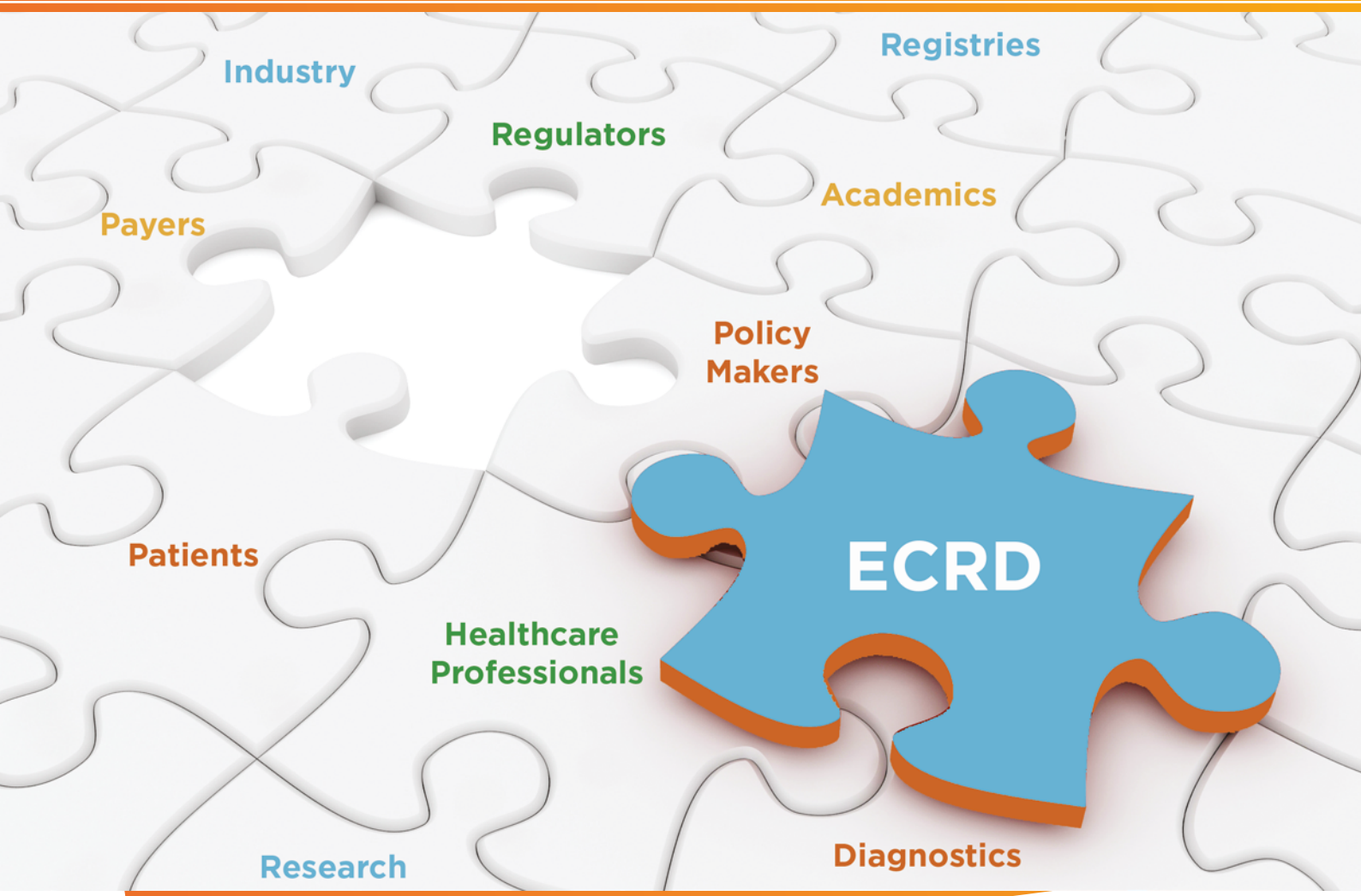

7th European Conference on Rare Diseases & Orphan Products

8-10 May 2014, Andel's Hotel, Berlin, Germany

The Rare Disease Puzzle:
Bringing the Picture to Life

In the past two decades we have identified the pieces that address the needs of the rare disease community. The challenge remains to combine those pieces into a picture that drives action forward with clarity and energy into a coherent and sustainable strategic plan

Executive Summary

A conference organised by

EURORDIS
Rare Diseases Europe

Co-organised by

FIFTY YEARS 1964-2014 DIA®
www.diahome.org

www.rare-diseases.eu

The European Conferences on Rare Diseases and Orphan Products

- 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 – patient representatives, academics, healthcare professionals, industry, payers, regulators and policymakers.
- It is a biennial event, providing the state-of-the-art of the rare disease environment, as well as on monitoring and benchmarking initiatives. It covers research, development of new treatments, healthcare, 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 with them, but efforts are complementary, fully respecting initiatives of all.

Speaker presentations and poster abstracts are available on the website:
www.rare-diseases.eu

EURORDIS
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ACKNOWLEDGEMENTS AND CREDITS

We wish to thank the following institutions for their active collaboration

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AFMTÉLÉTHON INNOVER POUR GUERIR

In partnership with

EUROPEAN MEDICINES AGENCY SCIENCE MEDICINES HEALTH

orphanet

ebe european biopharmaceutical enterprises

EuropaBio The European Association for Bioindustries

NORD National Organization for Rare Disorders

CORD Canadian Organization for Rare Disorders

achse Allianz Chronischer Seltener Erkrankungen

ESHG

European Federation of Internal Medicine

FDMIE SFIME FDIME

Continuing Education

DIA meetings and training courses are approved by the Commission for Professional Development (CPD) of the Swiss Association of Pharmaceutical Professionals (SwAPP) and the Swiss Society of Pharmaceutical Medicine (SGPM) and will be honoured with credits for pharmaceutical medicine. The ECRD has been awarded with 9 CPD credits from the Faculty of Pharmaceutical Medicine (FPM) of the Royal College of Physicians (RCP) of the UK. All participants are eligible for these credits.

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Miroslaw Zielinski
Chairman, Polish National Forum on the Treatment of Orphan Diseases (ORPHAN), Poland

OBJECTIVES:

- **Disseminate** the most up-to-date health 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 health issues related to the rare disease environment

PARTICIPANTS:

768 ATTENDEES

138 SESSION CHAIRS / SPEAKERS

38 SESSIONS

+200 POSTERS

43 COUNTRIES

KEY POINTS OF SUCCESS:

- **Pertinent conference programme**
  Discussion of hot topics: organisation of healthcare, research, development and availability of orphan medicinal products in Europe
- **High-level session chairs and speakers – Diversity and complementarity of topics**
- **Case studies from: Germany, Denmark, France, Belgium, Austria, UK, Portugal, Netherlands, Spain, Italy, Canada, USA, Sweden, Finland, Romania, Ireland, Norway and Hungary**
- **Involving a diversity of stakeholders: patient representatives, academics, healthcare professionals, industry, payers, regulators, and policymakers**

KEY FEATURES OF THE CONFERENCE

Highlights of the successful 7th European Conference on Rare Diseases & Orphan Products (ECRD 2014) include:

- **Highest participation since the first ECRD 2001 with 768 participants on-site**
- **Participants from 43 countries, including 26 from EU/EEA**
- **15 Eastern European countries**
- **A comprehensive and multi-stakeholder conference programme: 138 speakers, 38 sessions, +200 posters**

PARTICIPATION

A total of 768 participants attended ECRD 2014 Berlin (a 18.4% increase from 2012), including 138 speakers and session chairs. 40 full patient fellowships were awarded.

The participants at ECRD 2014 represented 43 countries, including 26 from the EU/EEA. The total number of participants attending from EU countries was 610 (compared to 521 in 2012), corresponding to 80% of the overall attendees. Participants attending from EEA countries corresponded to 82% of the overall number of attendees (629).

Representatives from 15 Eastern European states (Bosnia and Herzegovina, Bulgaria, Croatia, Czech Republic, Georgia, Hungary, Latvia, Republic of Macedonia, Poland, Romania, Russian Federation, Serbia, Slovakia, Slovenia, Ukraine) were present at the Conference. In total, participants from Eastern Europe represented 11.2% of the attendance.

Non-European participation from Australia, Brazil, Canada, India, Israel, Japan, South Africa, Taiwan, Province of China, and USA gathered a delegation of 53 international participants representing 7% of the total attendance.

By country of origin, Germany had the largest delegation of 129 people, representing 16.8% of participants. Mobilisation of German patient organisations via ACHSE, the German National Alliance for Chronic Rare Diseases, as well as interpretation of sessions in German and the early-bird registration fee that was made available throughout the registration process were all key factors for this success.

The other countries with an important presence at the Conference included the UK (93 participants), Italy (71 participants), France (48 participants), Belgium (48 participants), the Netherlands (39 participants) and Switzerland (33 participants). The number of Polish participants also saw a record high of 25 at the Conference. This can be explained by the close proximity and ease of access to Germany from Poland.

Interpretation of the opening and plenary sessions from English into five languages (French, German, Polish, German and Russian) was undoubtedly an important factor for registration: overall, delegates originating from countries where these languages dominate represented 64% of all delegates.

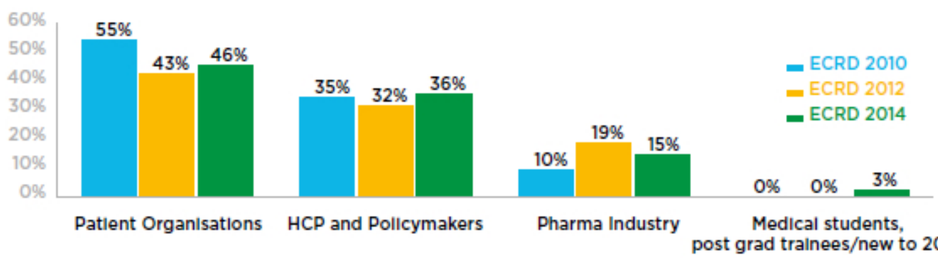
Evolution of the total number of participants to the European Conference on Rare Diseases since 2001

WHO WERE THE PARTICIPANTS?

In terms of delegates' categories, the composition of ECRD 2014 Berlin differed slightly compared to previous events: the introduction of a new category (medical students and post-graduate trainees) made up 3% of total figures. There were slightly fewer industry representatives (15% versus 19% in 2012). The proportion of healthcare professionals and policymakers was more or less stable (36%).

The majority of the official Conference partners were represented among the speakers and session chairs, including the DIA, ACHSE, CORD, Europa-Bio-EBE, EMA/COMP, ESHG and Orphanet.

Categories of delegates: Comparison between ECRDs 2010, 2012, and 2014



Country	*No. of delegates*
Australia	2
Austria	14
Belgium	48
Bosnia and Herzegovina	1
Brazil	2
Bulgaria	4
Canada	9
Croatia	2
Czech Republic	5
Denmark	19
Finland	17
France	48
Georgia	2
Germany	129
Greece	4
Hungary	10
Iceland	1
India	1
Ireland	13
Israel	1
Italy	71
Japan	3

Country	*No. of delegates*
Latvia	2
Luxembourg	9
Republic of Macedonia	1
Netherlands	39
Norway	18
Poland	25
Portugal	3
Romania	4
Russian Federation	16
Serbia	7
Slovakia	2
Slovenia	2
South Africa	1
Spain	25
Sweden	21
Switzerland	33
Taiwan, Province of China	1
Turkey	2
Ukraine	2
United Kingdom	93
United States	33
Non-specified	22

This year we introduced the ECRD Mobile App, enabling participants to access information about ECRD before, after and during the conference. The App's instant messaging feature allowed participants to contact each other on-site, and participants were able to access a wide variety of material via the App, including the programme, floor plans, posters, speaker presentations and glossary of acronyms. A total of 338 participants downloaded the App, 43% of all participants.

ECRD 2014 AT A GLANCE

Thursday 8 May 2014

14:30 – 17:00

Tutorial 1
HTA 101 for Rare Diseases

Tutorial 2
RD Connect (an integrated platform connecting registries, biobanks and clinical bioinformatics for Rare Disease research)

Tutorial 3
Supporting the pathway to trials for Rare Diseases: clinical trial design and other considerations

Tutorial 4
How to get the best out of Orphanet data

Friday 9 May 2014

09:00 – 14:00

Opening Session, Plenary Session, Poster Session

	Theme 01 Improving Healthcare Services	Theme 02 Knowledge Generation and Dissemination	Theme 03 Research from Discovery to Patients	Theme 04 State of the Art and Innovative Practices in Orphan Products	Theme 05 Emerging Concepts and Future Policies for Rare Disease Therapies	Theme 06 Beyond Medical Care
14:00 – 15:30	Session 0101 Centres of Expertise – Part 1 (models and practical examples)	Session 0201 The role, risks and relevance of registries in shaping therapy development to 2020	Session 0301 Shaping Rare Disease Research Policy	Session 0401 Current landscape of Policy Development on Orphan Products & Rare Disease Therapies	Session 0501 Early Dialogue and Horizon Scanning of Product development to address unmet medical needs	Session 0601 Identifying specific social challenges of rare diseases
16:30 – 18:00	Session 0102 Centres of Expertise – Part II (designation & evaluation)	Session 0202 Session 0202: A collaborative model to progress knowledge and research	Session 0302 Addressing the Gaps in Research at International Level to Identify Opportunities	Session 0402 Facts on current patient access challenges to orphan products	Session 0502 Session 0502: How to shape a better framework for orphan drug development: EMA/FDA collaboration	Session 0602 Different approaches to the social challenges of rare diseases: Social Policy

