EXECUTIVE SUMMARY

THE 10th EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS

ERDI

ONLINE on 14-15 May 2020

THE JOURNEY OF LIVING WITH a RARE DISEASE in 2030

Organised by EURORDIS RARE DISEASES EUROPE

Co-organised by orphanet Inserm

With the support of AFMTELETHON INNOVER POUR GUERIR

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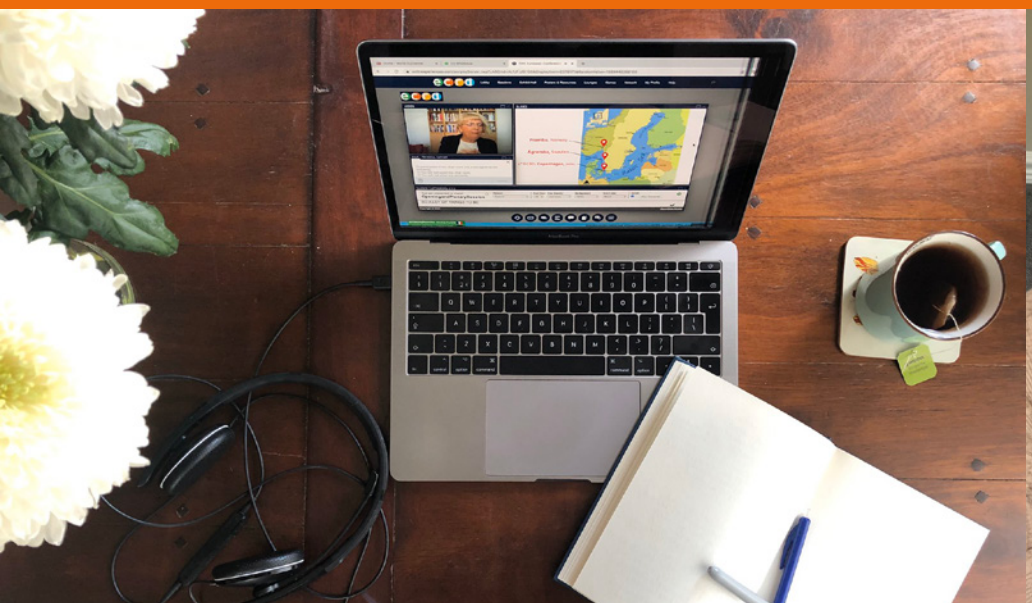

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WHAT ATTENDEES SAID:

"I think EURORDIS did a spectacular job of putting this together in such a short amount time. By the way, **having access to the presentations and discussions for a year** is really extremely helpful!"

KEY FACTS AND FIGURES

WHAT IS THE EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS (ECRD)?

The ECRD is recognised globally as the **largest, patient-led rare disease event** in which collaborative dialogue, learning and conversation takes place, forming the groundwork to shape future rare disease policies.

57 countries represented

1500+ online participants

120+ experts as session chairs, speakers & panelists

230+ posters displayed

3 European Commissioners

1042 unique visitors during opening session

15 exhibitors showcased

2 Ministers of Health

ECRD2020 used 3100 times on Twitter

CONTEXT

ECRD 2020, a ground-breaking virtual event

For the first time ever, the European Conference on Rare Diseases & Orphan Products 2020 was a fully virtual event.

An unprecedented success, 1,500+ people from 57 countries registered to attend ECRD online.

At a moment in history when people have experienced strong feelings of isolation, the event offered a timely opportunity for conference participants to come together and connect online with members of the rare disease community around the world.

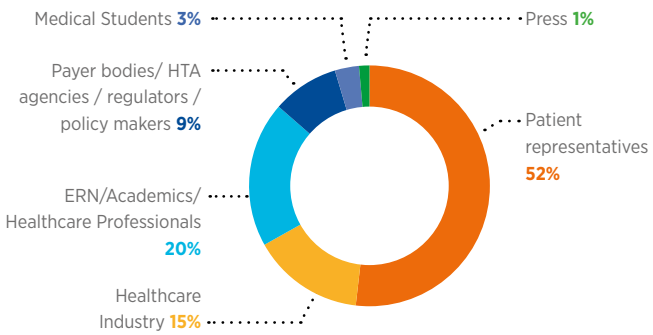
Based on the work carried out through the ongoing Rare2030 Foresight Study, ECRD brought together patient advocates, industry representatives, regulators and healthcare professionals to discuss how to build a better future for people living with a rare disease.

Through a custom-built platform, participants were able to attend from the safety of their own homes and participate in 29 sessions with 100+ speakers, chat with fellow attendees and participate in 'meet the speaker' sessions.

A huge thank you to ECRD attendees for joining, contributing to such high quality and productive discussions, and for sharing positive feedback of their ECRD 2020 online experience.

Leading, inspiring and engaging **all stakeholders** to take action, this Conference is an unrivalled opportunity to network and exchange invaluable knowledge with all stakeholders in the rare disease community.

Here is a breakdown of ECRD 2020's online participants:



All sessions available on demand

For anyone that was unable to attend the event, it is still possible to register for full access to all recordings via: www.rare-diseases.eu/register/. Learn about the policy options that can lead to better conditions for people living with rare diseases for the years ahead.

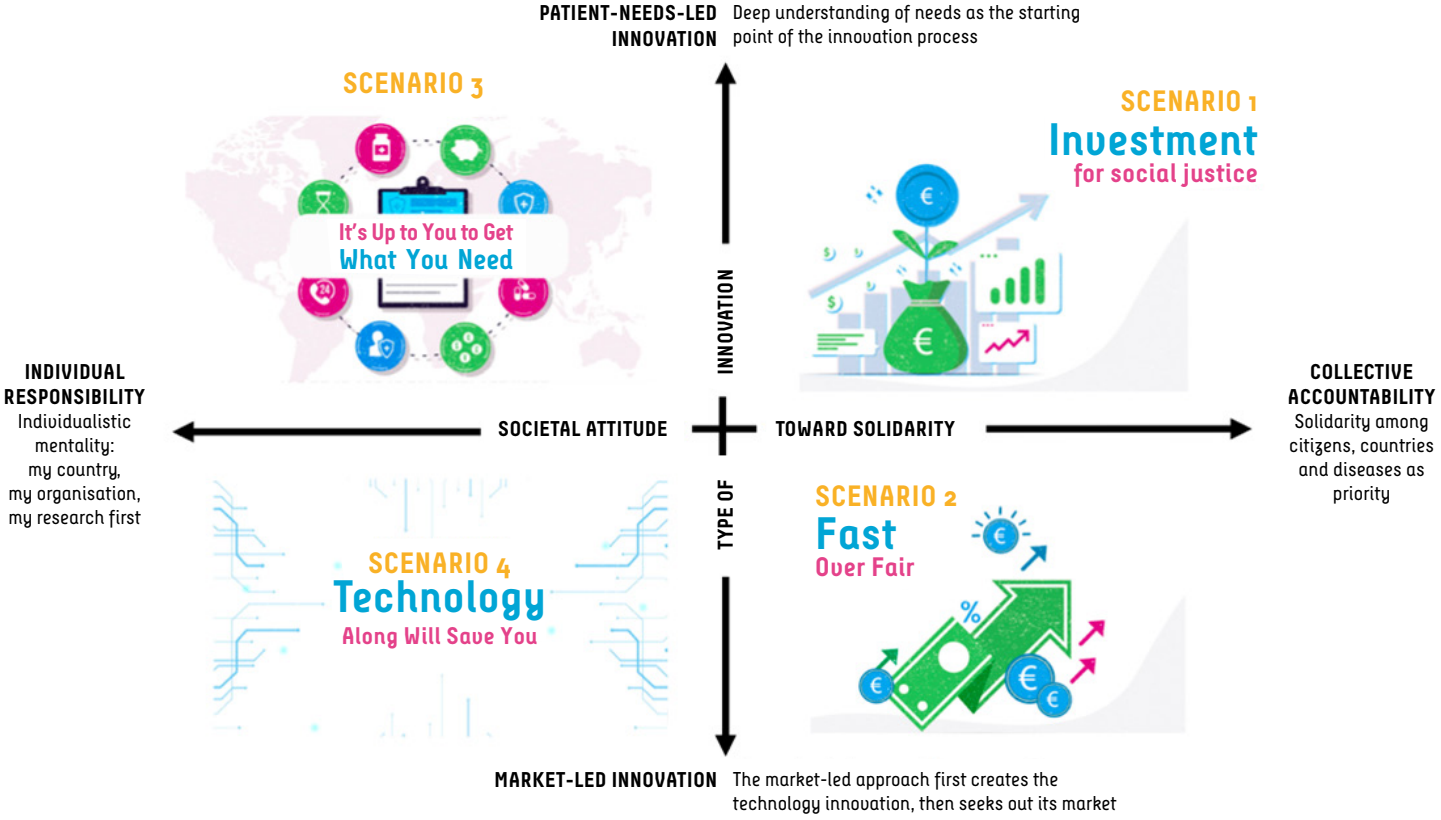
Follow all of EURORDIS' activities throughout the year, by signing up to our monthly newsletter available in English, French, German, Spanish, Italian, Portuguese and Russian! eurordis.org/enews

ECRD AND THE RARE2030 FORESIGHT STUDY

Since 2019 the partners of the Rare2030 project and its over 200-member consultative panels of experts, young citizens and patient advocates have embarked on a foresight study to prepare recommendations for a new policy framework for people living with rare diseases by 2030 and beyond. With the back drop of the COVID-19 pandemic, such uncertain times require - more than ever - the need to prepare for the future by identifying possible scenarios and co-designing the policies needed to reach those that are most preferable.

