATIENTS WORKING TOGETHER

THEME LEADERS:

Wim Goettsch, Executive Board Chair,
EUNetHTA, Netherlands

Jordi Linares Garcia, Head of Scientific and
Regulatory Management, EMA, EU

EURORDIS SUPPORT:

François Houyez, Treatment Information and Access
Director, Health Policy Advisor, EURORDIS-Rare
Diseases Europe

Matteo Scarabelli, Patient Engagement Manager –
HTA, EURORDIS-Rare Diseases Europe

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.

Participants were introduced to: the concept of the Community Advisory Boards (enabling a patient-driven involvement with clinical trial sponsors); the PRIME initiative; an overview of the orphan drug experience; and concepts such as drug repurposing. On the subject of HTA, participants discussed activities of EUNetHTA and also the European Commission's proposal for future European Cooperation on HTA.

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 Giannio 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)

- 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 EUROCAB 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 SUMMARIES

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

EURORDIS SUPPORT:

Elisa Ferrer, Patient Engagement Manager, EURORDIS-Rare Diseases Europe

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.

THEME SUMMARIES

THEME 4

QUALITY OF LIFE: MAKING WHAT MATTERS, MATTER

THEME LEADERS:

Lene Jensen, Chief Executive Officer, Rare Diseases Denmark, Denmark

Ursula Holtgrewe, Head of Work & Equal Opportunities, Zentrum für Soziale Innovation, Austria

EURORDIS SUPPORT:

Raquel Castro, Social Policy Senior Manager, EURORDIS-Rare Diseases Europe

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 SUMMARIES

THEME 5

ECONOMICAL PERSPECTIVES IN RARE DISEASES

THEME LEADERS:

Michael Schlander, Professor of Health Economics, University of Heidelberg, Germany

Ruediger Gatermann, Director, Healthcare Policy and External Affairs Europe, CSL Behring, Germany

EURORDIS SUPPORT:

Simone Boselli, Public Affairs Director, EURORDIS-Rare Diseases Europe

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.



- When it comes to the topic of what can be done and what needs to be done in the field, one aspect repeatedly discussed: **the potential of European Reference Networks to generate valuable data and insights**, and to contribute to the further evolution of the field to the benefit of the patients and those who care for them.
- 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 SUMMARIES

THEME 6

GLOBAL RARE EQUITY: ARE WE THERE YET?

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.





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