Saturday 10 May 2014

09:00 – 10:30	Session 0103 European Reference Networks (ERNs)	Session 0203 Making the invisible visible: The coding of Rare Diseases in Health Information Systems	Session 0303 Incentives to create a favourable eco-system	Session 0403 EMA / Health Technologies Assessment (HTA) interfacing on rare disease therapies	Session 0503 Progressive Patient Access Schemes & Patient Involvement in Benefit-Risk Assessment	Session 0603 Concrete Solutions to Social Challenges: Essential tools for the integration of rare diseases into Social Services
11:30 – 13:00	Session 0104 Addressing the Challenges of Healthcare Pathways	Session 0204 Delivering Help and Support in a virtual world: what will work best?	Session 0304 Breakthroughs in Science	Session 0404 Shortages in Authorised Medicines for Rare Diseases	Session 0504 Mechanism of Coordinated Access (MOCA) and Transparent Value Framework, Managed Entry Agreements	Session 0604 Can people living with a rare disease be independent? Inspiring personal stories
14:00 – 15:30	Session 0105 Advances in Diagnostic Possibilities for Undiagnosed Patients	Session 0205 Knowledge at the point of care: getting the facts just in time or just in case	Session 0305 Pre-competitive tools and resources / public-private partnership in the area of rare diseases	Session 0405 Understanding of Orphan Therapies Off-Label Uses and Their New Challenges	Session 0505 Emerging Ideas for Sustainable Access to Orphan Medicinal Products	Session 0605 Can people living with a rare disease be independent? Inspiring solutions by providers
15:45 – 17:15	Session 0106 Improving the Quality of and Access to Diagnostic Services	Session 0206 Hype, help or harm? The impact of media promotion of rare diseases	Session 0306 Whose data is it?: Stimulating Research and Removing Barriers	Session 0406 Empowering Patient Advocates in Drug Development	Session 0506 Rare disease treatments beyond medicinal products	Session 0606 How Centres of Expertise should/could interface with Social Services

OPENING SESSION

Friday 9 May 2014

09:00 – 9:45

Welcome address and opening remarks by co-organisers:
Avril Daly, Vice-President EURORDIS, Chair of Rare Diseases Ireland GRDO, CEO Fighting Blindness, Ireland
Jytte Lyngvig, Director, DIA Europe, Middle East & Africa, Switzerland

Keynote Addresses:
Annette Widmann-Mauz, Parliamentary State Secretary, Federal Ministry of Health, Germany
Christoph Nachtigäller, President, German National Alliance for Chronic Rare Diseases (ACHSE), Germany
Irene Norstedt, Head of Unit, Personalised Medicine, Directorate for Health Research at the DG Research & Innovation, European Commission, EU
Lesley Greene, Vice-Chair COMP, Vice-President, CLIMB, Co-Chair Programme Committee ECRD 2014, UK

PLENARY SESSION

Friday 9 May 2014

10:15 – 12:00

Session Anchor:
Durhane Wong Rieger, President, CORD, Canada

Results of the European Commission implementation report on the Council Recommendation on Rare Diseases
John F. Ryan, Director Public Health, DG Health & Consumers, European Commission, EU

Round table discussion: The main challenges that remain in the areas of organisation of healthcare, research and the development and availability of orphan medicinal products in Europe
Ségolène Aymé, Emeritus, Director of Research, Director of International Affairs, ORPHANET-INSERM, France
Serge Braun, Chief Scientific Officer, AFM (Association Française contre Les Myopathies), France
Kate Bushby, Professor of Neuromuscular Genetics, MRC Centre for Neuromuscular Diseases, Institute of Genetic Medicine, International Centre for Life, Newcastle Upon Tyne Hospital, UK
Pauline Evers, EGAN (European genetic alliances network), Dutch Federation of Cancer Patient Organisations, The Netherlands
Wills Hughes-Wilson, Chair of Task Force for Rare Diseases & Orphan Drugs of EuropaBio-EBE, Chief Patient Access Officer, Vice President External Affairs, sobi (Swedish Orphan Biovitrum AB), Sweden
Tsveta Schyns, European Network for Research on Alternating Hemiplegia (ENRAH), Belgium
Bruno Sepodes, Chair, Committee for Orphan Medicinal Products (COMP), University of Lisbon, Portugal

THEMES

THEME 1 | IMPROVING HEALTHCARE SERVICES

Kate Bushby, Professor of Neuromuscular Genetics, MRC Centre for Neuromuscular Diseases, Institute of Genetic Medicine, International Centre for Life, Newcastle Upon Tyne Hospital, UK

Véronique Héon-Klin, German Federal Ministry of Health, Germany

The development of Centres of Expertise and European Reference Networks in the field of rare diseases is encouraged in the Council Recommendation on an Action in the Field of Rare Diseases and in the Directive on the Application of Patients' Rights in Cross-border Healthcare as a means of organising care for the thousands of heterogeneous rare conditions affecting scattered patient populations across Europe. The aim is to link all these Centres of Expertise together through European Reference Networks (ERNs) in order to gather expertise and improve healthcare for rare disease patients.
Theme 1 will look at the experience gained so far in implementing these concepts. In Session 1, three good practical examples of different types of Centres of Expertise will provide the backdrop to a discussion on the organisation of these types of centres. In Session 2, speakers will focus on the quality criteria that should be considered when evaluating such centres. Session 3 will review where we stand with the establishment of European Reference Networks and what can be expected for rare diseases. Session 4 will explore the challenges of establishing seamless care pathways between the treating physician and the centres of expertise. In Session 5 and 6, the state of the art concerning advances and breakthroughs in diagnostic possibilities, such as the new generation sequencing, will be presented. The development of innovations in this field is very rapid and the challenge is now to ensure that these innovations benefit the people who need them most.

Session 0101
Friday 9 May, 14:00 - 15:30

CENTRES OF EXPERTISE – PART I (MODELS AND PRACTICAL EXAMPLES)
Interpretation DE + RU

Session Chair:
Hélène Dollfus, Professor of Medical Genetics, Faculty of Medicine, France

This session looks at the interpretation of the concept of Centres of Expertise in different countries. The role of Centres of Expertise in healthcare delivery for rare diseases is highlighted in the EUCERD recommendations, and a pillar of the national planning process. In addition, it is envisaged that Centres of Expertise will play a major role in the future European Reference Networks. In this session we explore the experience of two different models for Centres of Expertise- one focussed on a single disease and one with a much broader remit. Finally we will learn about the operation of Centres of Expertise as part of an advanced national plan with local networks in France.

Best Practices (example 1) – Specialised centre for epidermolysis bullosa (EB)
Leena Bruckner-Tuderman, Professor and Chair of the Department of Dermatology, University Medical Center, Albert-Ludwigs University of Freiburg, Germany

Best Practices (example 2)– Perspective of a Centre of Expertise with a broader remit than one rare disease
John Rosendahl Ostergaard, Clinical Professor, Centre of Rare Diseases, Aarhus University Hospital, Denmark

Best Practices (example 3): French Centre that demonstrates how they work in a broader healthcare system / how they interact with local networks
Pierre Sarda, Département de Génétique Médicale, Hôpital Arnaud de Villeneuve, France

Session 0102
Friday 9 May, 16:30 - 18:00

CENTRES OF EXPERTISE – PART 2 (DESIGNATION & EVALUATION)

Session Chair:
Enrique Terol, Policy Officer, DG SANCO, Health and Consumers Unit, European Commission, EU

This session will share best practice examples in the designation and evaluation of Centres of Expertise. Such centres are a key component of healthcare planning for patients with rare diseases, to provide improved capability for diagnosis and specialised management. In addition, Centres of Expertise will be core members of the European Reference Networks for Rare Diseases as planned under the Cross-Border Healthcare Directive.

Evaluation of Centres: The French experience since 2009
Sabine Sarnacki, Coordinator of expert centre on anorectal and rare pelvic malformations, Hopital Necker Enfants Malades, APHP and Paris Descartes University, France

Quality Criteria; Outcome Measures
Edmund Jessop, Medical adviser at NHS England, National Health Service, UK

Designation Process: How centres are selected; how to evaluate centres for rare diseases. How they plan to evaluate in the future
Enrique Terol, Policy Officer, DG SANCO, Health and Consumers Unit, European Commission, EU

Session 0103
Friday 9 May, 09:00 - 10:30

EUROPEAN REFERENCE NETWORKS (ERNS)

Session Chair:
Till Voigtländer, Clinical Institute of Neurology, Medical University of Vienna, Austria

In 2003/2004, the high-level European reflection process on cross-border healthcare initiated, amongst others, a discussion regarding the pan-European establishment and designation of highly specialised medical centres (so-called "Centres of Expertise") and their interlinking in European Reference Networks (ERN). Ten years later, the preparatory work for this concept has been largely finished with the implementation of the Cross-border Healthcare Directive (in October 2013) and the adoption and entering into force of accompanying legal acts (in May 2014). At the end of this year, the first call for proposals for ERNs will be published. At this transitional state, the session will highlight the current practices in the member states, as well as the details and implementation strategy of the ERN concept, followed by a panel discussion looking at existing experiences with the establishment and management of European networks in the fields of healthcare and research, as well as possible future strategies to ensure sustainability of ERNs once established.

Observatory Study – Building European Reference Networks in healthcare
Willy Palm, Dissemination Development Officer, European Observatory on Health Systems and Policies, Belgium

European Commission Report
Enrique Terol, Policy Officer, DG SANCO, Health and Consumers Unit, European Commission, EU

Round Table: Sustainability – Looking to the future
The example of the Austrian Epidermolysis Bullosa (EB) Centre and how it links into a broader network
Gabi Pohla-Gubo, Head of Epidermolysis Bullosa (EB) Academy, General Hospital Salzburg/Salzburger Landesklinikum (SALK), Paracelsus Medical University Salzburg (PMU), Austria

Past models such as TREAT-Neuromuscular Network (NMD): how this will change moving forward and the funding challenges
Kate Bushby, Professor of Neuromuscular Genetics, MRC Centre for Neuromuscular Diseases, Institute of Genetic Medicine, International Centre for Life, Newcastle Upon Tyne Hospital, UK

Session 0104
Saturday 10 May, 11:30 - 13:00

ADDRESSING THE CHALLENGES OF HEALTHCARE PATHWAYS
Interpretation DE + RU

Session Chair:
Sabine Sarnacki, Coordinator of expert centre on anorectal and rare pelvic malformations, Hopital Necker Enfants Malades, APHP and Paris Descartes University, France

The pathway from the General Practitioner (GP) , Primary Physician to Hospitals, Specialist Doctors, Centres of Expertise.

The Organisation of Healthcare Pathways around the Centres of Expertise: The French model
Sabine Sarnacki, Coordinator of expert centre on anorectal and rare pelvic malformations, Hopital Necker Enfants Malades, APHP and Paris Descartes University, France

Panel Discussion
Kate Bushby, Professor of Neuromuscular GeneticsMRC Centre for Neuromuscular Diseases, Institute of Genetic Medicine, International Centre for Life, Newcastle Upon Tyne Hospital, UK
Helena Kääriäinen, Research Professor, National Institute for Health and Welfare, Finland
Tsveta Schyns, European Network for Research on Alternating Hemiplegia (ENRAH), Belgium
Theda Wessel, Berlin Center for Rare Diseases, Charité – University Medicine Berlin, Germany

Session 0105
Saturday 10 May, 14:00 - 15:30

ADVANCES IN DIAGNOSTIC POSSIBILITIES FOR UNDIAGNOSED PATIENTS

Session Chair:
Tjitske Kleefstra, Clinical Geneticist, Radboud University Medical Center Nijmegen, the Netherlands

Whole exome and genome tests are rapidly being introduced in medicine. In this session examples of breakthroughs in next generation sequencing and their implication for research and clinic will be provided.

Sequencing: The Netherlands example
Tjitske Kleefstra, Clinical Geneticist, Radboud University Nijmegen Medical Centre, the Netherlands

UK Example: Deciphering Developmental Disorders (DDD) project and 100,000 Genomes
Wendy Jones, Wellcome Trust Sanger Institute / University of Cambridge, UK

FindZebra – What is currently available as computerised systems for diagnosis, what are their benefits and their limits
Ole Winther, Associate Professor, DTU Informatics, Technical University of Denmark, Denmark

Session 0106
Saturday 10 May, 15:45 - 17:15

IMPROVING THE QUALITY OF AND ACCESS TO DIAGNOSTIC SERVICES

Session Chair:
Gert Matthijs, Coordinator of EuroGentest, Laboratory for Molecular Diagnosis, Center for Human Genetics, Belgium

The new 'Massive Parallel Sequencing' or 'Next Generation Sequencing' (NGS) tools are rapidly being transformed from research applications to diagnostic methods. How will patients maximise profit from this evolution and how will healthcare systems cope with a new paradigm in genetic testing that will affect medical practice in its entirety? Clinical utility, technical validation and appropriate reimbursement models are a few of the key issues that deserve to be discussed at the community level and with all stakeholders.

How to Apply Next Generation Sequencing in Clinical Diagnostics: Challenges, guidelines and indications
Peter Bauer, Medical Specialist for Human Genetics, Head of Genomics Unit, Head of Molecular Genetic Diagnostics, Head of Core Unit for Applied Genomics, University of Tübingen, Germany

Ensuring Rapid Translation of Science to Services while Ensuring Quality and Affordability – Organisation of exome sequencing, an example from Belgium
Gert Matthijs, Coordinator of EuroGentest, Laboratory for Molecular Diagnosis, Center for Human Genetics, Belgium

Challenges of Central and Eastern European Healthcare Systems: Balancing the role of state and private sectors
Milan Macek, Professor, Charles University, Czech Republic

THEME 2 | KNOWLEDGE GENERATION AND DISSEMINATION

Ségolène Aymé, Director of Research, Director of International Affairs, ORPHANET-INSERM, France
Lesley Greene, Vice-President, CLIMB, Vice-Chair Committee for Orphan Medicinal Products (COMP), Volunteer Patient Advocate, EURORDIS, France

Without accessible, accurate information about rare diseases, diagnosis and appropriate care are delayed. Without raising awareness and improving education about rare diseases and their impact, clinicians do not have the tools to do their job effectively, patients and carers are unsupported and isolated and research and industry are starved of essential incentives and data to develop effective treatments. This theme covers all aspects of knowledge generation and dissemination, from coding and registries, to publicity through media.
The theme also examines the potential challenges and rewards from the perspective of all stakeholders.

Session 0201
Friday 9 May, 14:00 - 15:30

THE ROLE, RISKS AND RELEVANCE OF REGISTRIES IN SHAPING THERAPY DEVELOPMENT TO 2020

Session Chair:
Christine Lavery, Chief Executive, Society for Mucopolysaccharide Diseases, UK

Collecting data on diseases is a necessity at pre- and post-drug development stages to speed up clinical research, to provide data to regulatory and reimbursement bodies. Duplication of efforts should be avoided so as not to waste resources and expertise. Unified sources of data should be provided for diseases where several products are available, and advantage should be taken of technology to share data repositories, without ignoring the challenges raised by such an approach. Setting the scene – where we are today?