Rare2030
Foresight in Rare Disease Policy

The 10th European Conference on Rare Diseases & Orphan Products 2020 marked a unique online opportunity for the rare disease community to consider what the journey of people living with rare diseases will look like in four possible future scenarios that differed in their emphasis on solidarity and drivers of innovation.



An audience of over 800 delegates in the Rare2030 opening plenary session voted on the four Rare2030 scenarios. Discussions ensued throughout the two-day conference, indicating the following opinions:

- If we continue as we are we will find ourselves in the "Fast over Fair" scenario which forecasts high collective responsibility but with an emphasis on market-led innovation
- The majority of the rare disease community prefers a future scenario with continued high collective accountability but more of an emphasis on needs-led innovation, "Investments for Social Justice"
- A significant portion of the community agreed that a balance must remain with the market led attractiveness of the "Technology Along Will Save You" scenario.
- A scenario where "It's Up to You to Get What You Need" was least preferred by all.

RARE 2030 SCENARIOS VIDEO
https://youtu.be/WBvsj9E0U-g

These scenarios and Rare2030 foundations served as an instrument to help the audience consider how to prepare for the best possible journey in 2030.

WHAT ATTENDEES SAID:

"Such an achievement.
Can't say how **fantastic** it is to be here.
I couldn't have joined if it wasn't online"

KEYNOTE SPEECHES

View **full programme**

Click on the icons to watch the replay on YouTube

HRH PRINCESS VICTORIA
HRH Crown Princess Victoria of Sweden

Challenges like navigating through the healthcare system, finding the right doctors and treatments, and struggling with emotional and sometimes even financial stress that takes energy, strength, and resilience. That is why organisations such as yours, and meetings such as this, are so important: to give voice to those who need to be heard, to share knowledge and experience, and to remind us that, even when the world gives you plenty of reasons to feel that way, you are not alone.

TERKEL ANDERSEN
President, EURORDIS-Rare Diseases Europe

This epidemic has shown that if you fail to prepare, you prepare for failure, and we must prepare together across borders.

ANA RATH
Director, Orphanet

By looking for new ways to imagine science and care for the 300 million people living with a rare disease in the world today, the rare disease field is, despite its intrinsic diversity, a fantastic land of innovation and solidarity. This is a way of saying that we as a community are, to a degree, well-prepared for the new world after COVID-19.

MARIA MONTEFUSCO
President, Rare Diseases Sweden

For us in the disability and patient movement, the focus [during the COVID-19 pandemic] changed just overnight from pushing for policies for inclusion in society in general, to the right to life...we try to remind decision makers all the time that life and health are fundamental and at the same time the whole life of a person matters.

LENA HALLENGREN
Swedish Minister of Health & Social Affairs, Sweden

"It is crucial that we work together across national borders. By working together we can provide the best possible support to the patients with the rarest of diseases and make their journeys better."

"An effective, equal and accessible healthcare system is crucial for strong welfare society and especially important for those living with chronical diseases and other health problems."

KEYNOTE SPEECHES

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STELLA KYRIAKIDES
European Commissioner for Health and Food Safety

In the coming years we are carrying out a real stocktaking exercise, we will be guided by the Rare2030 Foresight Study. We need now to take lessons from COVID-19. We know that patients will be the driving force of our rare disease policy.

HELENA DALLI
European Commissioner for Equality

All people should be able to participate fully and equally in society and in the economy. It is not only their right, their participation also represents a huge contribution to the whole of society.

IRENE NORSTEDT
Director in DG Research & Innovation of the European Commission (Statement by **MARIYA GABRIEL**, Commissioner for Innovation, Research, Culture, Education & Youth)

Conferences such as this one provide an important forum to build the connections between researchers, innovators, clinicians, and people living with a rare disease across Europe and the world. We in the European Commission will keep supporting the scale of research collaborations needed in the area of rare diseases to deliver tangible impact to all those in need.

DAVID LEGA
Member of the European Parliament, Sweden

Living with a rare disease myself, looking back 100, 50 or only 10 years, I see that we have come a long way but we need to move society and its attitudes even further in the coming years...and the power to make a difference, really lies with you.

ANNA WEDELL
Member and Former Chair of the Nobel Committee for Physiology or Medicine, Sweden

We have tremendous opportunities thanks to new technology but we have to realise that it is not just technology itself. This is the result of many women and men over history, step by step, making these critical discoveries that really transform medicine.

GARETH BAYNAM
Clinical Geneticist, Genetic Services of Western Australia

The vision, the hope and the action is to combine the voice of the people, the voice of the doctor, Orthodox medical knowledge, ancient wisdom and new technology to deliver answers, community, trust and empowerment.

KEYNOTE SPEECHES

View **full programme**

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MILAN MACEK
Motol University Hospital and Charles University, Prague

The COVID-19 pandemic has taught us more than ever that we cannot predict the future but we can prepare for it. We have both the impetus of the EU Court of Auditors Report on the Implementation of the Cross Border Healthcare Directive and the upcoming opportunity created by the trio Presidency of the EU council by France, Czech Republic and Sweden in 2022 and 2023 which can provide a roadmap for a new policy framework for rare diseases.

REBECCA TVEDT SKARBERG
Osteogenesis Imperfecta Federation Europe (OIFE), Norway

Development is dependent on all of us! Doctors and researchers, social workers and policy makers, psychologists, teachers, parents and loved ones. It's about our society and what we want to see. The time is now. Do your part to steer towards a better world for rare disease.

JANA POPOVA
Young Patient Advocate, Bulgarian Association for Neuromuscular Diseases; European Patients' Forum Youth Group; European Alliance of Neuromuscular Disorders

The truth is that once you become a person living with a rare condition you have to face this hurricane of emotions and feelings that you have never experienced before. The good news is that there are many people who can help... and why for me the strongest message from this conference is that only if we continue to work together we will be able to provoke the positive change we want in the world and to provide a brighter future for the community of people living with rare diseases.

ADAM VOJTECH
Minister of Health, Czech Republic

At the EU-wide level our government is looking forward to the outcomes of this conference which will substantially inform the Rare2030 project!

YANN LE CAM
EURORDIS

We are preparing for the next decade. We are bringing forward solutions, created at the margins of society but becoming more mainstream. Rare2030 was the backbone of ECRD 2020. The outcome of the conference is a new impetus to initiate a new legislative framework for rare diseases.

WHAT ATTENDEES SAID:

"Kudos to the EURORDIS team for all the work you have done to take ECRD virtual amidst all of the other great work you are doing on behalf of the rare disease community and dealing with the pandemic!"

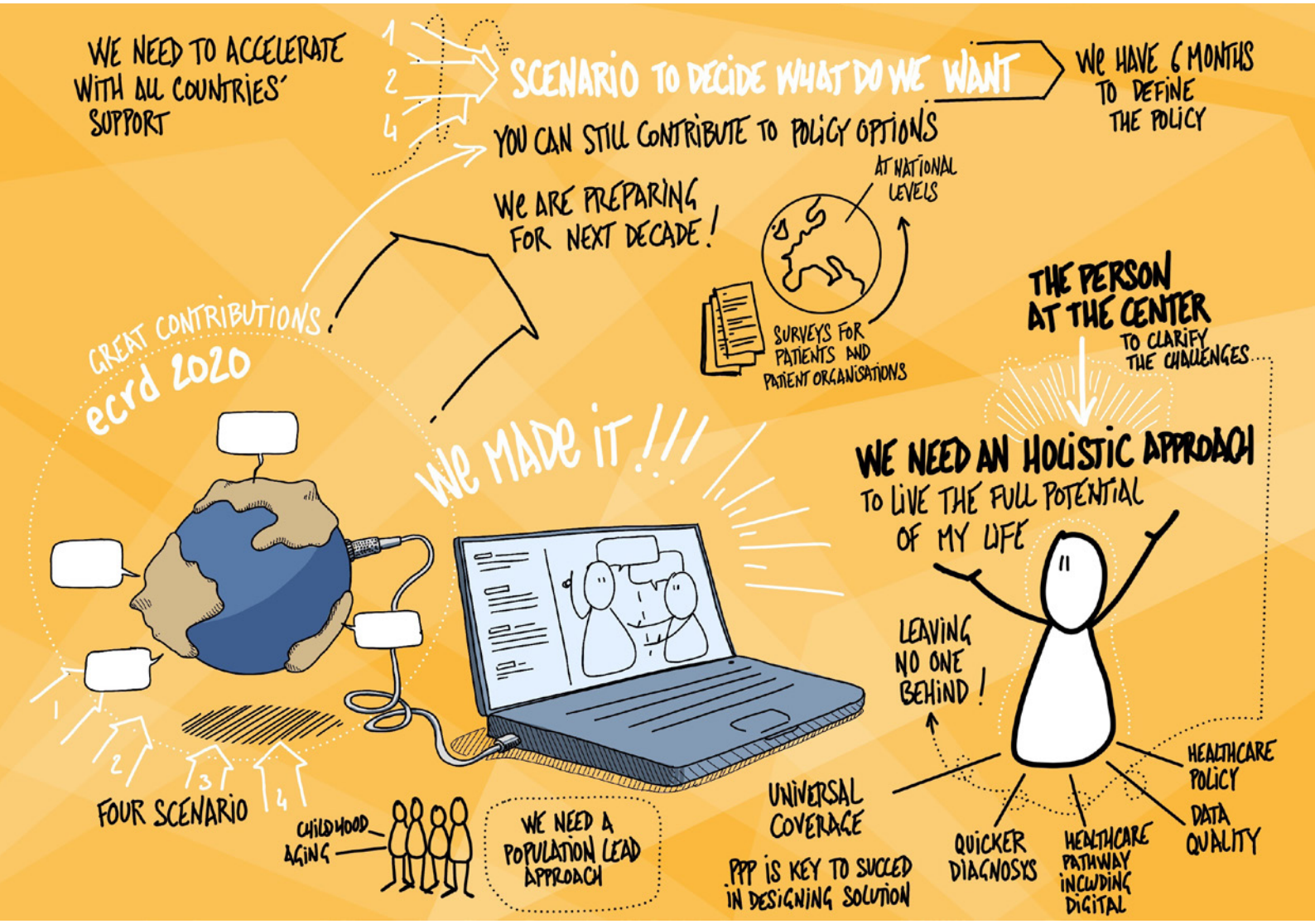
KEY TAKEAWAYS from EURORDIS CEO, Yann Le Cam