Landscape of Disease Registries in Europe and Challenges at Country Level
Elfriede Swinnen, Scientific Institute of Public Health, Belgium

The role and contribution of integrated registries as part of a holistic approach to rare disease treatments
Daniel Rosenberg, Senior Director, Head Epidemiology & Observational Studies, Actelion, Switzerland

Panel Discussion: Focus on Scientific and Procedural Hurdles for Therapy Development and Ongoing Matters in the Field of Orphans
Jeremy Manuel, OBE, Chair European Genetic Alliance, European Gaucher Alliance, UK
Carla Hollak, Professor of Inherited Metabolic Diseases in Adults, Academic Medical Centre, the Netherlands
Micheline Wille, Senior Director Medical Affairs, Shire, Switzerland
Leeza Osipenko, Head of Scientific Advice, Associate Director, NICE, UK

Session 0202
Friday 9 May, 16:30 - 18:00

A COLLABORATIVE MODEL TO PROGRESS KNOWLEDGE AND RESEARCH

Session Chair:
Stefan Schreck, Head of Unit, Health Information, DG SANCO, European Commission, EU

Rare disease patient registries and data collections need to be as internationally inter-operable as possible. The procedures to collect and exchange data need to be harmonised and consistent to allow pooling of data when necessary in order to reach sufficient statistically significant numbers for clinical research and public health purposes. Several countries have started to move in this direction and the European Commission supports a European platform that will be presented during this session.

Ispra Platform of Services for Rare Disease Registries
Ciarán Nicholl, Unit Head Public Health Policy Support, Institute for Health and Consumer Protection, Joint Research Centre, European Commission, Ispra (IT), EU

National Rare Disease Registries: Overview from France, Spain and Germany
Rémy Choquet, Project Manager, National Bank of Rare Diseases Data, Necker Hospital for Children, France
Manuel Posada, Director, Institute of Rare Diseases Research, Spain
Frank Ückert, Head of Medical Informatics, University of Mainz, Germany

Panel Discussion
Kay Parkinson, Chief Executive, Alström Syndrome, UK
Thomas Wagner, Pneumology/Allergology, Hospital of the Johann Wolfgang Goethe University, Germany

Session 0203
Saturday 10 May, 09:00 - 10:30

MAKING THE INVISIBLE VISIBLE: THE CODING OF RARE DISEASES IN HEALTH INFORMATION SYSTEMS
Interpretation DE + RU

Session Chair:
Ségolène Aymé, Emeritus Director of Research, Director of International Affairs, ORPHANET-INSERM, France

Much data is available in health information systems but data on rare diseases is not identifiable due to the lack of a proper coding system. The International Classification of Diseases has precise codes for only 240 rare diseases and the next edition, which should include all rare diseases, is not expected before 2017. Orphanet offers a coding system that can be implemented as a complement to the International Statistical Classification of Diseases and Related Health Problems (ICD10), the current version.
This would generate a lot of relevant information very quickly and at minimal cost. Experiences will be presented during this session.
The clinical spectrum of the diseases should also be recorded with a harmonised nomenclature to ensure the inter-operability of databases.

French and German Agencies' Experience of Coding Rare Diseases with Orpha Codes in Hospital
Stefanie Weber, Head of the Medical Classifications Unit, German Institute of Medical Documentation and Information (DIMDI), Germany.

Forecast for Having Rare Disease Codes in ICD11 and SNOMED-CT
Ana Rath, Managing Editor, Orphanet-Inserm, Rare Disease Platform, France

Harmonisation of Coding Systems to Describe Disease Expression and International Efforts
Peter Robinson, Professor of Medical Genomics, Charité, Universitätsmedizin Berlin, Germany

Session 0204
Saturday 10 May, 11:30 - 13:00

DELIVERING HELP AND SUPPORT IN A VIRTUAL WORLD: WHAT WILL WORK BEST?
Interpretation DE + RU

Session Chair:
Lesley Greene, Vice-President, CLIMB, Vice-Chair Committee for Orphan Medicinal Products (COMP), Volunteer Patient Advocate, EURORDIS, France

Technology has not only made vast amounts of previously elusive knowledge accessible to the population at large, it has also enabled those seeking greater information and support regarding rare diseases to approach help through multiple media. Is there still a place for the traditional phone helpline or are the opportunities offered through email, the internet and social network sites of greater value and importance? In this session we examine the challenges involved and which sources and deliveries best suit both patient and practitioner.

The Challenges for Helplines, the 116 Number and Why We Are Advocating It
Dorica Dan, President, Romania Prader Willi Association, Romanian National Alliance for Rare Diseases, Romania

The Importance of Helplines in National Plans
Monica Mazzucato, Rare Diseases Coordinating Centre – Veneto Region Rare Diseases Registry, Italy

Results of the Caller Profile Analysis
Georgi Iskrov, Institute of Rare Diseases, Bulgaria

Session 0205
Saturday 10 May, 14:00 - 15:30

KNOWLEDGE AT THE POINT OF CARE: GETTING THE FACTS JUST IN TIME OR JUST IN CASE

Session Chair:
Peter Farndon CBE, Director, National Health Service, National Genetics and Genomics Education Centre, UK

It is universally acknowledged that current and accurate information is the key to speedy diagnosis and appropriate care. How can this be managed at point of care, specifically in an emergency situation where the expert may not be involved at the initial assessment. This session examines different approaches to secure the best outcome for the patient involved.

Overview of RARE-Best Practices EU Project
Domenica Taruscio, Director, National Centre for Rare Diseases, Italy

Emergency Guidelines and Emergency Cards
– Practical experience for clinician and patient
Ana Rath, Managing Editor, Orphanet-Inserm, Rare Disease Platform, France

Saturday Night at a University Hospital
– How to handle a patient with a rare disease
Tino Münster, Project Manager, Orphan Anesthesia, Germany

Panel Discussion
Lesley Greene, Vice-President, CLIMB, Vice-Chair Committee for Orphan Medicinal Products (COMP), Volunteer Patient Advocate, EURORDIS, France

Session 0206
Saturday 10 May, 15:45 - 17:15

HYPE, HELP OR HARM? THE IMPACT OF MEDIA PROMOTION OF RARE DISEASES

Session Chair:
Denis Costello, Web Communications Senior Manager & RareConnect Leader, EURORDIS, Spain

This session will look at the opportunities and challenges that have emerged as a result of the growing awareness in society of the unmet medical needs faced by people living with a rare disease, the strengthened shared identity between rare disease patients through the work of national alliances and events such as Rare Disease Day, as well as the more challenging debate on the sustainability of care to rare disease patients and families. It will share perspectives from national alliances, national media and also put forward the merits of a European Year of Rare Diseases in 2019.

Benefits and pitfalls of mainstream media coverage of rare diseases
Rinke van den Brink, Dutch Television, the Netherlands

Why a European Year of Rare Diseases 2019? What it brings to the table
Avril Daly, Vice-President, EURORDIS, Chair, Genetic & Rare Disorders Organisation (GRDO), Chief Executive Officer, Fighting Blindness, Ireland

Communicating about rare diseases research to the general public
Alessia Daturi, Patient Organisation Liaison Officer, Telethon Italia, Italy

THEME 3 | RESEARCH FROM DISCOVERY TO PATIENTS

Kay Parkinson, Chief Executive, Alström Syndrome, UK
Serge Braun, Chief Scientific Officer, AFM (Association Française Contre Les Myopathies), France

Rare disease research is still too fragmented and compartmentalised. This leads to lack of integration, duplication of efforts, lack of critical mass, thinking in “silos” and waste of resources. It also hinders progress towards better diagnosis and therapy for rare disease patients despite many opportunities offered by new technological developments. To improve the situation and ensure
a rapid translation of discoveries into operational diagnostic and therapeutic tools, several initiatives have emerged at local, regional, national and international level which will be reported along with their outcomes during the sessions in this Theme.

Session 0301
Friday 9 May, 14:00 - 15:30

SHAPING RARE DISEASE RESEARCH POLICY
Interpretation DE + RU

Session Chair:
Ségolène Aymé, Director of Research, Director of International Affairs, ORPHANET-INSERM, France

At a European and international level, new funding opportunities have been launched and new initiatives have been taken to foster research and development in the field of rare diseases. A critical appraisal of whether or not they match the identified needs in the field will be discussed in this session.

EU Horizon 2020: Focus on Research
Irene Norstedt, Head of Unit, Personalised Medicine, Health Research Directorate, DG Research and Innovation, European Commission, EU

Addressing the needs of the rare disease research community: The E-Rare perspective
Daria Julkowska, e-Rare Coordinator, INSERM, Fondation Maladies Rares, France

International Rare Diseases Research Consortium (IRDiRC), State of the Art
Paul Lasko, Chair, International Rare Diseases Research Consortium (IRDiRC), Canada

Panel Discussion
Kay Parkinson, Chief Executive, Alström Syndrome, UK
Ralph Schuster, DLR Project Management Agency, Germany

Session 0302
Friday 9 May, 16:30 - 18:00

ADDRESSING THE GAPS IN RESEARCH AT INTERNATIONAL LEVEL TO IDENTIFY OPPORTUNITIES
Interpretation DE + RU

Session Chair:
Milan Macek, Professor, Charles University, Czech Republic

The International Rare Diseases Research Consortium has reviewed what should be the roadmap for the years to come to ensure an optimal use of research opportunities for rare diseases. Three areas will be presented in this session: the area of diagnostics, the area of therapy development and the area of infrastructures supporting research in general.

IRDiRC road map based on gaps and solutions focusing on three scientific committee areas:

Facilitating the Diagnosis of Most Rare Diseases by 2020: IRDiRC’s path forward
Kym Boycott, Investigator, the Children’s Hospital of Eastern Ontario (CHEO), Canada

Unlocking the Potential toward 200 New Rare Disease Therapies by 2020
Yann Le Cam, Chief Executive Officer, EURORDIS, France

Infrastructural requirements for Rare Disease Research within IRDiRC
Hanns Lochmüller, Chair of Experimental Myology, Institute of Genetic Medicine Newcastle University, UK

Panel Debate: Current progress and ways to utilise synergies between committee activities

Session 0303
Saturday 10 May, 09:00 - 10:30

INCENTIVES TO CREATE A FAVOURABLE ECO SYSTEM

Session Chair:
Serge Braun, Chief Scientific Officer, AFM (Association Française Contre Les Myopathies), France

Win-win situations are possible through partnerships and initiatives between industry, public institutions and charity organisations, which open a new model to develop advanced therapeutic medicinal products. Examples will be given of successful ecosystems that contributed to the marketing of innovative treatments of rare diseases. This includes successful private fund raising for translational research centres involving all stakeholders; local and transnational bioclusters, as well as new infrastructure models for drug development.

An Innovative Model for Early Stage Rare Disease Therapy Financing and Development
Erik Tambuyzer, Founding Member, Biopontis Alliance Rare Disease Foundation (BARD), Belgium

Care for Rare
Kym Boycott, Investigator, the Children’s Hospital of Eastern Ontario (CHEO), Canada

Public-Private Initiative to Generate Diagnostic and Therapeutic Solutions
Virginie Miath, Project and Investment Manager, Conectus, France

Session 0304
Saturday 10 May, 11:30 - 13:00

BREAKTHROUGHS IN SCIENCE

Session Chair:
Gertjan van Ommen, Department of Human Genetics Leiden University, Director, Centre for Medical Systems Biology (CMSB), Director BioBanking and Biomolecular Research Infrastructure (BBMRI-NL), the Netherlands

This session will cover a number of advances in rare disease therapy design and development including the status and advances in stem cell therapy, the use of animal models to assist in functional assessment of the role of exome variants in rare diseases and the latest progress and pitfalls in the development of exon skip therapy.

An Overview on the Status of Stem Cells in Therapy / Stem Cell Research Breakthroughs
Christine Mummery, Head of Department, Professor of Developmental Biology, Leiden University Medical Centre, the Netherlands

Use of Animal Models for Exome Prioritisation of Rare Disease Genes
Damian Smedley, Wellcome Trust Sanger Institute, UK

Promises and Status of Exon Skipping In Broad Sense
Gertjan van Ommen, Leiden University Medical Center, the Netherlands

Panel discussion
Serge Braun, Chief Scientific Officer, AFM (Association Française Contre Les Myopathies), France

THEME 4 | STATE OF THE ART AND INNOVATIVE PRACTICES IN ORPHAN PRODUCTS

Bruno Sepodes, Chair, Committee for Orphan Medicinal Products (COMP), Portugal
Emmanuelle Lecomte-Brisset, Quality Assurance, Regulatory Affairs, Head International Regulatory Strategy, Shire, Switzerland

Fourteen years have passed since the adoption of the European Regulation on Orphan Medicinal Products. Success in the stimulation of the research, development and bringing to the market of appropriate medications for orphan diseases has been achieved beyond expectation. However, work remains to continue to improve the legislative framework, to ensure patients have access to these treatments.

The aim of Theme 4 is to examine the latest initiatives and discuss innovative practices in orphan medicinal products at all stages of the development chain. It also addresses the main challenges being faced in accessing and securing the availability of rare disease treatments to patients.

The theme will include presentations and panel discussions, enriched with the experience and expertise of the main stakeholders shaping the orphan landscape today.

Session 0401
Friday 9 May, 14:00 - 15:30

CURRENT LANDSCAPE OF POLICY DEVELOPMENT ON ORPHAN PRODUCTS & RARE DISEASE THERAPIES

Session Chair:
Bruno Sepodes, Chair, Committee for Orphan Medicinal Products (COMP), Portugal

In this session, regulators will provide the latest status of current and emerging regulatory tools to support orphan therapies development and emerging regulatory tools to support orphan therapies development and approval processes.

Current Landscape – Overview by European Medicines Agency
Stiina Aarum, Acting Head of Orphan Medicines, European Medicines Agency, EU

State of Play from the US Perspective
Debra Lewis, Deputy Director of Office of Orphan Products Development, FDA, USA

Development of International Orphan Drug Policies
Emmanuelle Lecomte-Brisset, Quality Assurance, Regulatory Affairs, Head International Regulatory Strategy, Shire, Switzerland

Panel Discussion
Marlene Haffner, Chief Executive Officer, Haffner Associates, USA

Session 0402
Friday 9 May, 16:30 - 18:00

FACTS ON CURRENT PATIENT ACCESS CHALLENGES TO ORPHAN PRODUCTS

Session Chair:
Thomas Heynisch, Deputy Head of Unit, Unit Food & Healthcare Industries Biotechnology, DG Enterprise, European Commission, EU

Session 0305
Saturday 10 May, 14:00 - 15:30

PRE-COMPETITIVE TOOLS AND RESOURCES / PUBLIC-PRIVATE PARTNERSHIP IN THE AREA OF RARE DISEASES (INCLUDING INNOVATIVE MEDICINES INITIATIVE)

Session Chair:
Nathalie Seigneuret, Senior Scientific Project Manager, Innovative Medicines Initiative (IMI), Executive Office, EU

Despite the high quality of public research and the existence of centres and networks of excellence in rare disease research, there is a lack of innovative translational approaches for the development and marketing of new orphan medicinal products. Gaps can be filled by public-private collaborations including academics, patient organisations and biotech/pharmaceutical partners. Complementary expertise is increasingly merging with a common objective: catalysing innovation and efficiency. Three experiences will be presented in this session that will set the basis for interactive discussions about public-private partnership challenges and achievements.

Supporting the Drug Development Pathway for Rare Diseases – The experience of the-Neuromuscular Network (NMD) advisory committee for therapeutics (TACT)
Kate Bushby, Professor of Neuromuscular Genetics, MRC Centre for Neuromuscular Diseases, Institute of Genetic Medicine, International Centre for Life, Newcastle Upon Tyne Hospital, UK

European Bioinformatics Institute (EBI) Initiative
Justin Paschall, Team Leader, Variation, European Bioinformatics Institute, UK

The Experience of a Charity in Translating the Results of Basic Research to Therapies for Patients
Lucia Monaco, Chief Scientific Officer, Fondazione Telethon, Italy

Session 0306
Saturday 10 May, 15:45 - 17:15

WHOSE DATA IS IT? STIMULATING RESEARCH AND REMOVING BARRIERS

Session Chair:
Kay Parkinson, Chief Executive, Alström Syndrome, UK

This session will provide opportunities to hear how new ways of working and new technologies are helping to stimulate research for diseases that were often marginalised, whilst also highlighting some of the legal and ethical barriers that have to be overcome.

Stimulating Research and Monitoring Patients
Phil Beales, Professor of Medical Genetics, UCL, UK

The Responsible Use and Indication Criteria for Next Generation Sequencing Diagnostics in Clinical Practice
Hans Scheffer, Associate Professor Clinical Molecular Genetics, Radboud University Nijmegen Medical Centre, the Netherlands

The EU Data Protection Law Reform and Scientific Research: What's new?
Gauthier Chassang, Lawyer EU and International Law, INSERM, France

Challenges in orphan medicinal product access are increasing in times of austerity. Viewpoints and engagement of main stakeholders involved in the process at country and European level will be shared in this session.

Cost Containment Measures for Medicines in the European Economic Crisis
François Houyez, Treatment Information and Access Director, EURORDIS, France

Greece's Health Crisis: from austerity to denialism
Alexander Kentikelenis, Research Associate, Department of Sociology, University of Cambridge, UK

Panel Discussion: Viewpoint and engagement of different stakeholders involved
Paolo Siviero, Head of Economic Strategy and Pharmaceutical Policy, Italian Medicines Agency (AIFA), Italy
Heidi Wagner, Senior Vice President, Global Government Affairs, Alexion Pharmaceuticals, USA

Session 0403
Saturday 10 May, 09:00 - 10:30

EUROPEAN MEDICINES AGENCY (EMA) & HEALTH TECHNOLOGIES ASSESSMENT (HTA) INTERFACING ON RARE DISEASE THERAPIES
Interpretation DE + RU

Session Chair:
Spiros Vamvakas, Head of Scientific Advice, Human Medicines Special Areas, European Medicines Agency, EU

Growing interfacing and early dialogue between the European Medicines Agency (EMA) and Health Technology Assessment (HTA) is an opportunity for developers to receive simultaneous feedback from both sides on their development plans and reduce the time of orphan therapy availability to patients.
During this session, available regulatory options, practical experiences gained so far on parallel protocol assistance with HTA bodies, as well as procedural trends for the future will be presented.

Experience to date on interfacing in rare diseases, status with protocol assistance and where are we going – future directions
Spiros Vamvakas, Head of Scientific Advice, Human Medicines Special Areas, European Medicines Agency, EU

Experience of the Parallel European Medicines Agency (EMA) Health Technology Assessment-(HTA) Scientific Advice (SA) from an Orphan Disease Point of View
Samuel Rigourd, Global Program Regulatory Director, Novartis Pharma, Switzerland

Panel Discussion: Scientific and procedural hurdles for orphans
Leeza Osipenko, Head of Scientific Advice, Associate Director, NICE, UK

Session 0404
Saturday 10 May, 11:30 - 13:00

SHORTAGES IN AUTHORISED MEDICINES FOR RARE DISEASES

Session Chair:
Jeremy Manuel, OBE, Chairman European Gaucher Alliance, UK

There is a growing challenge of shortages in authorised medicines for rare diseases and this session will provide the opportunity to share viewpoints from patients, regulators and industry on how best to secure rare disease treatments.

Experience with Fabry Shortage
Carla Hollak, Professor of Inherited Metabolic Diseases in Adults, Academic Medical Center, the Netherlands

Regulatory Perspective
Brendan Cuddy, Scientific Administrator, European Medicines Agency, EU

Genzyme, a Sanofi Company – How a company should/could respond
Carlo Incerti, Senior Vice President, Head Genzyme Global Medical Affairs, Italy

Panel Discussion
François Houyez, Treatment Information and Access Director, EURORDIS, France
Paolo Siviero, Head of Economic Strategy and Pharmaceutical Policy, Italian Medicines Agency (AIFA), Italy
Heidi Wagner, Senior Vice President, Global Government Affairs, Alexion Pharmaceuticals, USA

Session 0405
Saturday 10 May, 14:00 - 15:30

UNDERSTANDING OF ORPHAN THERAPIES OFF-LABEL USES AND THEIR NEW CHALLENGES

Session Chair:
André Lhoir, Member of Committee for Orphan Medicinal Products (COMP), Belgium

It is recognised that there is a current trend to reduce access to off-label drugs at national levels. This session is devoted to presenting the views of different stakeholders on how to secure the off-label use and how in this context, data collection could benefit the healthcare system.

Challenges Associated with Healthcare System: Reimbursement
Yves Juillet, Secretary General, Academy of Medicine Foundation, France

Off-label Use: Good and bad practices
Marc Dooms, Senior Orphan Drug Specialist, University Hospitals Leuven, Belgium

National Institute of Health (NIH) Guidelines that Include Offlabel Use
Greet Musch, Director General DG PRE – authorisation, Federal Agency for Medicinal and Health Products, Belgium

Panel Discussion: Support Systems to Get Off-Label Use and Evidence Generating

Session 0406
Saturday 10 May, 15:45 - 17:15

EMPOWERING PATIENT ADVOCATES IN DRUG DEVELOPMENT
Interpretation DE + RU

Session Chair:
Nick Sireau, Chairman, AKU Society, UK

Patients and advocacy groups provide a great deal of value in drug development, particularly in rare diseases. This session will develop ideas on how best to empower them.

Education & Training Initiatives in Drug Development for Patients
Maria Mavris, Director Therapeutic Development, EURORDIS, France

Understanding and Using Health Technology Assessment to Make a Case for Better Patient Care
Elena Nicod, Research Officer, LSE Health and Social Policy, London School of Economics, UK

A "How-to" Guide to Help Patient Groups Drive the Drug Development Process
Tony Hall, Co-Founder, Findacure, UK

THEME 5 | EMERGING CONCEPTS AND FUTURE POLICIES FOR RARE DISEASE THERAPIES

Jordi Llinares Garcia, Head, Product Development Scientific Support Department, European Medicines Agency, EU
Paolo Siviero, Head of Economic Strategy and Pharmaceutical Policy, Italian Medicines Agency (AIFA), Italy

Complementing Theme 4, Theme 5 will look to the future at what practical policies are being developed to facilitate access to treatments for rare diseases.

Participants will hear the perspectives of a range of stakeholders, including patient representatives, regulators, HTA bodies, payers and industry. The theme will include exchanges of views on how early dialogue is working in practice and how we can ensure a continuous dialogue throughout the life-cycle of a drug; what level of cooperation between the EMA and the FDA is possible or desirable to speed up drug development; how we can ensure that the voice of the patient is heard throughout all aspects of the processes; whether we are making the best use of the current regulatory framework; and whether patient access can be improved through increased collaboration at all stages.

To conclude, the theme will examine the external influencing factors that can either support or threaten the continued availability of rare disease treatments and what the future holds in terms of potential alternatives in the development and availability of therapeutic interventions.

Session 0501
Friday 9 May, 14:00 - 15:30

EARLY DIALOGUE AND HORIZON SCANNING OF PRODUCT DEVELOPMENT TO ADDRESS UNMET MEDICAL NEEDS

Session Chair:
Paolo Siviero, Chair, MEDEV and Head of Economic Strategy & Pharmaceutical Policy, AIFA, Italy

Early and continuous dialogue: what is it; what does it mean; where can it take place; who should be involved; why do we need it; when do we need it ?

Panel Discussion
Jordi Llinares Garcia, Head, Product Development Scientific Support Department, European Medicines Agency, EU
François Meyer, Advisor to HAS' President, International Affairs, French National Authority for Health, Haute Autorité de Santé (HAS), France
Ad Schuurman, Head of the Business Contact Centre and International Affairs of the National Healthcare Institute, the Netherlands
Jan Geissler, European Patients' Academy on Therapeutic Innovation (EUPATI), Germany
Christine Mayer-Nicolai, Merck KgaA, Germany

Session 0502
Friday 9 May, 16:30 - 18:00

HOW TO SHAPE A BETTER FRAMEWORK FOR ORPHAN DRUG DEVELOPMENT: EMA/FDA COLLABORATION
Interpretation DE + RU

Session Chairs:
Jordi Llinares Garcia, Head, Product Development Scientific Support Department, European Medicines Agency, EU
Debra Lewis, Deputy Director of Office of Orphan Products Development, FDA, USA

Improving the effectiveness of collaboration efforts within the rare disease community is key to addressing rare disease therapy development worldwide. This session will describe options, perspectives, and case studies to address the collaborative regulatory and research efforts with the EU and USA to advance drug development for rare diseases.

Common or Coordinated Guidelines – Why do we need collaboration and how could it be achieved?
Patricia Hurter, Senior Vice President, Global Pharmaceutical Development, Vertex, USA

How Trans-Atlantic Collaboration can Speed up Efficient Drug Development: The Cystic Fibrosis (CF) Clinical Trial Networks
Kris De Boeck, Paediatric Pulmonology, University Gasthuisberg, Belgium

Case Study: Duchene Muscular Dystrophy
The European Perspective: **Elizabeth Vroom**, Parent Project, the Netherlands
The US Perspective: **Pat Furlong**, Parent Project Muscular Dystrophy, USA

Panel Discussion
Spiros Vamvakas, Head of Scientific Advice, Human Medicines Special Areas, European Medicines Agency, EU

Session 0503
Saturday 10 May, 09:00 - 10:30

PROGRESSIVE PATIENT ACCESS SCHEMES & PATIENT INVOLVEMENT IN BENEFIT-RISK ASSESSMENT

Session Chair:
Jaroslaw Waligora, Policy Officer, Directorate-General for Health and Consumers, European Commission, EU

Are we making the most of the regulatory and other tools in the European framework to secure timely patient access to needed rare disease treatments? Or are we asking rare disease patients to take more risks than is reasonable? How can we best balance the needs and the responsibilities of all of the actors to secure the best healthcare outcomes?

Are we ready? What is missing and what is needed? A regulator's perspective
Luca Pani, Director General, Italian Medicines Agency (AIFA), Italy

Patient View on Progressive Patient Access Schemes
Pauline Evers, EGAN (European genetic alliances network / Dutch Federation of Cancer Patient Organisations), the Netherlands

Panel Discussion
Mark Rothera, Chief Commercial Officer, PTC Therapeutics, USA
Yann Le Cam, Chief Executive Officer, EURORDIS, France

Session 0504
Saturday 10 May, 11:30 - 13:00

MECHANISM OF COORDINATED ACCESS (MOCA) AND TRANSPARENT VALUE FRAMEWORK, MARKET ENTRY AGREEMENTS

Session Chair:
Thomas Heynisch, Deputy Head of Unit, Unit Food & Healthcare Industries Biotechnology, DG Enterprise, European Commission, EU

The process and concept: a promise of smoother and timelier patient's access based on collaborative and cooperative work. Where are we and will the promise be delivered?

Concepts & MOCA Pilots (feedback from the process around the first pilots)
Wills Hughes-Wilson, Chief Patient Access Officer & Vice President External Affairs, Sobi, Sweden

Managed Entry Agreements
Luca Pani, Director General, Italian Medicines Agency (AIFA), Italy

Panel Discussion
Yann Le Cam, Chief Executive Officer, EURORDIS, France
Ri de Ridder, Director General, RIZIV-INAMI, Belgium

Session 0505
Saturday 10 May, 14:00 - 15:30

EMERGING IDEAS FOR SUSTAINABLE ACCESS TO ORPHAN MEDICINAL PRODUCTS
Interpretation DE + RU

Session Chair:
Kerstin Westermark, Senior Expert, Committee for Orphan Medicinal Products (COMP Member), Medical Products Agency, Sweden.

What are the environmental threats to the continued sustainable availability of treatment for rare diseases and how can we navigate them ?