FUTURE POLICY OPTIONS FOR AN IMPROVED JOURNEY OF LIVING WITH A RARE DISEASE IN 2030

As the ECRD unfolded our community was on the frontline responding to the impact and challenges of the COVID-19 pandemic on people living with rare diseases and our healthcare and social care systems. We continue to prepare for the future by bringing forward innovative concepts and models created at the margins of the healthcare system but with benefits for all.

The ECRD conference marked a turning point in our vision for the future of people living with rare diseases in the future. The high-level collective goal is that every person living with a rare disease in Europe live the full potential of their life. This ambition to leave no one behind calls for a holistic approach that responds to a person's needs all along their lifetime and results in improved health and social outcomes. The United Nation's Sustainable Development Goals (already appropriated by the EU and WHO Europe) provides the rare disease community with an existing framework based on such a holistic approach as well as human rights and disability conventions.

Click to watch the replay



To achieve this ambitious goal, the following policy considerations are required:

An international vision – efforts at the local, regional, national and international levels are all connected and must remain concerted for success.

A tailored approach – we now know that although the rare disease community must continue to stay united to overcome its common challenges tailored approaches to care, research and treatment may be appropriate for some groups such as those living with cancers, very rare diseases, or rare genetic diseases versus rare infectious diseases for example.

Policies and technologies for accelerated diagnosis – The revision and update of public health policies across Europe such as on newborn screening and the clinical implementation of genome sequencing techniques will lead to earlier diagnosis and better health outcomes – especially for people with currently "undiagnosed" rare diseases. Awareness raising efforts amongst frontline healthcare professionals and greater harmonisation across the EU remain key.

Accelerated translation of technologies into available, accessible and affordable treatment - 20 years ago the first patient was treated for SCID with a curative treatment. Today uptake of new technologies remains too slow and the decision making processes need to be adapted in order to make the curative treatments available. More knowledge gathering for common assessment through an Health Technology Assessment (HTA) legislation and a greater critical mass and bargaining power of Member States into a European collaborative effort with an organised and continuous approach to evidence generation through the product lifecycle and structured negotiations are required.

Public private partnership – solutions must be co-designed by the public and private sector, including civil society and all private actors.

A person-centred approach – By considering the person at the core of all policies the lines between sectors disappear and the journey of a person living with a rare disease becomes smoother from diagnosis, to specialised care and primary care, to integration in society, to being not just empowered and engaged but an equal partner with all the other actors in creating treatments and solutions for a better quality of life.

Adequate policies to support the highest excellence in clinical care – Adequate healthcare pathways will require EU level support working in coordination with a bottom up effort from Member States and other countries in Europe. On the local level people living with rare diseases and their health care providers require additional support to navigate their regional and national health and social care systems and connect primary to specialised care and treatments that may exist across borders. To achieve this, improved implementation of the EU Cross Boarder Healthcare Directive is required. In addition, support for European Reference Networks as a sustainable ecosystem - well integrated with national healthcare systems - is required.

Funding that adequately meets ambitions – for clinical research, healthcare pathways from primary to specialised care, social research and the technology (including telemedicine and Artificial Intelligence applications) that links them all. This adequate funding is particularly important for the infrastructure of European Reference Networks.

Such policies need to be flexible, agile and open in order to adapt to rapidly changing scientific and societal landscapes. We don't want policy driven solely by the offer of public services like in the "Social Justice for all" Rare2030 scenario or exclusively by the market like in the "Fast Over Fair" or "Technology Alone Will Save You" scenarios, or by competencies of bodies of administrations limited by their boundaries such as those in the "It's Up to You to Get What You Need" scenario. We need to go beyond these boundaries. We need to be driven by the response to our community's needs, to the person's needs, with the right incentives, rewards, funding for all actors – all health technology industries, medical and social care professionals, policy makers, payers and patient organisations.

The ECRD has been a first opportunity to consider how to prepare for a better future for people living with rare diseases. Several opportunities in the Rare2030 Foresight Study to continually include the input of all stakeholders into these recommendations will continue throughout 2020:

- Deliberations of the 200+ members of expert panels
- Regional workshops taking place in 6 Member States with upcoming EU presidencies
- A workshop and survey with European Reference Health Care Professionals and EU healthcare policy makers
- A survey for people living with rare diseases and their advocates
- A Young Citizen Conference

These inputs will converge into recommendations presented at a final conference during Rare Disease Week 2021.

CONFERENCE THEMES

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THE FUTURE OF DIAGNOSIS:
NEW HOPES, PROMISES AND CHALLENGES

THEME 2

OUR VALUES, OUR RIGHTS, OUR FUTURE:
SHIFTING PARADIGMS TOWARDS INCLUSION

THEME 3

SHARE, CARE, CURE: TRANSFORMING
CARE FOR RARE DISEASES BY 2030

THEME 4

WHEN THERAPIES MEET THE NEEDS:
ENABLING A PATIENT-CENTRIC APPROACH
TO THERAPEUTIC DEVELOPMENT

THEME 5

ACHIEVING THE TRIPLE AS BY 2030: ACCESSIBLE,
AVAILABLE AND AFFORDABLE THERAPIES FOR
PEOPLE LIVING WITH A RARE DISEASE

THEME 6

THE DIGITAL HEALTH REVOLUTION:
HYPE VS. REALITY

WHAT ATTENDEES SAID:

"**Great conference**, rich in contributions and vision. Thank you all. **Exciting** to see such a wide and deep engagement on rare diseases. Lots of energy and purpose. Let us all maintain this momentum in Europe and beyond!"

THEME 1
THE FUTURE OF DIAGNOSIS: NEW HOPES, PROMISES AND CHALLENGES

THEME LEADERS:

Prof. Christine Patch, Clinical Lead for Genetic Counselling, Genomics England, UK

Virginie Bros-Facer, Scientific Director, EURORDIS

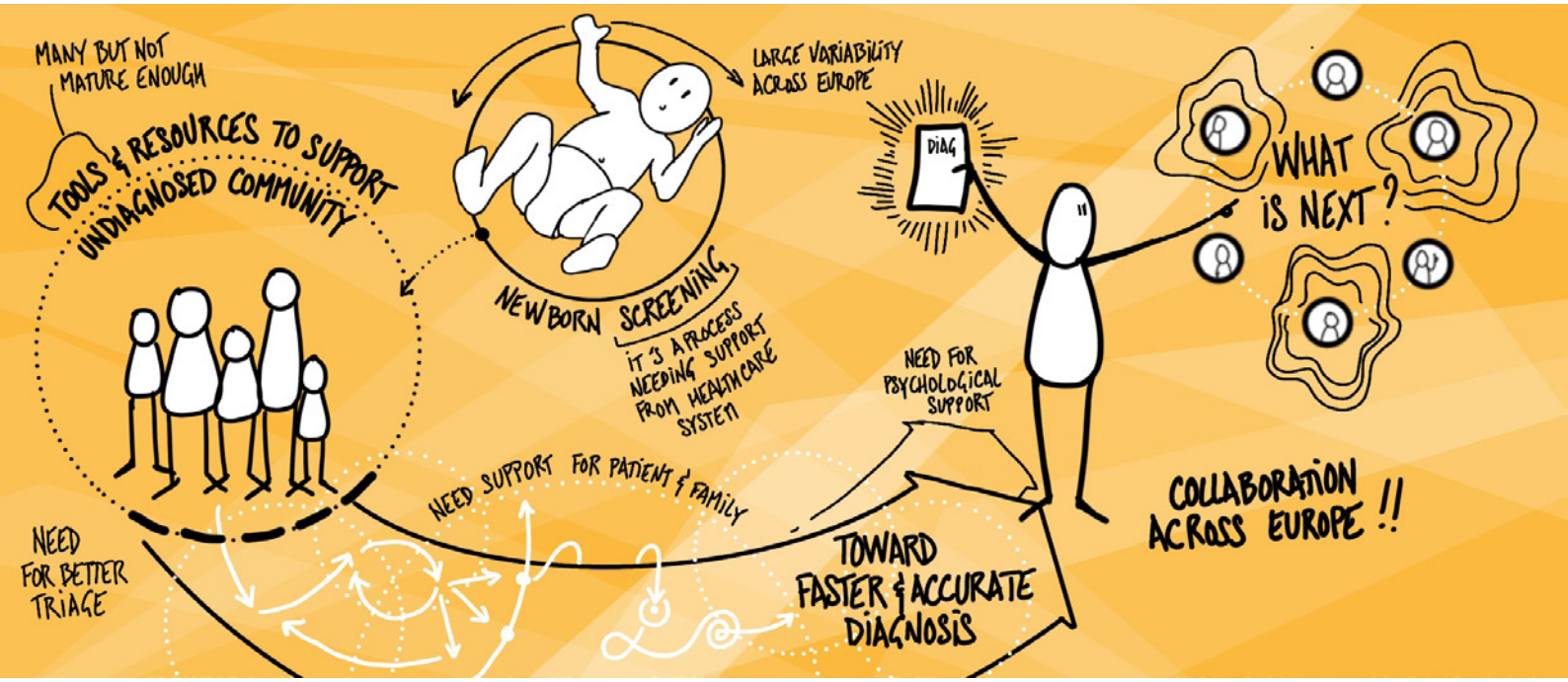
THEME SUPPORT :