Differential Pricing – A way to demonstrate social solidarity to alleviate the impact of the financial crisis
Flaminia Macchia, Director European Public Affairs, EURORDIS, Belgium

Licensing, Orphan Status and Reimbursement - Can we harmonize the evidence required?
Brigitte Bloechl-Daum, Associate Professor and Deputy Head of the Department of Clinical Pharmacology at the Medical University of Vienna; Austrian Delegate to the COMP , Vice-Chair of the Committee on Reimbursement of Drugs of the Austrian Social Securities Association, Austria

Panel Discussion
Adam Heathfield, Senior Director, Worldwide Policy, Pfizer, UK
Rembert Elbers, Member of the Committee of Orphan Medicinal Products (COMP), Member of the Scientific Advice Working Party, Former Head of Federal Institute for Drugs and Medical Devices (BfArM) Oncology Unit, Germany

Session 0506
Saturday 10 May, 15:45 - 17:15

RARE DISEASE TREATMENTS BEYOND MEDICINAL PRODUCTS

Session Chair:
Lesley Greene, Vice-President, CLIMB, Vice-Chair Committee for Orphan Medicinal Products (COMP), Volunteer Patient Advocate, EURORDIS, France

Advances in technology, design and computer programming all support more effective and targeted medical devices to deliver diagnostics and treatments for patients with rare diseases. These medical devices can play a vital role in optimising health and quality of life, by enabling quicker and more accurate diagnosis. Some medical devices can also allow home treatment rather than hospitalisation and the ability to travel for pleasure or work which would have been impossible with older systems. Other medical devices are designed for more targeted, more effective or more convenient delivery of a therapy. This session examines these issues from the point of view of the different stakeholders.

Round table discussion
Radoslaw Kaczmarek, Member of the EHC Steering Committee, European Haemophilia Consortium (EHC), Belgium
Peter Rutherford, Medical Director - Europe, Middle East and Africa, Baxter Healthcare, Switzerland
John Wilkinson OBE, Director of Devices, Medicines and Healthcare Products Regulatory Agency (MHRA), UK
Kerstin Westermark, Senior Expert, Committee for Orphan Medicinal Products (COMP Member), Medical Products Agency, Sweden.
Smail Hadj-Rabia, Department of Dermatology, Hôpital Necker - Enfants Malades, France

THEME 6 | BEYOND MEDICAL CARE

John Dart, Chief Operating Officer, DEBRA International, UK
Gabor Pogany, President Rare Diseases Hungary, Hungary

Whilst high quality, accessible clinical care is essential for people with rare diseases, we are far more than just patients. This Theme aims to explore social and other non-medical issues that impact on quality on life and access to full citizenship, to identify innovative solutions and programmes that address these issues and to make the case for embedding best practice in these areas into European and national policies and provisions.
The approach will be participative, with platform speakers and delegates sharing knowledge, experience and aspirations, feeding into EURORDIS' future strategy on specialised social services. The need for National Plans and Centres of Expertise to recognise and include non-medical care will be emphasised and the guiding principles and experience gained so far evaluated.
Innovative schemes that may serve as models for promoting independent living will be described, together with personal experiences of service users.

Session 0601
Friday 9 May, 14:00 - 15:30

IDENTIFYING SPECIFIC SOCIAL CHALLENGES OF RARE DISEASES

Session Chair:
Stein Are Aksnes, Leader of Norwegian Advisory Unit on Rare Disorders, Oslo University Hospital, Norway

This session will present the importance of coordination, interdisciplinary and individual care as well as demonstrate how National Plans and legislation can be a powerful national advocacy tool that
can lead to change and improvement in patients' holistic care.

Current Challenges and Issues
Dorica Dan, President, Romania Prader Willi Association, Romanian National Alliance for Rare Diseases, Romania

How National Plans can Address These Issues
Simona Bellagambi, UNIAMO, Italy

Case Study: The experience of France
Christel Nourissier, Alliance Maladies Rares, France

Session 0602
Friday 9 May, 16:30 - 18:00

DIFFERENT APPROACHES TO THE SOCIAL CHALLENGES OF RARE DISEASES: SOCIAL POLICY

Session Chair:
Helena Kääriäinen, Research Professor, National Institute for Health and Welfare, Finland

Social services face challenges when dealing with individuals with rare diseases. Depending on the type of the service and the life situation of the individual, adaptation of the available services is often needed. For that, information on the rare disease, specifically tailored to the needs of social services, is needed, including experiences from the individuals who have
provided or received such services. This session gives examples of different approaches to these challenges.

Orphanet Disability Projects
Myriam de Chalendar, Orphanet, France

Social Profiles Project by Rare Diseases Denmark
Birthe Holm, Rare Diseases Denmark, Denmark

Online Platform for Patients to Share Innovative Solutions/ Discoveries
Pedro Oliveira, University of Lisbon, Portugal

Session 0603
Saturday 10 May, 09:00 - 10:30

CONCRETE SOLUTIONS TO SOCIAL CHALLENGES: ESSENTIAL TOOLS FOR THE INTEGRATION OF RARE DISEASES INTO SOCIAL SERVICES

Session Chair:
Lene Jensen, Chief Executive Officer, Rare Diseases Denmark, Denmark

Even though systems for social support differ among the European countries, rare disease patients' needs for specialised social services are similar. This workshop presents the results of EUCERD workshops on principles for specialised social services and for training of the providers. Key actors will share their experience of working with specialised social services in practice. The participants in this session are invited to not only learn from this experience but also share their own experiences. The contribution of each participant will be valued and an active network among patients, families, social and healthcare professionals is encouraged.

Guiding Principles for Specialised Social Services and Guidelines for the Training of Social Service Providers
Dorica Dan, President, Romania Prader Willi Association, Romanian National Alliance for Rare Diseases, Romania

Panel Discussion
Terry Dignan, Serious Fun Network, Ireland
Norbert Hödebeck-Stuntebeck, Charitable Foundation for People with Disabilities, Prader-Willi Syndrome, Germany
Lisen Julie Mohr, Frambu, Norway
Anders Olauson, Ågrenska, Sweden

Session 0604
Saturday 10 May, 11:30 - 13:00

CAN PEOPLE LIVING WITH A RARE DISEASE BE INDEPENDENT? INSPIRING PERSONAL STORIES

Session Chair:
Gabor Pogany, President Rare Diseases Hungary, Hungary

People living with rare diseases often find themselves struggling with common daily life tasks, due to the fact that structures and society at large are not always prepared and adapted to deal with the different kinds of disabilities resulting from complex diseases. This session gives voice to some inspiring personal stories from people who are the living proof that it is possible to be more autonomous with some creative and innovative solutions.

The Balance between Needing Care and Living Independently for a Young Man with a Degenerative Rare Disease
Hanka Meutgeert, Mother of a young adult, living with a degenerative rare disease, the Netherlands

My Story
Peter Ryan, Fighting Blindness, Dublin, Ireland

Let to Fly – Independent life camps for rare disease youths with intellectual disabilities
Beata Boncz, Hungarian Williams Syndrome Association, Hungary
Krisztina Pogany, Living with Williams Syndrome, Hungary

Session 0605
Saturday 10 May, 14:00 - 15:30

CAN PEOPLE LIVING WITH A RARE DISEASE BE INDEPENDENT? INSPIRING SOLUTIONS BY PROVIDERS
Interpretation DE + RU

Session Chair:
Christoph Nachtigäller, President, ACHSE (German National Alliance for Chronic Rare Diseases), Germany

This session will present some solutions developed by patient organisations in order to provide support to daily life challenges of people living with a rare disease, helping them to achieve a higher level of autonomy.

Online Psychological Support for people living with a rare disease
Alba Ancochea, The Spanish Federation of Rare Diseases (FEDER), Spain

Ristoro Fantasia – Creating employment for people living with a rare disease
Renza Barbon Galluppi, President of Italian Federation of Rare Diseases (UNIAMO), Italy

Empowerment Weekends for Young Adults with Anorectal Malformations
Annette Lemli, Vice-President SoMA (Patient Organisation for People with Anorectal Malformations), Germany

Panel Discussion
Denis Costello, Web Communications Senior Manager & RareConnect Leader, EURORDIS, Spain

Session 0606
Saturday 10 May, 15:45 - 17:15

HOW CENTRES OF EXPERTISE SHOULD/COULD INTERFACE WITH SOCIAL SERVICES
Interpretation DE + RU

Session Chair:
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National Centres of Expertise are key to the delivery of high quality services, especially in the field of rare diseases. Integration of social care into the package of support available is essential. In this session we will explore some of the challenges and opportunities in doing this, including practical examples of how various countries are approaching the provision of seamless care.

The Proposed Role of Centres of Expertise Based on European Union Committee of Experts on Rare Diseases (EUCERD) Recommendations
Kate Bushby, Professor of Neuromuscular Genetics, MRC Centre for Neuromuscular Diseases, Institute of Genetic Medicine, International Centre for Life, Newcastle Upon Tyne Hospital, UK

Case Study: The experience of France
Christel Nourissier, Alliance Maladies Rares, France

Panel Discussion and Lively Debate
Francesc Palau, CIBERER, Spain
Annette Grüters-Kieslich, Medical Director of the Charité Center for Women and Child Health and Human Genetics, Germany

SUMMARY OF OPENING SESSION

The 7th European Conference on Rare Diseases and Orphan Products (ECRD 2014 Berlin) was opened by **EURORDIS Vice President, Avril DALY,** who pointed out how significantly the event has grown since the first ECRD was held in 2001 in Copenhagen. This time around, the event gathered over 750 participants representing all stakeholders, demonstrating the growing interest in rare diseases (RD).

The planning and the content of the Conference involved a collaboration between EURORDIS with the co-organiser, DIA Europe (Drug Information Association), and partnerships with the European Commission, the European Medicines Agency (EMA), the National Organization for Rare Disorders (NORD), the Canadian Organization for Rare Disorders (CORD), the German National Alliance for Chronic Rare Diseases (ACHSE), ORPHANET, the European Society for Human Genetics (ESHG) and industry through the EBE-EuropaBio, as well as members of the Programme Committee and supporters.

Ms Daly reminded the audience that the ECRD provides a multi-national and multi-stakeholder policy forum through which participants can learn from each other, exchange and network, and contribute to shaping the direction of future policy actions. Delegates are in attendance from all over Europe as well as the USA, Canada, South America, Asia, Africa, Australia and New Zealand, in recognition of the global necessity to develop a policy framework addressing rare diseases and the need to work together to tackle collective concerns.

Sharing a vision and a common goal for the betterment of society has proven to lead to success. Ms Daly explained that while one focus of the event is to ensure access to therapies in a timely fashion, not everybody affected by a rare disease will have a treatment in their lifetime; hence, ECRD 2014 Berlin will equally focus on developing a more comprehensive approach to care and services that will improve quality of life.

Since the last ECRD, many National Plans for Rare Diseases have been adopted or are on the verge of adoption. The EU Committee of Experts on Rare Diseases (EUCERD), established in 2009 and active until July 2013, aided the European Commission to develop and implement the Community activities in the field of rare diseases. The seven patient representatives to the EUCERD advocated within the Committee on behalf of the rare disease community. The EUCERD adopted Recommendations and one opinion aimed to give guidance on rare disease policies across EU countries. They included recommendations on: Quality Criteria for Centres of Expertise for Rare Diseases in Member States; Improving Informed Decisions based on the Clinical Added Value of Orphan Medicinal Products (CAVOMP) Information Flow; European Reference Networks for Rare Diseases; Rare Disease Patient Registration and Data Collection; Core Indicators for Rare Disease National Plans/Strategies, and Opinion on Potential Areas for European Collaboration in the Field of Newborn Screening.

Within the EUCERD Joint Action, EURORDIS continues to support the development of National Plans (Work Package 4, "EUROPLAN") by supporting and coordinating the organisation of 20 National Conferences in the EU and in 4 European countries outside the EU.

Advancement in research on rare diseases is another strategic pillar of EURORDIS, and the adoption of the new EU Research and Innovation Framework Programme, Horizon 2020, was a welcome development. EURORDIS is active within the International Rare Diseases Research Consortium (IRDiRC) where it has strengthened its presence in the governing bodies and meetings. IRDiRC teams up investors in rare disease research in order to deliver 200 new therapies for rare diseases and means to diagnose most rare diseases by the year 2020. Furthermore, as a partner of consortia and projects in the field, EURORDIS supports integrated data repositories, biobanks and data infrastructures that reflect patients' best interests by involving patients both at the governance and clinical research level.

The regulatory process is essential to bring therapies to patients: EURORDIS holds a unique position with the presence of its volunteer patient representatives in the EMA Committees and Working Parties. This year alone, EURORDIS patient representatives contributed to the examination of 413 dossiers as part of assignments of the Committee they belong to. Additionally, 16 patient representatives were invited by the EMA Scientific Advice Working Party to provide first-hand experience on relevant outcome measures and endpoints in clinical trials. EURORDIS patient representatives are supported by the Therapeutic Action Group (TAG).

Annette Widmann-Mauz, Parliamentary State Secretary, Federal Ministry of Health, Germany

The Drug Information, Transparency and Access task force (DITA) is a group of volunteers trained through the EURORDIS Summer School, advising patient representatives in EMA Committees and Health Technology Assessment (HTA) bodies. Indeed, the Summer School that EURORDIS has held each year since 2008 is a well-established training course and a core component of its capacity-building activities. In addition, EURORDIS has expanded its collection of online e-learning tools while patients' involvement has continued in the DIA Patient Fellowship Programme, EUPATI (European Patients' Academy on Therapeutic Innovation) and ECRIN-IA (European Clinical Research Infrastructure Network – Integrating Activity).

EURORDIS believes strongly in a comprehensive approach to the delivery of care and essential services for those affected by rare diseases, their families and carers, and has established a broad patient movement across Europe with more than 600 member organisations in over 55 countries, creating a strong, unified voice of rare disease patients central to policy development.

Ms Daly recalled that the European regulatory and policy framework is now well developed and, while continuing to maintain it, focus must shift now to the implementation of national plans and strategies at the national/local levels. EURORDIS is preparing for this next phase by strengthening its movement and aligning National Alliances and EURORDIS on Common Goals and Mutual Commitments, and by involving isolated patients through the moderated online patient forum RareConnect. Other initiatives include the campaign for the European Year for Rare Diseases in 2019, aimed to increase awareness and political support; international groups, such as NORD, CORD, IPOPI; preparing a long-term strategy on European Reference Networks, Centres of Expertise and Healthcare Pathways, as well as patient mobility across the EU and their links to integrated research infrastructures; and always being mindful of potentially emerging ethical and societal challenges that cannot be ignored.