Gulcin Gumus, Research and Policy Project Manager, EURORDIS

View **full programme**

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

- There is a need for better **triage**, better **screening tools** and better **classification of data**. Also, **equity of access** to genetic testing should be promoted. A balance between fostering creativity (technological tools/initiatives) and a coordinated and integrated response using the tools will speed up access to diagnosis.
- Newborn screening is not just a test but a complex and **multidisciplinary system** that needs adequate **information** and **communication** with families and the general public along with adequate **training for healthcare professionals**. Timely information and treatment are needed. Perspectives need to be attuned to harmonise newborn screening across Europe. **Scientific advancements** should be taken into account when discussing the expansion of the screening panel. The potential of **genome sequencing** for newborn screening should be further explored.
- Getting a diagnosis answers an important question but often triggers a thousand more: it is just the start of the next phase of the **patient journey**. Therefore, patients need better connections to information support communities and to research.
- Interventions to help improve **clinical management systems** and other initiatives such as **medical apps** using decision support systems will contribute to deciphering unsolved and unsolvable diseases.
- Future specialised health services need to a) **integrate liaison officers** for rare diseases to support the follow up care of newly diagnosed/undiagnosed rare disease patients and b) better **support expectation management** and information by setting up principles and recommendations. The novel care pathway approaches, knowledge created by the **European Reference Networks**, need to be reflected and implemented at national levels to support clinicians to better care for their patients.



THEME 2

OUR VALUES, OUR RIGHTS, OUR FUTURE: SHIFTING PARADIGMS TOWARDS INCLUSION

THEME LEADERS:

Maria Montefusco, President, Rare Diseases Sweden, Sweden

Valentina Bottarelli, Public Affairs Director and Head of European and International Affairs, EURORDIS

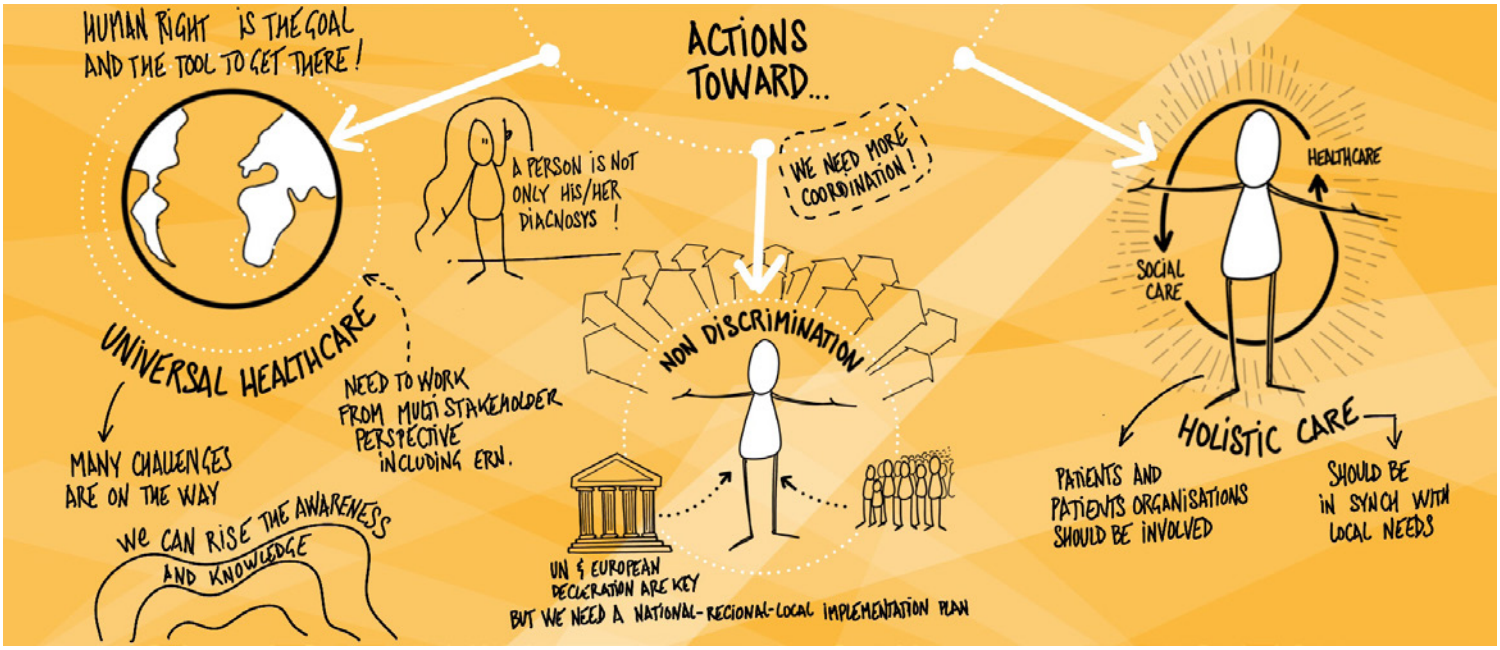
THEME SUPPORT :

Clara Hervas, Public Affairs Manager, European and International Affairs, EURORDIS

View **full programme**

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

- One of the **key paradigm** shifts needed is the one that will take us from looking at people living with a rare disease from the strictly medical point of view to looking at them as more than their diagnosis: as individuals that need **inclusion and participation in society** and the **respect of their human rights**, including the **rights to life, liberty, security of person, education, work, adequate standard of living** and the enjoyment of the **highest attainable standard of physical and mental health**. The fight for human rights in the rare disease community has long focused around access to care and medicines, from a resource perspective, but taking a **holistic, person-centred human rights approach** that strives for all rights can produce better outcomes.
- **Human rights** are a goal to be attained, but they are also a tool in themselves. It is essential to integrate the human rights approach at every level - from the **local, national, European to the international** – and to consolidate commitments into legal frameworks and agreements. One such important international agreement is the **UN Political Declaration on Universal Health Coverage** which acknowledges the right to health of all, and puts a spotlight on vulnerable populations such as people living with a rare disease. In order to make UHC a reality for them, it is key to operationalise it. In the EU context, this is achieved through legislative instruments like the **Orphan Drug Regulation and the Cross-Border Healthcare Directive**, as well as tools like the **European Reference Networks**, which need to all combine as a part of the same overall goal.
- One of the key barriers to inclusion and participation in society is the existence of silos that are not well connected and do not coordinate within the health and welfare system, as well as a lack of exchange with the patient. **Holistic care** offers an approach that addresses the **micro level** (the person receiving the service he/she needs via multidisciplinary care teams) and the **macro level** (the person being involved in the organisation, design and evaluation of the structures through patient representatives). There are currently **barriers to holistic care**, including the **rarity of expertise, the separation of budgets and portfolios between ministries** and the **fragility of national strategies** that are not planned for the long-term. However, if adapted to local needs and circumstances, holistic care offers the possibility to provide **better care delivery** and to **lower costs** to the care system.
- People living with a rare disease and disability often face **discrimination**, and it is important to understand disability from a human rights perspective and **break down barriers** embedded in the system, which includes **bad attitudes and stigma**. For instance, disability assessment should not be diagnosed-based but **needs-based** and include all external barriers. **Legal and strategic** tools are available and under construction, such as the **UN Convention** on the rights of persons with disability, or the **European Declaration of fundamental rights**, the **EU social pillar** and the **European disability strategy**. These can form the basis for clear and well-structured plans for addressing the rights and needs of people with disabilities, including **intellectual disability**, which should be implemented and monitored at the **local, national and international levels**.



THEME 3

SHARE, CARE, CURE: TRANSFORMING CARE FOR RARE DISEASES BY 2030

THEME LEADERS:

Prof. Alberto Pereira, Coordinator of the European Reference Network on Rare Endocrine Conditions (Endo-ERN) & Leiden University Medical Centre, The Netherlands.

Dr. Birute Tumiene, Clinical Geneticist, Coordinator for Competence Centres, Vilnius University Hospital Santaros Clinics, Lithuania.