Ms Daly concluded her introduction by inviting the Conference delegates to reconvene in 2016 for the next ECRD, which will be held in Edinburgh, Scotland, at which time the results of this implementation process will be observed.

Jytte Lyngvig, Director for DIA Europe (Drug Information Association), welcomed the audience by evoking the slogan of ECRD 2014 Berlin: "The Rare Disease Puzzle: Bringing the Picture to Life" and the challenge of how to combine the pieces into an overall picture. Quoting Antoine Saint-Exupéry's book The Little Prince at different times, Ms Lyngvig reminded the audience that they were gathered to work together to forward actions with clarity and energy into a coherent and sustainable strategic plan.

Identifying and understanding uniqueness in the mass is the secret for driving development forward in an area labelled "rare". Ms Lyngvig closed her speech with a reminder for the conference participants: "The most beautiful things in the world are felt with the heart".

Christoph Nachtigäller, President of ACHSE (Allianz Chronischer Seltener Erkrankungen), the German National Alliance, partner of ECRD 2014 Berlin, recalled the progress made in the field of rare diseases since 2006, when the EURORDIS Membership Meeting last took place in Berlin. He highlighted the growing number of participants and the increasing awareness of rare diseases amongst stakeholders.

Mr Nachtigäller described the role that ACHSE has played since 2004 in Germany advocating for special needs and the recognition of expert knowledge of people living with rare diseases, as well as in pursuing patient empowerment from all angles: participation, legal equality and equal opportunities. These activities have won the organisation the official status of stakeholder in the German healthcare sector, as well as recognition in the rare disease movement, including the EURORDIS Patient Organisation Award in 2014. ACHSE is a very lively network of 120 members, representing approximately 4 million patients.

The German Alliance played a crucial role in the process that led to the adoption of the German National Plan of Action for Rare Diseases, adopted in 2013 after three years of concerted effort with all stakeholders working together in the National Action League for people with rare diseases (NAMSE). Mr Nachtigäller praised the patient-centred approach that characterised the work of the NAMSE members and recalled the complex issues addressed: access to qualified and clear information, shortening the path to diagnosis, identification and mapping of rare disease competence, appropriate diagnostics and consideration for patients without diagnosis, and research.

Areas still to be addressed for ACHSE include how to make information barrier-free; equal opportunities in accessing therapies including orphan drugs and off label medicines; identification of Centres of Expertise and related quality standards; improving patient quality of life through research.

Mr Nachtigäller evoked the essential support and stimulus from the EU where policy initiatives have spurred successful national processes, as well as the collaboration with EURORDIS and its members to learn from each other, share information and best practices, devise solutions and carry on joint efforts.

The Parliamentary State Secretary to the Federal Minister of Health, Germany, Annette Widmann-Mauz, welcomed the delegates on behalf of the Minister, acknowledging the ECRD as the most important European discussion forum for rare diseases and commented that Germany is proud to host the event. Ms Widmann-Mauz recalled the paradox of rare diseases: while only a few people suffer from each disease, altogether 4 million people in Germany alone are affected and possibly 36 million in Europe. Ms Widmann-Mauz stressed the need to give special attention to people living with a rare disease because for too long they have been neglected by public concern. People living with a rare disease not only suffer from their disease but also from the cost of treatments, difficult access to care and other disadvantages.

She recalled the role of the EU Council Recommendation on an Action in the Field of Rare Diseases adopted in 2009, and the important consultation process carried out for a national plan in Germany with the almost 30 partners in NAMSE. Over 50 proposals were agreed upon that aim to improve the life of people living with a rare disease and numerous projects of those identified in the plan will receive public funding, including those contributing to the standardisation of rare disease codification and to setting up of the JRC European platform on registries.

It is important, added the Parliamentary State Secretary to the Federal Minister of Health, that international and national specialists work together in order to decipher the puzzle that rare diseases present. The German National Plan will not only collect information at the national level, but also will contribute to cross border knowledge through its national Centres of Expertise that will form the basis of the European Reference Networks.

The Head of Unit for Personalised Medicine at the European Commission, DG Research and Innovation, Irene Norstedt, began by stressing that the leitmotiv for the work on rare diseases is collaboration. Rare diseases are a challenge far too big to master alone. Unmet needs are still huge, and there is much more to be done, not only in Europe. Collaboration is key, as resources in the field are scarce and scattered.

Ms Norstedt summarised the increase in funding that rare disease research received from the European Commission: from 64 million EUR and 47 projects funded in its 5th Research Framework Programme (FP5), the Commission invested 620 million EUR and supported close to 120 collaborative projects in its 7th Framework Programme (FP7) that ended in 2013. This adds to more than 100 individual fellowships, grants and training networks.

EC-funded research for rare diseases range from natural history and pathophysiology, to in vitro/in vivo models, registries & bio-banks, identification of biomarkers, clinical trial methodologies for small populations, -omics for rare diseases and linking data, as well as development of preventive, diagnostic and therapeutic interventions.

Ms Norstedt mentioned some exemplary projects, such as:

- ALPHA-MAN that received funds over three Research Framework Programmes, from FP5 to FP7, having progressively advanced research over time from biochemical characterisation of mutations in the alpha-mannosidase gene (MANB) to clinical trials in patients
- EuroGentest started in FP6 and then under FP7 contributed to the harmonisation, validation and standardisation in genetic testing, helping bring genetic testing to healthcare
- ENRAH set up a European Network for research on Alternating Hemiplegia in Childhood and was a good example of patients taking the lead. As a result of the project, de novo mutations were identified as the primary cause of AHC, also offering insight into disease pathophysiology
- E-RARE is an example of networking activity pulling together resources and funders from EU Member States and beyond

Ms Norstedt then presented the achievements of the International Rare Disease Research Consortium (IRDiRC), an international collaborative initiative launched by the European Commission and the NIH to stimulate, coordinate and maximise outputs of rare disease research efforts around the world. IRDiRC was created in 2011 with the ambitious goals of delivering 200 new therapies for rare diseases and the means to diagnose most rare diseases by 2020. Collaboration is crucial to achieve these objectives: to date IRDiRC counts almost 40 partners from four continents and a variety of actors – regulators, patients, industries, agencies, charities.

Ms Norstedt added that the European Commission continues its commitment to research under the new Research Framework Programme, HORIZON 2020, which sets aside over 7 billion EUR to health research only. The new Framework Programme will continue to support rare disease research and the actions necessary to meet the commitments made in the framework of the IRDiRC. She closed her intervention by emphasising that the European Commission wants therapies to come to patients and, to this end, the integration of patients in research is vital.

Lesley Greene, Vice-Chair of the EMA Committee for Orphan Medicinal Products (COMP), Vice-President, CLIMB, Volunteer Patient Advocate EURORDIS, UK.
Speaking on behalf of the rare disease patient communities, Ms Greene recalled how her journey in the rare disease movement started in 1980 when her daughter was diagnosed with cystinosis. Significant progress has been made since that time in many areas: healthcare, research diagnosis, therapy, palliative and social care, regulatory affairs and policy. Yet all the pieces still need to be brought together, which explains the Theme of ECRD 2014 Berlin: The Rare Disease Puzzle - Bringing the Picture to Life. ECRD 2014 Berlin aims to present all the pieces of that puzzle in six related themes and build on the active contributions of all participants in putting those pieces together into a coherent whole.

The 800 stakeholders attending ECRD 2014 Berlin also represent the individual pieces of a puzzle, with each participant looking for a solution to the same jigsaw. Patients and families are forced into this search: the day they receive the news that they or someone they love is affected by a rare disease, their life changes and from that day forward they spend vast amounts of time and energy looking for information, targeted research, care, treatments and cure. Other stakeholders, from researchers to industry, are driven both by inquisitiveness and the desire to relieve suffering to try and transform the untreatable into the manageable and curable.
Ms Greene welcomed and thanked the pool of speakers and panelists delivering the themes of the conference, sharing their expertise to help unlock the 'mystery box' where the treasured solutions are contained. Yet the Conference relies on the participation of all present to complete the puzzle and secure a better future for patients, families, and ultimately for all those committed to the rare disease community in whatever capacity.
Ms Greene also took a moment to remind the audience of how frequently chronic and untreatable rare diseases and the related long-term care they require can lead to permanent splits in families, whose pieces cannot be brought together anymore. There is often a sense of isolation following a diagnosis of a rare disease, but ECRD 2014 Berlin is about joining together, sharing, and collaborating – and with the presence and participation of everybody at the conference, bringing the rare disease picture to life.

SUMMARY OF PLENARY SESSION

The Plenary Session and roundtable discussion were chaired by **Durhane Wong-Rieger, President of the Canadian Organization for Rare Disorders (CORD),** who welcomed participants to the ECRD 2014 Berlin, remarking that it offers a tremendous opportunity to take stock of what the EU has accomplished in the field of rare diseases and orphan medicines. While the USA brought "orphan drugs" onto the horizon, the EU made rare diseases a reality and this conference is a testimony of how far Europe has come in a short period of time. Not long ago, stakeholders in the field were invited to a consultation on the challenges ahead, the need for national plans and strategies and the value of Centres of Expertise. Today EU programmes and discussions are focussed on the modelling of these centres.

It is exciting to have the opportunity not only to discuss what has been happening, but also to look at the challenges that still lie ahead, including the extent to which these challenges are outside Europe and how to address them on an 'international' scale. Orphanet is a huge testament to this. Around the world progress is being made. Canada is going to have a national plan for rare diseases this year and a policy dedicated to orphan medicines. An Asian-Pacific Alliance for rare diseases is taking shape and developing and emerging countries also benefit from this huge movement in support of rare diseases.

John Ryan, Director for Public Health at the European Commission, DG Health and Consumers (DG SANCO), evoked the Commission Communication of 2008 and the Council Recommendation that followed the year after. Pulling limited resources, the European Commission focused on the recognition and visibility of rare diseases with the aim of supporting policies in Member States and coordination among them. This led to the adoption of a "soft law" on the definition of rare diseases, codification, European Reference Networks, gathering expertise at the EU level, empowerment of patient organisations and sustainability. The European Commission (EC) is publishing a report on the implementation of this Recommendation in the Member States.

The number of people affected by rare diseases in the EU is estimated between 27 and 36 million; rare diseases are a public health priority as an area of high EU added-value: pulling resources in public health projects supported by the European Commission so far showed the added value of working together.

The Council recommended the adoption of national plans and strategies, preferably before the end of 2013, to help overcome the lack of initiatives in certain European healthcare systems that result in late diagnosis and inadequate care. The EC-funded project EUROPLAN aimed to support the development of national plans and strategies, a support that is now secured through the EUCERD Joint Action: Working for Rare Diseases.
Before 2009, only four Member States had such a plan (France, Portugal, Bulgaria and Spain) but by January 2014, 16 EU countries had adopted a national plan or strategy, while seven more were at an advanced stage of preparation. The implementation of these plans varies considerably across Member States, because funding often comes from the general healthcare budgets, at the present time heavily under pressure due to the economic crisis. Moreover, plans/strategies for rare diseases in Europe are at different stages of the process: some have just been adopted, others are more advanced. Additionally, there is some confusion as to the areas that are covered in the plans. Notably, rare cancers are part of the scope in some countries and excluded in others.

When it comes to the definition of rare diseases, there are still some discrepancies, as not all EU countries adopt the recommended definition that any disease affecting fewer than 5 people in 10,000 is considered rare. In Sweden and Denmark, for instance, it is required that no more than 500 people in the overall country population are affected by a disease for it to be considered rare.
Regarding codification, all Member States are using the World Health Organization ICD-9 or ICD-10 codification systems from which most rare diseases are absent. Recently in some countries Orpha Codes have been added to integrate the ICD codification and the EC is looking at ways to support such initiatives.

Concerning access to proper information for patients, Orphanet has expanded to include over 6000 diseases, as well as features for queries and a database for best practice guidelines.
DG SANCO works very closely with the EC Directorate-General for Research (DG Research) to be able to fund large research projects. 120 projects were funded from 2007 to 2013 under the seventh Framework Programme for Research (FP7) in very different rare disease areas. Also, the EC supports international collaboration in research for rare diseases with the International Rare Disease Research Consortium (IRDiRC) that launched in 2011 with the objective to deliver 200 new therapies for rare diseases as well as diagnosis for most rare diseases by the year 2020.

At the national level, only a few Member States have specific programmes targeting rare diseases while in most countries support for research on rare diseases comes from the general research programmes.

At this time, 588 rare disease registries have been identified, most of which are established in public or academic institutions and only a minority managed by industries, biotechs or patient organisations. A platform to improve quality, comparability and sustainability of registries and databases for rare diseases will be created in collaboration with the Joint Research Centre (JRC). The platform's main objectives will be to provide a central access point to relevant information for all stakeholders, as well as to support new and existing registries, to develop their interoperability by developing the necessary IT tools and to secure their surveillance. The EC hopes that the initiative will lead to a substantial quality increase and long-term sustainability of rare disease registries, in the same way the EC is doing in relation to cancer data.

Concerning Centres of Expertise, Directive 2011/24/EU on Patients' Rights in Cross-border Healthcare, that ensued rulings of the European Court of Justice on the matter, recognises the right of patients to seek treatment in other EU Member States. The Directive clarifies patients' rights to seek treatment abroad and establishes firm bases for cooperation among national authorities: it is the first time that collaboration between centres is laid out so clearly. In compliance with the Directive, the EC in March 2014 adopted the Delegated and Implementing Acts that set out the criteria and the conditions for European Reference Networks (ERNs) and for those healthcare providers joining the networks.

Article 13 of the cross-border healthcare Directive addresses rare diseases specifically and provides specific tools for patients affected by these conditions travelling within the EU.
In relation to Centres of Expertise (CEs), Member States have taken different approaches within their healthcare systems. Some countries have officially designated Centres at national and/or regional level. Criteria vary across countries but altogether they are often in line with those laid out in the EUCERD Recommendations on Quality Criteria for CEs.

On the topic of patient empowerment, the extent to which patients are the "motor" of the process is recognised - their activism is crucial. According to Orphanet, 2512 specific patient organisations exist that are established at the EU, national and regional levels. All national authorities when asked by the EC have declared engaging patient groups in their policies on rare diseases.

For the treatment of rare diseases, the Orphan Drug Regulation is still the reference legislation. However, most rare diseases lack a specific treatment. More than 1000 products have been designated in the EU as 'Orphan Medicinal Products' and over 90 have been granted marketing authorisation at the time of the ECRD 2014 Berlin. Despite the existing incentives, Orphan Medicinal Products are not equally available to patients in all Member States, as their availability depends on the national systems, with economic conditions having an impact as well.

Working Groups have been established in order to ensure better coordination among Member States in the evaluation of orphan medicinal products, with the objective to boost real access in EU countries.

For newborn screening, in the 2008 Communication on rare diseases, the European Commission committed to conducting an evaluation of current population screening (including newborn screening) schemes for rare diseases and potential new strategies. The mapping covered centres, disorders screened, and the number of infants covered. The results highlighted a very diverse landscape across Europe. The EUCERD adopted an Opinion on the areas of potential collaboration amongst Member States including, for instance, Standard Operating Procedures for communication to patients, guidelines for patient management, information material for parents training schemes, and networking between laboratories. The establishment of key public health indicators would also be important.

Regarding governance at the EU level, the European Commission established a Committee with an advisory role, the EU Expert Committee on Rare Diseases, EUCERD, which was mandated from 2009 to 2013 and delivered five sets of Recommendations, a bimonthly newsletter and annual reports on the state of the art of rare diseases in Europe.

The new Commission Group of Experts on Rare Diseases, which replaced the EUCERD, met in February 2014 for the first time and hopefully will be instrumental in advancing rare disease policy in the EU.

Mr Ryan was pleased to see that many people came to ECRD 2014 Berlin from outside Europe. The EU and its Member States are seen as leaders in developing actions on rare diseases and it will be beneficial if they can influence other countries. There are examples of specific actions outside Europe: for instance, Orphanet is a truly global source of information, currently embedded in the EUCERD Joint Action, and taking part in the update of ICD-10; IRDiRC is another example of European co-leadership at the international level.

In conclusion, progress has been made and cooperation between MS and stakeholders has improved. The preparation of national plans and strategies for rare diseases proved to be stimulating and effective even though some Member States do not yet have a plan, as many are in the final stages of development. The adoption and implementation of National Plans remains a key priority for the European Commission.

The Commission intends to maintain its coordination role in support of national plans and strategies. It also wishes to use its main relevant funding programmes, the Public Health Programme and Horizon 2020, to support rare diseases as a priority, and specifically the IRDiRC, the codification of rare diseases, the development of European Reference Networks, the identification of e-health solutions in the area of rare diseases, as well as the creation of a platform for rare disease registration, an initiative that will demonstrate leadership at the global level.
Rare Disease Day is an example of effective mobilisation and the Commission would be happy to find a way to support this initiative.

Round Table

The round table focused on the main challenges that remain in the areas of organisation of healthcare, research, and the development and availability of orphan medicinal products in Europe.

Kate Bushby, Professor of Neuromuscular Genetics at the Institute of Genetic Medicine of the University of Newcastle, UK, identified three key challenges for the time ahead: the harmonisation of standards of care; the way expertise, advice and services are supported and care is coordinated; and the pathways for patients to access diagnostics and treatment.

The process of concentration of expertise has been taking place for some time and it is now time to build networks to make sure that patients are diagnosed and offered care. Investment in Centres of Expertise and European Reference Networks is necessary as there are still major gaps. Filling these gaps is possibly the greatest challenge both for well-known and less understood diseases. In the process of designation of Centres of Expertise the challenge is to support and define them. A driver in this process is the possibility for a Centre to become part of a European Reference Network in the future. Pr Bushby stated that in her experience networks can work: it is now a priority to ensure support for these networks, especially financial support, in order to make them a reality and give all rare disease patients a home in the long run. In particular, funding European Reference Networks is a big challenge in a context of numerous economic pressures.

Centres that are to be part of European Reference Networks need to show that they can meet standards over a certain period of time. For the functioning of European Reference Networks, it is vital to secure "glue money" for the networks: this is especially important where services are less developed or for areas/diseases that are less developed.

John Ryan recalled that EU Health Ministries are concerned by the financial and demographic crisis, its impact on healthcare systems and the difficulties to maintain standards of care. Chronic diseases, in particular, add pressure and it is unavoidable that policymakers must make choices on what services to maintain. Financing new services appears therefore particularly challenging. A "European legal route" would help EU Ministries when negotiating budgets, as having clear EU guidance on a given matter does carry some weight and has an impact on national Ministers' financial decision-making.

SUMMARY OF PLENARY SESSION

There is a strong case for improving care closer to patients: expert centres still need to develop these guidelines, which should take into account input from patients and families.

A representative of an Italian patient group shared their experience in mapping centres for their patients, as those officially designated do not always correspond to the "real" ones, i.e., those offering actual expertise for their disease. Guidelines from the EU would make a difference. Pr Bushby stressed that patients know where centres are and the task for the years to come is to make the designation process mature.

Serge Braun, Chief Scientific Officer, AFM (Association Française contre Les Myopathies), France, mentioned the close cooperation established between patients and Centres in France that are sometimes co-funded by patient organisations. It is important to rethink the economic basis for centres that need to stay as "Centres of Expertise", as sometimes it is preferable to concentrate all expertise in one place instead of travelling across the country.

Dr Ségolène Aymé, Director of Research and International Affairs at ORPHANET-INSERM, France, reported that due to sequencing, research on rare diseases has significantly accelerated its pace, but it is still almost impossible to interpret data as most of the background data that are necessary are not known. Databases storing data on sequencing are in ten major repositories around the world that do not communicate with each other. Hence we lack the global picture of what is currently known. Databases for this purpose should be created and dialogue should be made possible. These solutions are expensive however and require specific decisions to be taken.

In order to speed up the discovery of natural history and pathophysiology of diseases, while it is not possible to set up registries for all rare diseases, creative solutions need to be found: for example by optimising costs, linking electronic health records that already exist or solving the coding issues to assign a clear diagnosis to each given patient. This latter component is on its way: progress is to be expected in 2016.

Finally, Dr Aymé called for a cultural change that consists of the acknowledgement of the need for academics and industry to work together. In isolation, academic research is not as successful as hoped and industry struggles to develop research in all fields. Resistance to this cultural shift should be overcome by creating a common pot and common approach as the best way to make sure that years of research and resources are not wasted.

Serge Braun recalled that numbers are vague, but rare disease patients in Europe are estimated to be between 27 and 36 million. It is fundamental to be exhaustive in registration and make sure that all have access to healthcare systems. When patients self-report, this should happen in a curated and managed way.

IRDiRC objectives imply that we still need some additional 100 OMPs on the market in the next six years. Many developers struggle through the regulatory process and creative solutions are also needed in the regulatory field. We need a learning process where all stakeholders are collaborating, including the EMA. The European Medicines Agency, in particular, needs to interact more closely with the FDA, and other collaborations need to be fostered and improved: patient organisations should cooperate more in public-private partnership and with industry, as there are plenty of opportunities out there. For industry it is important to remember that rare diseases are a model for more common disorders as well.

Tsveta Schyns, Executive Secretary and Founder of the European Network for Research on Alternating Hemiplegia (ENRAH), spoke about the numerous genes discovered, the new technologies bringing more information and the role of patients in this fast evolving scenario. A new approach to the design of bio-repositories needs to be developed and put in place. With the availability of new sequencing technologies, the discovery of new variants and genes, if patients are not fully involved in the process, results may be faulty and patients will not receive the information on the research they are concerned about, and, in many cases, to which they have contributed.
It is also important that what is discovered then finds an application to have an impact on healthcare and the effective use of therapies. At this stage, researchers need to share results with patients. Ms Schyns recalled the experience of AHC (alternating hemiplegia of childhood): when the pathophysiology was discovered, everything changed and a new investigation approach was required to move to the next phase, in which patients were also involved. AHC patients and families have been "on the move" all the way through the process.
Wills Hughes-Wilson, Chief Patient Access Officer at SOBI, Sweden, focused on therapies development and what needs doing in this area, inviting the audience to consider patient access to orphan medicinal products as a pathway. She called for a collaborative and multi-stakeholder approach from start to finish, whereby all initiatives are brought together for ensuring better and uniform access to orphan therapies. Ms Hughes-Wilson stressed that we should work with the end in mind: the fruit of research is treatments in the hands of patients. If years ago each player strived to do their part in the overall pathway, today it is clear that that all actors need to think of the whole path and how they are going to connect with each other and other endpoints along the way.

Initiatives such as the CAVOMP, adaptive licensing, parallel Scientific Advice/Protocol Assistance, and multi HTA advice are all extremely positive, but they are not going to deliver if all those concerned do not join together to deliver the end goal. Every stakeholder has to play their role: if someone has to do the first steps, all actors are invited "to go and dance on the dance floor".

Pauline Evers, from EGAN, the European Genetic Alliances Network, underlined the importance of taking into account how the lives of patients are affected by new medicines. Patients have many stories to tell that are relevant for the entire process of new medicines development. Not only chemical parameters and statistics, but also patient reported outcomes, with their real life elements, need to be part of the picture. Patients need to be involved from the very start of the programme. Insurers should also be involved so they can help make sure that products do not remain on the pharmacy shelves.

Bruno Sepodes, Chair of the COMP (Committee for Orphan Medicinal Products) of the European Medicines Agency, recognised the role of patients in showing value to all stakeholders involved. With 200 therapies due for 2020, the workload for the COMP is remarkable. However, the regulator is keen to support the development of products that can make it to the finish line. Dr Sepodes compared the whole process to a 'marathon' that needs to be run side by side. Scientific Advice and Protocol Assistance are wonderful services available to sponsors and are instrumental to the achievement of 200 medicines on the market by 2020. All stakeholders need to share knowledge, and dialogue with regulators needs to start as early as possible in the process to increase the chances of success.

As for patient participation in the regulatory process, the COMP Chair recalled how this is essential in his Committee and expressed a wish that patient representatives will also soon join the CHMP.

Durhane Wong Rieger wrapped up the roundtable discussion by stressing that the agenda needs to be pushed further by partners willing to play together. Policymakers from different regions also need to work together. This would be a great step forward and provide an example of how to bring the pieces of the puzzle together.

THEME SUMMARIES

THEME 1 | IMPROVING HEALTHCARE SERVICES

The development of Centres of Expertise and European Reference Networks in the field of rare diseases is encouraged in the Council Recommendation on an action in the field of rare diseases and in the Directive on the Application of Patient's Rights in Cross-border Healthcare as a means of organising care for the thousands of rare conditions affecting scattered patient populations across Europe. The overall aim involves linking the Centres of Expertise through European Reference Networks in order to gather expertise and improve healthcare for rare disease patients. Theme 1 looks at the experience gained so far in implementing these concepts.

Centres of expertise (CE) are major pillars for rare disease (RD) planning organisation across the EU and major pieces of the future puzzle for European Reference Networks (ERNS). Major differences in size, organisation, disease coverage, scopes, overall care, diagnosis, access and research are observed according to country size and healthcare systems, including resources and expectancies. **In the 1st session**, chaired by Pr Hélène Dollfus, Professor of Medical Genetics, Faculty of Medicine, University of Strasbourg, France, three outstanding examples of CE illustrated this situation:

The Centre for Epidermolysis Bullosa (EB), a condition inducing extreme skin fragility, is run by Pr Leena Bruckner-Tuderman, Professor and Chair of the Department of Dermatology, University Medical Center, Albert-Ludwigs University of Freiburg, Germany. Her personal interest in this condition was the basis for this now internationally recognised centre, ensuring for Germany and other countries, clinical, pathologic and genetic diagnosis combined with very high level research. An EU rare disease network encompassing a dozen of disease groups would be the natural continuation to develop this highly specialised care and translational research.

The Aarhus University Hospital Centre of Rare Diseases, headed by Pr John Rosendahl Ostergaard, is one of two rare disease centres in Denmark and treats more than 100 different rare diseases with a « hands on » dedicated team. The difficult transition from childhood care, when the developing child is considered as a whole, versus the various organ specificities of adult care was stressed, emphasising the need to implicate many medical specialities for the best management of rare diseases.

The last example, presented by Pr Pierre Sarda, Département de Génétique Médicale, Hôpital Arnaud de Villeneuve, France, described a regional initiative in the French region of Languedoc-Roussillon with a population of 2 million, of whom 3% face the daily difficulties and challenges of living with developmental anomalies and/or intellectual disabilities. An alliance was created to complement the rare disease centres in order to meet the need for social, educational, and family support and especially the training of educational professionals.

The 2nd session, chaired by Enrique Terol, Policy Officer, DG SANCO, Health and Consumers Unit, European Commission, was dedicated to the designation and evaluation processes for Centres of Expertise (CE), a crucial topic considering that Centres of Expertise are key components of healthcare planning for patients with rare diseases and will become core elements of the future European Reference Networks (ERN).

The French experience since 2009 was reported by Pr Sabine Sarnacki, Coordinator of the expert centre on anorectal and rare pelvic malformations, Hôpital Necker Enfants Malades, Paris. France, has the most comprehensive network of such Centres of Expertise, which were designated through a call for proposals during the first national plan for rare diseases (2005-2008), and evaluated at two time points: an auto-evaluation after 3 years, and an external evaluation with site visits performed after 5 years. During the second plan, the evaluation process was simplified to include an annual activity report and an external visit after 4 years only if problems are detected.

The English experience, presented by Dr Edmund Jessop, Medical Adviser, National Health Service, UK, was less rare disease focused as England has established commissioned specialised services, which are not specific to rare diseases. The evaluation focusses on the clinical outcome (mortality, quality of life, recovery), safety, and the patient experience. For each centre, a small number of items are selected for routine monitoring.

In the last presentation Enrique Terol, Policy Officer, DG SANCO, Health and Consumers Unit, European Commission, introduced the audience to the criteria which will be applied to select the Centres of Expertise constituting the future ERNs: 6 sets of criteria for networks, 5 sets of general criteria for centres, and 2 sets of specific criteria for centres. The assessment will be performed by an independent body.

The 3rd session, chaired by Dr Till Voigtländer, Clinical Institute of Neurology, Medical University of Vienna, Austria, looked at the situation for the European Reference Networks. The concept of European Reference Networks (ERN) was developed through discussions between the Member States and the Commission, and was inspired by the documents produced by the Rare Disease Task Force and the EUCERD. It is now embedded in the Cross-border Healthcare Directive as a way to improve access to diagnosis and provision of high-quality healthcare and to be applied not only to rare diseases but also to complex care (transplantation, highly specialised surgery) and chronic infrequent diseases, such as Parkinson's disease.