Matt Bolz-Johnson, ERN and Healthcare Advisor, EURORDIS

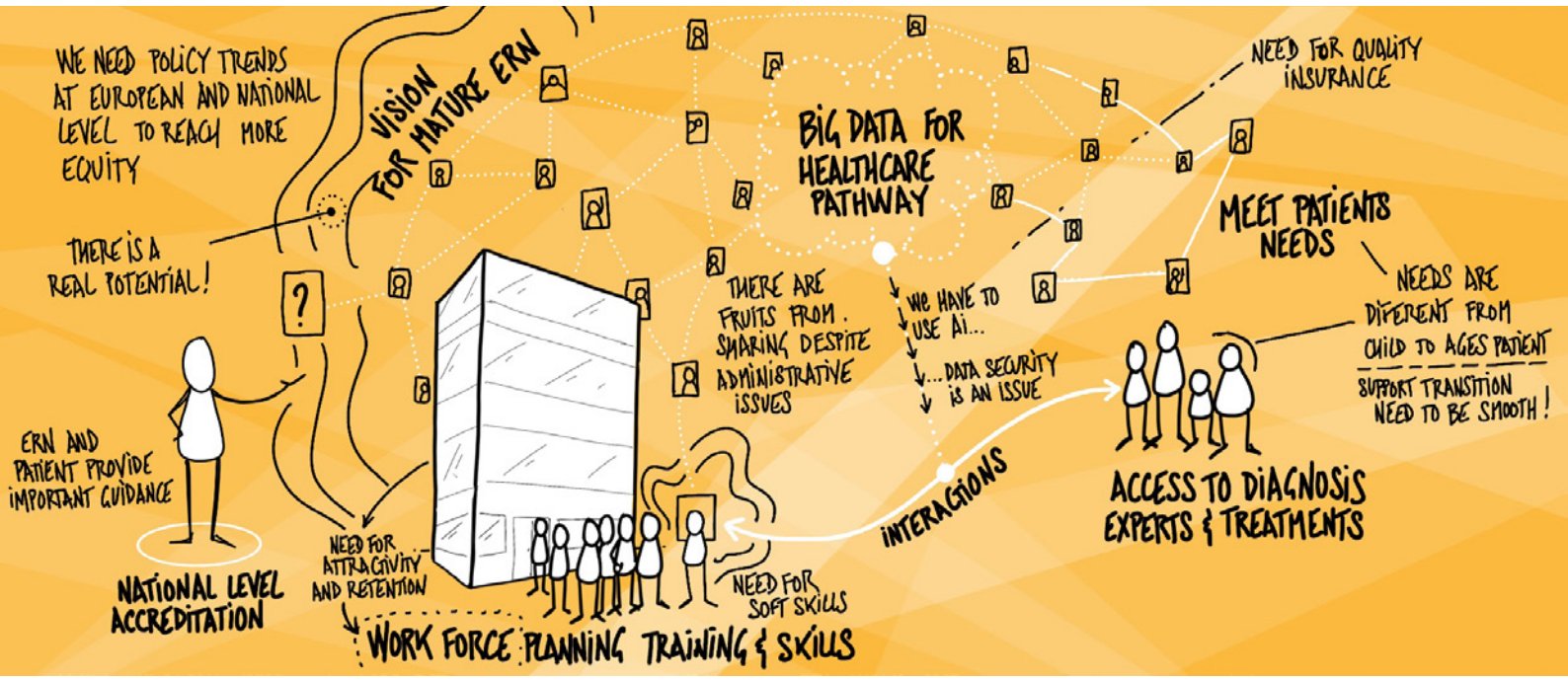
THEME SUPPORT :

Ines Hernando, ERN and Healthcare Director, EURORDIS

View **full programme**

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

- Patients and the rare disease community expect more **European solidarity**, greater **EU competencies** to develop pan-European policies and more **equity in access** to healthcare. At the same time, Member States should strengthen their individual approaches to rare diseases via a new **national planning process**. A renewed focus in national plans will force them to have a defined approach to rare diseases.
- Today we have a highly diverse **quality assurance** landscape across Europe for rare disease Centres of Expertise. The 2011 EUCERD recommendation on quality of such Centres remains relevant and the cornerstone and basis for quality standards of **Centres of Expertise**. The implementation of these recommendations varies across the EU and further funding and resources are required to support the implementation of these **quality standards** and a sound quality assurance process in all rare disease Centres of Expertise.
- **Artificial Intelligence** holds great promises to support **clinical decisions**, establish risk-adjusted **digital healthcare pathways**, improve **care coordination**, and eventually improve **cost effectiveness**. However, further research into the use of technology to support innovative digital care pathways and tools is needed to substantiate these claims.
- Organising care for rare diseases requires a "**whole workforce**" approach and greater **European integration** into workforce planning and forecasting. Policy makers should design bundles of policy interventions that take into consideration retention and motivation along with supply and adequate skill-mix. This will allow flexibility between roles, training and continuous **education of experts, nurses, social workers** and **GPs**, and the role of expert patients and **community care planning**, etc.



THEME 4

WHEN THERAPIES MEET THE NEEDS: ENABLING A PATIENT-CENTRIC APPROACH TO THERAPEUTIC DEVELOPMENT

THEME LEADERS:

Diego Ardigo, Global Rare Diseases R&D Head, Chiesi Farmaceutici, Italy & Chair, Therapies Scientific Committee of IRDiRC

Virginie Hivert, Therapeutic Development Director, EURORDIS and Vice-Chair, Therapies Scientific Committee of IRDiRC

THEME SUPPORT:

Maria Cavaller, Patient Engagement Junior Manager, EURORDIS

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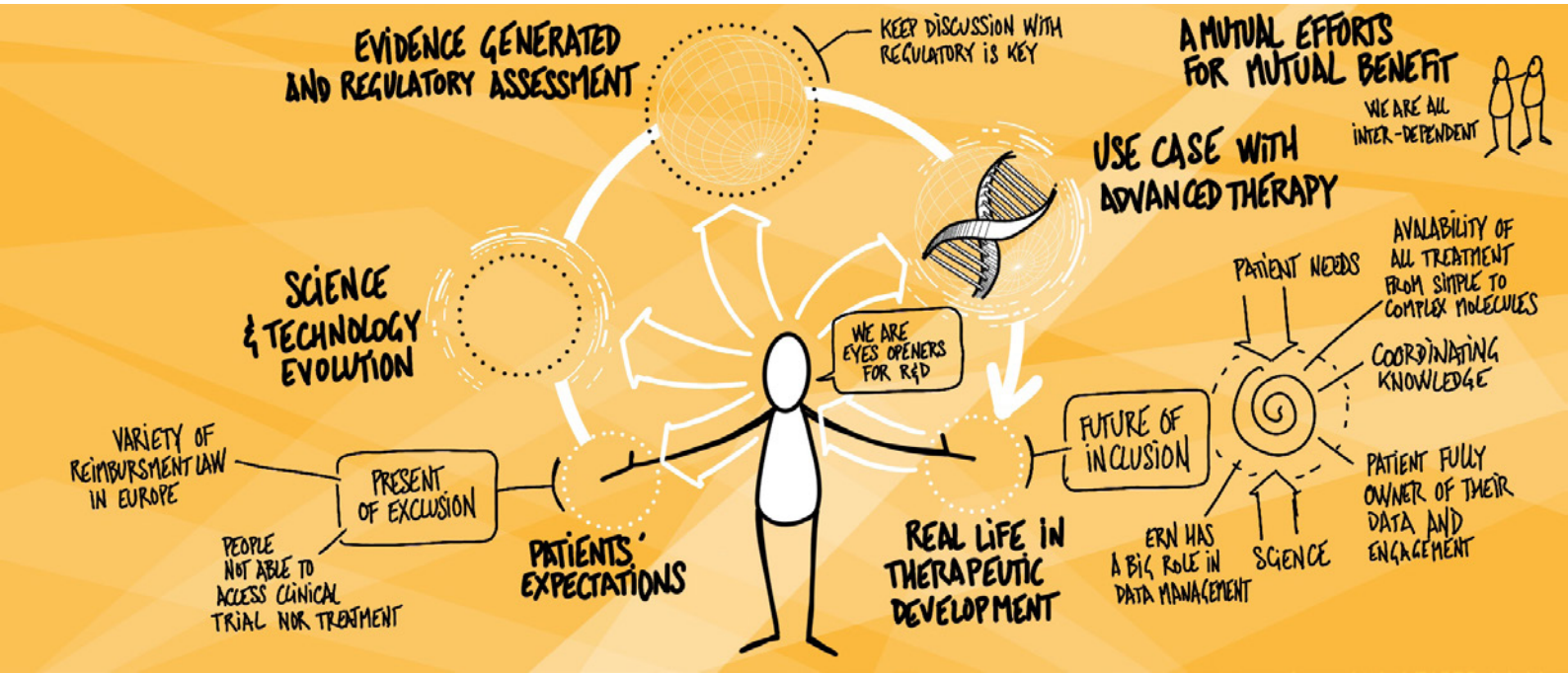
The main conclusions that came through in this Theme suggest that:

- Few **rare diseases** are getting all the attention and the **majority of them are still being disregarded** (no diagnosis, no public research, and no medicine development). This is either because they are too rare to attract research grants and efforts, there is too limited organised (available, accessible, usable, published) information, or it is too technologically complex to tackle. Identically, **patients are suffering from inequality in terms of access** to clinical trials, early access programmes or marketed treatments and these inequalities are even more striking in some countries.
- We have been looking at a '**future of inclusion**' which builds upon the right balance between the push of science, technology and business and the pull of patient's needs. This will be possible a) by generating a **coordinated research agenda** helping to redirect public and private investments and efforts towards currently disregarded diseases; b) by having **patients even more empowered and systematically involved** into the research and development processes from design to execution including on the aspects pertaining to data collection/data sharing; c) by integrating the **information collected** through real world evidence together with well-designed clinical trials in order to fulfil the **scientific and regulatory standards** while covering all gaps in knowledge and enhancing the value of medicinal products; and d) by considering the **whole spectrum of approaches** from the development of repurposed drugs to the most advanced therapies.
- To **ensure this transition**, Theme 4 showed that a lot of tools, initiatives and resources are **already available**. However, there is the need for **consolidation of these assets**, for **collaboration** amongst stakeholders and amongst countries and for **sharing risks and decisions** for the individual and collective benefit. It was highlighted **how crucial the role of patients is**, in understanding their condition and the clinical relevance and real value of a drug, but also in being owners of their data and full contributors to the research & development programmes or to the regulatory & HTA assessments. To make the transition successful, we also need to **leverage the existing networks and initiatives** such as the European Joint Programme for Rare Diseases (EJP-RD), to sustain the work conducted in IRDiRC – International Rare Diseases Research Consortium (in particular the **Orphan Drug Development Guidebook**) and to keep fostering the development of the **European Reference Networks**. The latter have not only been instrumental in structuring the healthcare pathways but are also bearing tremendous potential in terms of data collection, including for patient reported quality of life and clinical outcome data, and in increasing their capacity to perform clinical research and establish actual **Clinical Research Networks**.
- '**Mutual effort for mutual benefit**': to foster the development of **therapies that meet the needs of the patients**, we need the **collaboration of all stakeholders**, allowing complementarity of skills and resources while respecting the scope and specificities of each of them. 'How independent we can all be if we support each other and we also recognise our inter-dependence.'



THEME 5

ACHIEVING THE TRIPLE AS BY 2030: ACCESSIBLE, AVAILABLE AND AFFORDABLE THERAPIES FOR PEOPLE LIVING WITH A RARE DISEASE

THEME LEADERS:

Dimitrios Athanasiou, World Duchenne Organisation and European Patient Forum, Greece

Prof. Josep Torrent-Farnell, Universitat Autònoma de Barcelona, Spain

THEME SUPPORT:

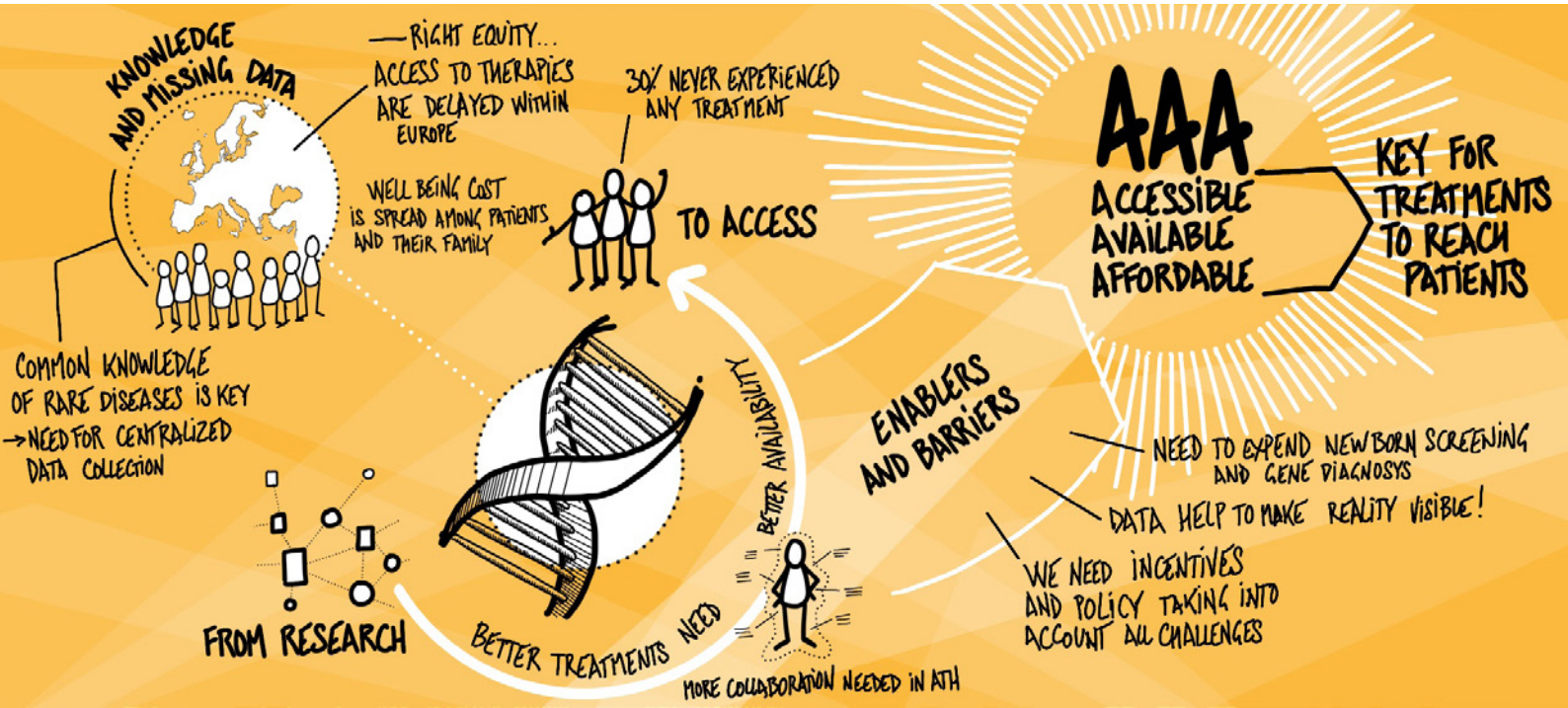
Simone Boselli, Public Affairs Director, EURORDIS

Ana Palma, Senior Director Global HTA & Patient Access Lead, SOBI

View **full programme**

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

- Access to therapies is **delayed** in many countries and the approved treatments are only available in some countries and not in others, posing a question of **equal rights under the EU law**. In the most recent **EURORDIS Rare Barometer Voices survey**, 31% of the 7,500+ respondents still have never received any treatment either because there is **no treatment available**, they could not take part in the **clinical trial** or the treatment was **not affordable**. For rare disease patients, the numbers reinforce the reality of the lives that they live, the impact of the condition and also the need to make changes in all aspects of the healthcare systems, particularly with respect to how we make the therapies **accessible, available and affordable**.
- New disruptive technologies like **gene therapies** are generating a lot of expectations from our community to have real transformative and potentially **curative treatment**, but they are creating challenges to healthcare systems. From 2025, the FDA is expected to approve 20 gene therapies per year, but there are still a number of barriers notably regarding the identification of patients. **Newborn screening** will need to be expanded and sometimes even a **genetic diagnosis** will be required. There are still clear gaps between regulators and HTA bodies in the evidence requirements. There should be more flexibility on the use of pre-existing data for HTA, patients and doctors could work on core outcomes for the conditions to facilitate assessment and for the use of registries.
- To reach a **common approach** and **reduce the inequalities in access**, European collaboration should be fostered, to overcome key common barriers such the **lack of trust** between different stakeholders, fragmentation of **data accessibility** and sharing, which are still impeding better processes that neither lead to better treatment nor to greater availability. However there are also key enablers, such as multi-stakeholders and community approaches, and **public private partnerships** with good governance to achieve common goals. Particularly on health technology assessment (HTA), the resounding support for a common European approach was heard as optimal, as well as initiatives to reduce perceived lack of data with the use of best data sets available, including **real world evidence** and patient driven evidence generation.
- When applying all of the discussion to the **Rare 2030 scenarios**, whilst the polling of participants indicated that **investment for social justice** is most preferable, scenario 2 **(fast over fair)** emerged as the more realistic in the context of current healthcare systems. A very worrisome scenario for rare diseases, one which is to a large extent already experienced today and that calls for patient organisations to advocate more because access will not be assured. Continuing to work together and to **develop reliable structures** for **data generation** will support patients with evidence based advocacy, inform **research needs** and regulatory HTA and payers decision-making, thus positively impacting access and availability as well as increasing trust.



THEME 6
THE DIGITAL HEALTH REVOLUTION: HYPE VS. REALITY

THEME LEADERS:

Julián Isla, Data and Artificial Intelligence Resource Manager, Microsoft & Chief Scientific Officer, Dravet Syndrome European Foundation, Spain.

Justina Januševičienė, Head of Healthcare Innovation Development Centre, Lithuanian University of Health Sciences, Lithuania.

Brian O'Connor, Chair, European Connected Health Alliance, UK/Ireland

THEME SUPPORT :

Denis Costello, Executive Director, CML Advocates Network, Spain.

Marta Campabadal, RareConnect Manager, EURORDIS

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

- There is a significant role and opportunity now for patients in the **digital health field**. Patients and patient organisations can make an impactful difference to avail now of the **technological enablers** that are coming on stream. They should be empowered and proactive in the use of these models and **digital technologies**. The idea of patient organisations as intermediaries and data curators was presented and the role of the **FAIR principles** (guidelines to improve the **Findability, Accessibility, Interoperability, and Reuse of data**) was raised in that context. At the same time, patients have no incetive to contribute data. Yet, they need such incentives, as well as some control over the use of their data. Patient organisations can help create an environment where this issue is tackled.

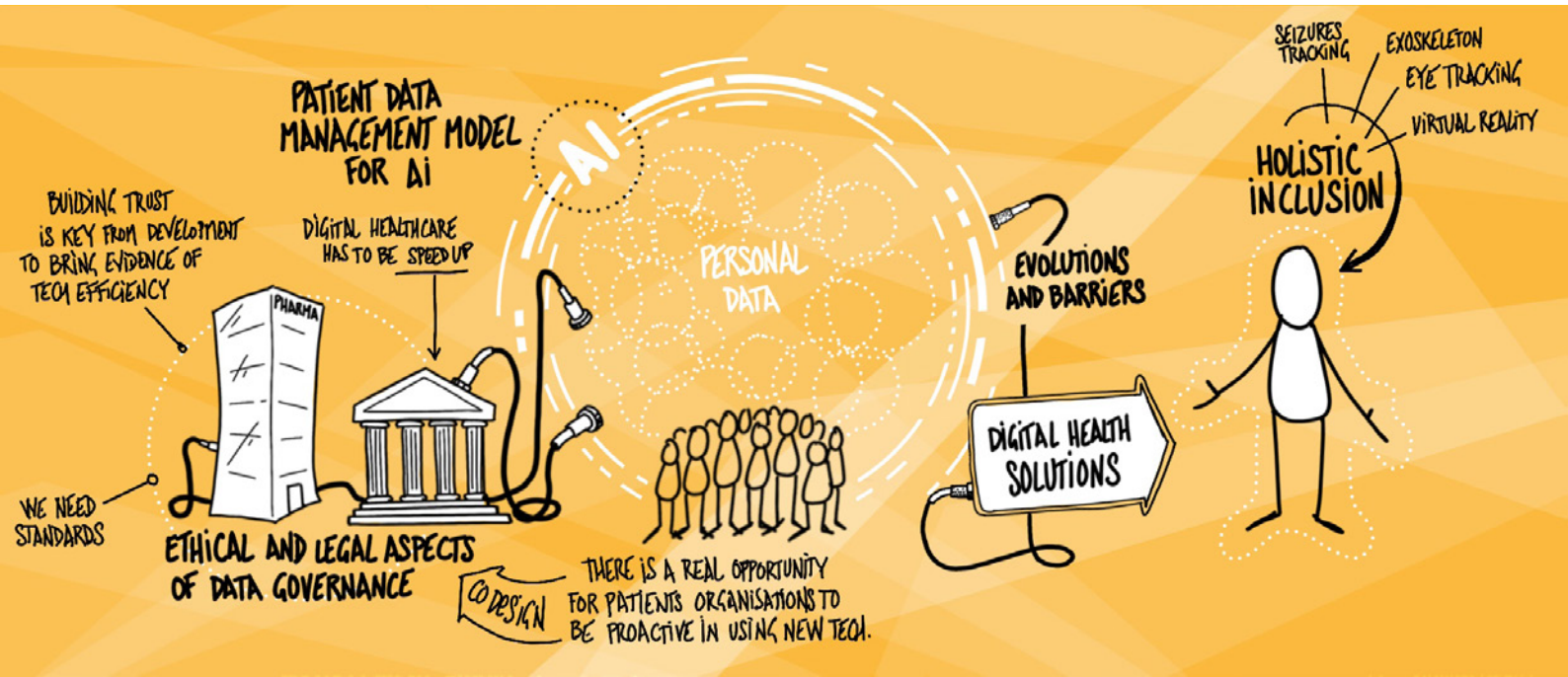
- Hospitals have traditionally been collecting data and defining the type of data that is relevant for their work. There should be a shift to allow data to **be person-focused** rather than scattered between institutions that only focus on the data that are useful for their own functioning, which might be different to what is useful or important for an individual. Patients and citizens should have access to all the data that concerns them.

- The **data sharing debate** should shift from trying to make people share their data more freely, to also looking at hurdles other actors create (public and private institutions) by not cooperating with each other and keeping data only for their own purposes. There are different ways in going forward and **EU Member States are taking various national approaches.** Although these are not incompatible, still a greater **European-level coordination** is needed.

- **Privacy is a big issue** for patients when deciding whether or not to share data. The underlying reason for this concern is the potential **discrimination that may occur at many levels (insurance, housing, employment, education)**. At the same time, the need for a solution is acute for most people living with a rare disease. An important addition to this debate to shift policies towards creating a **safer environment free of discrimination**, rather than increasing privacy requirements which often have an **impact on research and innovation**.

- Policies are not catching up with reality, creating a situation where **incredible technological advancements clash against little scale-up and a lot of uncertainty** about the integration of those solutions into routine care. **More investment** should be made to properly evaluate solutions that are available and then provide funding to scale them up and fully integrate them into the healthcare system. For the moment **many obstacles** are in place, but they might differ from country to country (ranging from concerns regarding **liability and ensuring privacy, to reimbursement issues, and interoperability hurdles**).

- There is a need to promote **digital health equally amongst all European regions**, as it is clear that advanced applications like Artificial Intelligence are not represented at the same level or stage of development in the different regions.



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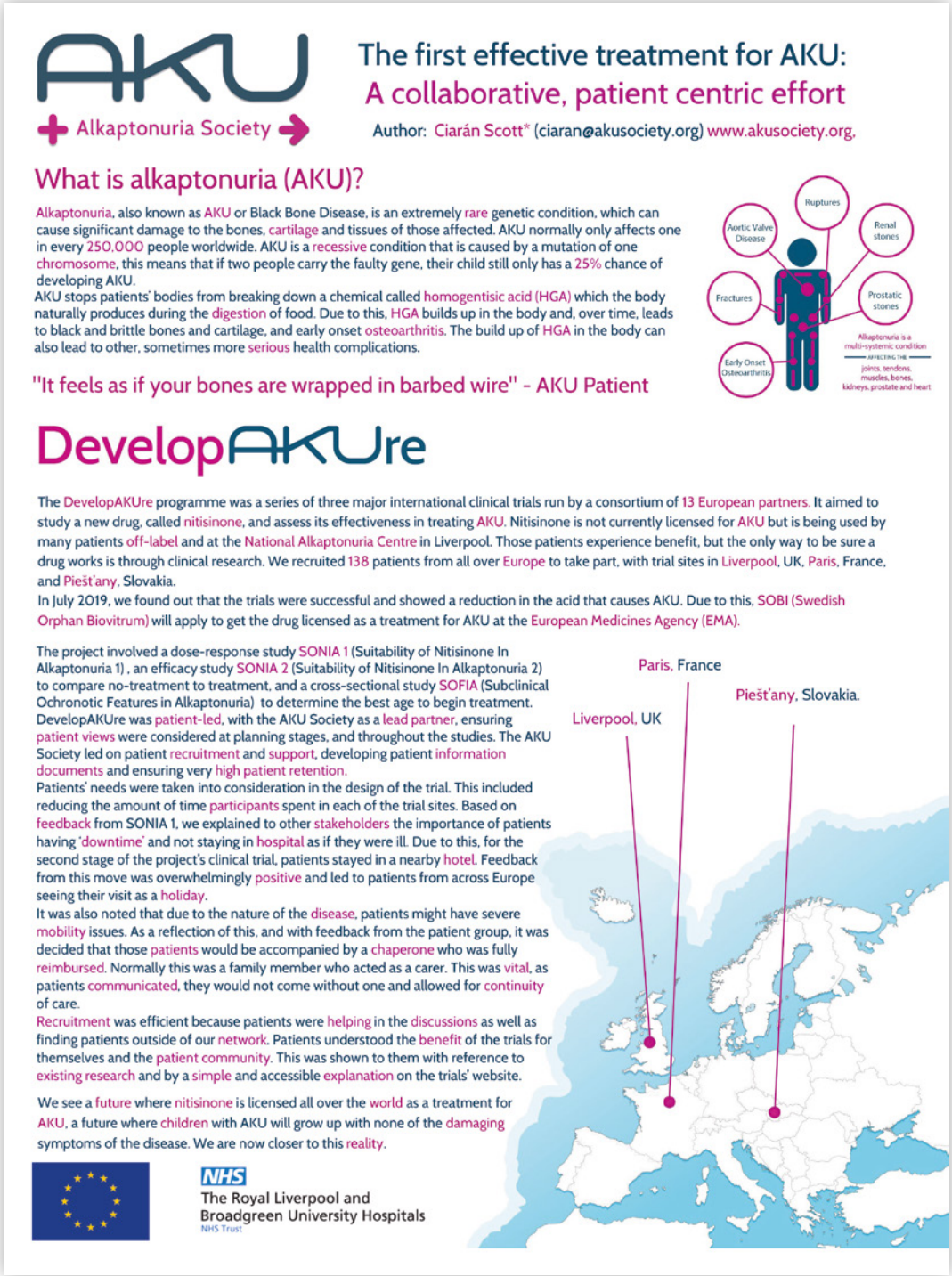
POSTER **WINNERS**

1st Place: P 163

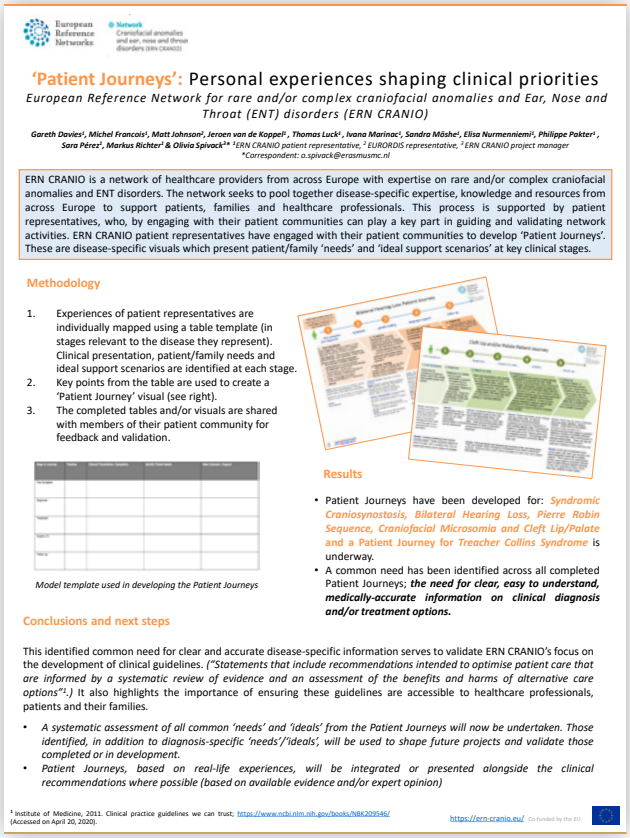
A collaborative and patient-centric effort to find the first effective treatment for alkaptonuria. (Nick Sireau)



CONFERENCE POSTERS

2nd Place: P 267

'Patient Journeys': Personal experiences shaping clinical priorities (Olivia Spivack)



'Patient Journeys': Personal experiences shaping clinical priorities

European Reference Network for rare and/or complex craniofacial anomalies and Ear, Nose and Throat (ENT) disorders (ERN CRANIO)

Gareth Davies[1], Michel Francois[1], Matt Johnson[2], Jeroen van de Koppel[2] , Thomas Luck[1] , Ivana Marinac[1], Sandra Möshe[2], Elisa Nurmenniemi[2], Philippe Pokter[1] , Sara Pérez[3], Markus Richter[2] & Olivia Spivack[2,4] [1]ERN CRANIO patient representative, [2] EURORDIS representative, [3] ERN CRANIO project manager
*Correspondent: o.spivack@erasmusmc.nl

ERN CRANIO is a network of healthcare providers from across Europe with expertise on rare and/or complex craniofacial anomalies and ENT disorders. The network seeks to pool together disease-specific expertise, knowledge and resources from across Europe to support patients, families and healthcare professionals. This process is supported by patient representatives, who, by engaging with their patient communities can play a key part in guiding and validating network activities. ERN CRANIO patient representatives have engaged with their patient communities to develop 'Patient Journeys'. These are disease-specific visuals which present patient/family 'needs' and 'ideal support scenarios' at key clinical stages.

Methodology

1. Experiences of patient representatives are individually mapped using a table template (in stages relevant to the disease they represent). Clinical presentation, patient/family needs and ideal support scenarios are identified at each stage.
2. Key points from the table are used to create a 'Patient Journey' visual (see right).
3. The completed tables and/or visuals are shared with members of their patient community for feedback and validation.

Model template used in developing the Patient Journeys

Results

- Patient Journeys have been developed for: *Syndromic Craniosynostosis, Bilateral Hearing Loss, Pierre Robin Sequence, Craniofacial Microsomia and Cleft Lip/Palate* and a Patient Journey for *Treacher Collins Syndrome* is underway.
- A common need has been identified across all completed Patient Journeys; ***the need for clear, easy to understand, medically-accurate information on clinical diagnosis and/or treatment options.***

Conclusions and next steps

This identified common need for clear and accurate disease-specific information serves to validate ERN CRANIO's focus on the development of clinical guidelines. *("Statements that include recommendations intended to optimise patient care that are informed by a systematic review of evidence and an assessment of the benefits and harms of alternative care options"[1].)* It also highlights the importance of ensuring these guidelines are accessible to healthcare professionals, patients and their families.

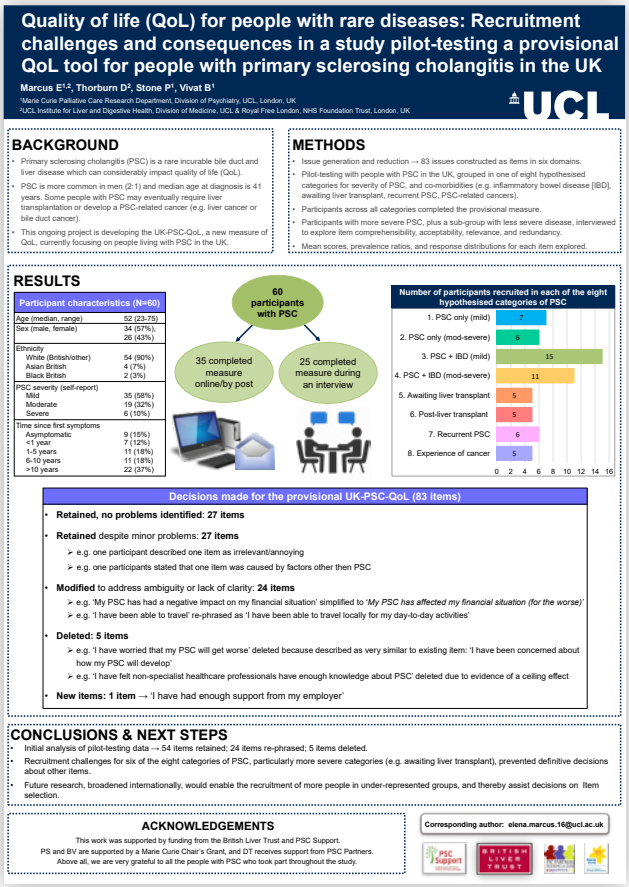
- *A systematic assessment of all common 'needs' and 'ideals' from the Patient Journeys will now be undertaken. Those identified, in addition to diagnosis-specific 'needs'/'ideals', will be used to shape future projects and validate those completed or in development.*
- *Patient Journeys, based on real-life experiences, will be integrated or presented alongside the clinical recommendations where possible (based on available evidence and/or expert opinion)*

[1] Institute of Medicine, 2011. Clinical practice guidelines we can trust; https://www.ncbi.nlm.nih.gov/books/NBK209546/ (Accessed on April 20, 2020).

https://ern-cranio.eu/ Co-funded by the EU

3rd Place: P 268

Quality of life (QoL) for people with rare diseases: Recruitment challenges and consequences in a study pilot-testing the UK-PSC-QoL, a provisional QoL tool for people with primary sclerosing cholangitis (PSC) in the UK (Elena Marcus)



Quality of life (QoL) for people with rare diseases: Recruitment challenges and consequences in a study pilot-testing a provisional QoL tool for people with primary sclerosing cholangitis in the UK

Marcus E[1,2], Thorburn D[2], Stone P[1], Vivat B[1]
[1]Marie Curie Palliative Care Research Department, Division of Psychiatry, UCL, London, UK
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BACKGROUND

- Primary sclerosing cholangitis (PSC) is a rare incurable bile duct and liver disease which can considerably impact quality of life (QoL).
- PSC is more common in men (2:1) and median age at diagnosis is 41 years. Some people with PSC may eventually require liver transplantation or develop a PSC-related cancer (e.g. liver cancer or bile duct cancer).
- This ongoing project is developing the UK-PSC-QoL, a new measure of QoL, currently focusing on people living with PSC in the UK.

METHODS

- Issue generation and reduction → 83 issues constructed as items in six domains.
- Pilot-testing with people with PSC in the UK, grouped in one of eight hypothesised categories for severity of PSC, and co-morbidities (e.g. inflammatory bowel disease [IBD], awaiting liver transplant, recurrent PSC, PSC-related cancers).
- Participants across all categories completed the provisional measure.
- Participants with more severe PSC, plus a sub-group with less severe disease, interviewed to explore item comprehensibility, acceptability, relevance, and redundancy.
- Mean scores, prevalence ratios, and response distributions for each item explored.

RESULTS

Participant characteristics (N=60)	
Age (median, range)	52 (23-75)
Sex (male, female)	34 (57%), 26 (43%)
Ethnicity	
White (British/other)	54 (90%)
Asian British	4 (7%)
Black British	2 (3%)
PSC severity (self-report)	
Mild	35 (58%)
Moderate	19 (32%)
Severe	6 (10%)
Time since first symptoms	
Asymptomatic	9 (15%)
<1 year	7 (12%)
1-5 years	11 (18%)
6-10 years	11 (18%)
>10 years	22 (37%)

60 participants with PSC → 35 completed measure online/by post; 25 completed measure during an interview

Number of participants recruited in each of the eight hypothesised categories of PSC

Decisions made for the provisional UK-PSC-QoL (83 items)

- **Retained, no problems identified**: **27 items**
- **Retained** despite minor problems: **27 items**
  - e.g. one participant described one item as irrelevant/annoying
  - e.g. one participants stated that one item was caused by factors other then PSC
- **Modified** to address ambiguity or lack of clarity: **24 items**
  - e.g. 'My PSC has had a negative impact on my financial situation' simplified to *'My PSC has affected my financial situation (for the worse)'*
  - e.g. 'I have been able to travel' re-phrased as 'I have been able to travel locally for my day-to-day activities'
- **Deleted: 5 items**
  - e.g. 'I have worried that my PSC will get worse' deleted because described as very similar to existing item: 'I have been concerned about how my PSC will develop'
  - e.g. 'I have felt non-specialist healthcare professionals have enough knowledge about PSC' deleted due to evidence of a ceiling effect
- **New items: 1 item** → 'I have had enough support from my employer'

CONCLUSIONS & NEXT STEPS

- Initial analysis of pilot-testing data → 54 items retained; 24 items re-phrased; 5 items deleted.
- Recruitment challenges for six of the eight categories of PSC, particularly more severe categories (e.g. awaiting liver transplant), prevented definitive decisions about other items.
- Future research, broadened internationally, would enable the recruitment of more people in under-represented groups, and thereby assist decisions on Item selection.

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Above all, we are very grateful to all the people with PSC who took part throughout the study.

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