Two legal Acts were recently published to define what is expected of the ERN and to define the designation procedure. The main principles to be applied will be: clear and solid eligibility criteria of the networks; a key role for Member States: endorsement and approval; voluntary participation and a commitment to the rules; a transparent and efficient process; and a strong independent technical assessment. The first call for proposals is expected in 2015. As an illustration of what an ERN could be in the future, the experience of TREAT-NMD was presented. While this network can be seen as an model to follow, this is unlikely to happen as the ERN have no funding anticipated to support their networking activities, while TREAT-NMD had the necessary budget to carry out its activities.

In Session 4, Pr Sabine Sarnacki, Coordinator of the expert centre on anorectal and rare pelvic malformations, Hôpital Necker Enfants Malades, Paris, France, presented the new organisation of Centres of Expertise (CE) in France, a network aiming to clarify the current landscape which consists of 131 Centres of Expertise and more than 500 centres of competence. The main objective is to improve the continuum between diagnosis, care, research and therapeutic development, medical and social management and to integrate the Centres of Expertise with the health system. These healthcare networks include not only Centres of Expertise but also centres of competence, forgotten specialities (such as pathology or imaging) patient associations, medical and social workers, research bodies, learned societies and colleges. Twenty-three rare disease healthcare networks ("Filière de Santé Maladie Rare") are now recognised by the Ministry of Health and will be granted

for coordination activities. Inter-regional bodies dedicated to relations with the educational, medical and social sectors will complement this national organisation to support the patients in their life pathway.

The definition of a healthcare pathway (HP) was clarified to mean the coordination of the multiple medical or paramedical specialities required for the management, treatment and well-being of the patient and their family. Pr Kate Bushby, Institute of Genetic Medicine, Newcastle Upon Tyne Hospital, UK, presented their healthcare pathway organisation for neuromuscular diseases and Theda Wessel (Berlin Centre for Rare Diseases, Charité-University Medicine Berlin) reported a simple method they used to simplify the healthcare pathway in their rare disease centre, by obtaining from the hospital direction one single medical file for each patient gathering all speciality reports. The experience of the healthcare pathway referent or complex cases manager of the AFM (France) is cited and should be ideally expanded to all rare diseases generating complex medico-social situations. The implementation of such a professionalisation of care should be adapted to the rare disease policy and the financial constraints of each country.

In the 5th session, chaired by Dr Tjitske Kleefstra, Clinical Geneticist, Radboud University Medical Center Nijmegen, Netherlands, examples of novel diagnostic possibilities were presented. Dr Kleefstra discussed how implementation of whole exome sequencing (all protein coding regions of the human genome) was introduced in the diagnostic process of heterogeneous disorders as hearing loss, vision loss, movement disorders and intellectual disability in the Department of Human Genetics of Radboud UMC Nijmegen. First a general, generic, approach is applied where the whole exome is sequenced in all conditions. This is followed by a specific analysis of selected gene panels for the respective conditions. The diagnostic yield has increased considerably for all conditions. The number of disorders for which whole exome sequencing can be requested in Nijmegen has increased after the positive experience for the initial 6 groups of disorders and now includes amongst others, cranio-facial anomalies, epilepsy, disorders of sexual development, and a group of 'unknown disorders'. Ethical aspects with regard to incidental findings are evaluated continuously.

Dr Wendy Jones, University of Cambridge, UK, presented data on the 'Deciphering Developmental Disorders' project. Within this project the aim is to establish diagnoses by applying current technologies as array CGH and whole exome sequencing, and systematically looking for the presence of uniparental disomy and mosaicism. Data from over 1000 trios (child and parents) have been collected and diagnostic yield so far is 16%. Examples on findings of novel causes and clinical reporting processes were highlighted.

Dr Ole Winther, Associate Professor, DTU Informatics, Technical University of Denmark presented the development of the search engine FindZebra. The internet has become a primary information resource about illnesses and treatments for both medical and non-medical users. Standard web search is by far the most common interface for such information. It is therefore of interest to find out how well web search engines work for diagnostic queries and what factors contribute to successes and failures. Among diseases, rare (or orphan) diseases represent an especially challenging and thus interesting class to diagnose as each is rare, diverse in symptoms and usually has scattered resources associated with it. In the presentation, the need for specialised search engines with simple user interfaces was underscored and some perspectives and examples of how information technology can be used to change the way diagnosis is approached were provided. The FindZebra search engine is available at http://www.findzebra.com/.

The new 'Massive Parallel Sequencing' or 'Next Generation Sequencing' (NGS) tools are rapidly being transformed from research applications to diagnostic methods. Clinical utility, technical validation and appropriate reimbursement models are a few of the key issues that have to be addressed, to pave the road towards the medical use of these technologies. The clinical application of NGS could help a lot of rare disease patients and families, and shorten the journey to diagnosis for many of them.

At the European level, EuroGentest has taken an initiative to write guidelines for the application and validation of diagnostic NGS tests. **In Session 6**, chaired by Pr Gert Matthijs, Coordinator of EuroGentest, Laboratory for Molecular Diagnosis, Center for Human Genetics, Belgium, the Pr Peter Bauer, who leads the Molecular Genetic Diagnostics laboratory and the Core Unit for Applied Genomics at the University of Tübingen (Germany), gave a summary of the possibilities and challenges of NGS. He explained features such as 'diagnostic routing', 'gene core lists' and proposed a scoring system for NGS tests, which would allow doctors, patients and policymakers to compare tests. Pr Gert Matthijs raised the issue of how NGS tests will eventually be reimbursed by the national healthcare systems. In Belgium, gene panels have tentatively been included in the current system for reimbursement of genetic tests. But the budget cannot accommodate the costs for whole exome or whole genome sequencing. This is a pity, because rare disease patients and their families would especially benefit from the reimbursement of the tests. However, it is not just a matter of prioritisation; it is also a matter of safeguarding what has been accomplished by the national healthcare systems in different countries.

Pr Milan Macek Jr, Chair of Biology and Medical Genetics, University Hospital Motol, Charles University, Prague, Czech Republic, and former president of the European Society of Human Genetics, presented case studies of inappropriate use of patients' and (grand)parents' confidence in the healthcare system and imbalanced marketing of diagnostic tests by certain organisations that offer genetic tests directly to primary care physicians and the public. These offers fall into the category of 'genetic horoscopes'. It is important to apply both a top down (regulatory) and a bottom up (informing patient groups) approach to this issue, which otherwise threatens to undermine public confidence in genetic testing.

THEME SUMMARIES

THEME 2 | KNOWLEDGE GENERATION AND DISSEMINATION

Without accessible, accurate information on rare diseases, diagnosis and appropriate care are delayed. Without raising awareness and improving education about rare diseases and their impact, clinicians do not have the tools to do their job effectively, patients and care-givers are unsupported and isolated and research and industry are deprived of essential incentives and data to develop effective treatments. Theme 2 covers all aspects of knowledge generation and dissemination, from coding and registries, to publicity through media. The theme also examines the potential challenges and rewards from the perspective of all stakeholders.

In Session 1, chaired by Christine Lavery, Chief Executive, Society for Mucopolysaccharide Diseases, UK, short presentations on Rare Disease Registries (Where we are today and how they might look in 2020) were made by Elfriede Swinnen, Scientific Institute of Public Health, Belgium; Daniel Rosenberg, Senior Director, Actelion; and Tarek Hiwot, Queen Elizabeth Hospital, Birmingham, UK.

Collecting data on rare diseases is necessary for pre- and post-marketing medicines development stages to speed up clinical research and provide data to regulatory and reimbursement bodies. However, there are important questions to address such as: "Can these registries better serve the rare disease community, in particular patients, if not maintained by industry?" and "How do we take advantage of new technologies to share data repositories?" There was a robust panel discussion with members, including Jeremy Manuel, European Gaucher Alliance UK; Carla Hollak, Academic Medical Centre, Amsterdam; Micheline Wille, Shire, Switzerland; and Leeza Osipenko, Nice UK.

The overwhelming conclusion of the workshop was that industry, clinicians, payers, regulators and patient organisations should lose no time in recognising the need and moving towards disease registries independent of the pharmaceutical industry which still meet regulatory responsibilities for pre- and post-marketing approval. There was a call for the European Medicines Agency (EMA) to show leadership and start a process that requires companies to contribute to an independent disease registry rather than develop their own product registry. There was also a call for the EMA's Committee for Medicinal Products for Human Use to clearly state what they are asking for, as their advice appears vague in this area.

RD patient registries and data collections need to be as internationally interoperable as possible and the procedures to collect and exchange data need to be harmonised and consistent to allow pooling of data when it is necessary to reach sufficient numbers for clinical research and public health purposes.

In Session 2, chaired by Stefan Schreck, Head of Unit, Health Information, DG SANCO, European Commission, examples of efforts in this direction were presented. The European Commission supports the development of a European platform on rare disease registration at their Joint Research Centre in Ispra, Italy. This platform is intended to serve as an aggregation point for all rare diseases, across all European countries, and will become a centre of rare disease-knowledge generation, acting as a reference for policymakers, which will also ensure coherence with international initiatives. The platform will provide IT tools, guidelines, standards and training.

Three countries presented their initiatives to collect data at the national level. The French national data repository for rare diseases collects a minimum dataset coming from patient files from the national Centres of Expertise (CE). It currently stores data from 235,000 patients from 62 CE, affected by 4,136 distinct disorders. The Spanish national rare disease registry is in development. It will be populated by data coming from disease-specific registries and from regional population-based registries. The German solution is to develop an open-source registration system at the disposal of the rare disease community. This session showed that there are several ways to fill the data gap and that it is of importance to share these experiences with other countries so they may make an informed decision on the best solution for their needs, keeping in mind that the interoperability between all these systems is essential.

Session 3 focused on "Making the invisible visible: the coding of rare diseases in Health Information Systems Interpretation" and was chaired by Ségolène Aymé, Emeritus Director of Research, Director of International Affairs, ORPHANET-INSERM, France. Much data is available in health information systems but the data concerning rare diseases is not identifiable due to the lack of specific codes for most rare diseases. The International Classification of Diseases (ICD 10) has precise codes for only 466 rare diseases and the next edition, which should include all rare diseases, is not expected before 2017. SNOMED-CT has codes for 2,883 rare diseases as listed in Orphanet and arrangements are in place to expand the list in both SNOMED-CT and ICD 11, through collaboration between Orphanet, ICD and SNOMED-CT. Orphanet offers a coding and classification system which is multi-hierarchical and cross-referenced with ICD, SNOMED-CT, UMLS, MeSH, MedDRA and OMIM. This is freely available for download at www.orphadata.org. Orphanet suggests the addition of Orpha codes to ICD codes in national health information systems until the international nomenclatures are comprehensive for rare diseases. This would generate a significant volume of relevant information very quickly and at minimal cost.

This session was the occasion to review the experience of Germany where it has been decided to code rare diseases with ICD 10-GM (German modification) and Orpha numbers in routine settings. In its pilot study, DIMDI has cross-referenced ICD10-GM with Orpha codes in order to identify the missing codes in ICD and add these into its Index database. The evaluation of the project and of its implementation within the German system will be available in 2016. Another presentation gave an overview of the importance of using one language to make the most of existing data and information generated by science via the sequencing effort, as well as through electronic health records which are being introduced progressively by many countries. The proposed unified language to describe the phenomes is HPO, the Human Phenome Ontology, together with Orphanet codes for the final diagnosis.

Session 4, chaired by Lesley Greene, Vice-President, CLIMB, Vice-Chair Committee for Orphan Medicinal Products (COMP), Volunteer Patient Advocate, EURORDIS, sought to answer the question: "Delivering Help and Support in a Virtual World: What Will Work Best?" The European Network of Rare Disease Helplines Caller Profile Analysis was led by EURORDIS to examine how the quality of helpline support could be improved. Twelve services from eight countries collaborated, sharing experiences and expertise to provide their informed support for emerging services in Europe.

The cross-sectional survey covered all enquiries through October 2013 and identified the unmet needs of enquirers, which patients hope will be met by Centres of Expertise. The importance of helplines in national plans was discussed. Different stakeholders seek multiple sources of information. The internet has played a valuable role but helplines have an added-value in the "internet era" by being tailored to user needs and profile. Websites give information but do not communicate with support. They cannot listen and react in an appropriate way. Studies show internet use can increase anxiety, and it is not accessible to poorer, less educated communities or delivered in local languages. Helplines can represent excellent observation tools through which Health Authorities can receive feedback both from patients and professionals concerning the functioning and possible improvements of the rare disease policies put in place.

The proposal for a European 116 helpline was examined. Based on studies and needs, there is obvious value and interest. A 116 helpline would provide high visibility for the rare disease community, support the CBHD and a stronger helpline network. Concerns remain, however, regarding the essential interaction with Member State ministries and national rare disease alliances. Funding, training, monitoring, owning, and sustaining, the service remains a concern.

Despite progress made by services in the field of rare diseases, and increased use of social media, patients still value the traditional helpline, effectively empowering them by offering the capacity to communicate, collaborate and advocate for their community's needs. The combined use of different media can reduce inequalities in access to the services but patients with a rare disease still favour the telephone. People need people.

The 5th session, entitled "Knowledge at the point of care: getting the facts just in time or just in case", chaired by Peter Farndon CBE, Director, National Health Service, National Genetics and Genomics Education Centre, UK, considered how people with rare diseases and their clinicians need information and knowledge - not only for an understanding of the disease but also to provide optimal care in emergency situations. The Chair referred to these two scenarios as "just in case" and "just in time" information. Health professionals acquire the former traditionally through their pre-registration training, but once in practice immediate access to information and knowledge to deal with (perhaps unexpected) clinical situations is required. Four experts shared their experiences in practice and conference participants engaged fully in the discussions which followed.

Domenico Taruscio, Director, National Centre for Rare Diseases, Italy, highlighted a project to identify the procedures for the development and evaluation of clinical practice guidelines for rare diseases, and to train professionals to use them. A particular challenge is developing methodology to take into account a relative lack of publications and to assess information based on a limited number of patients or the experience of clinical experts. Such a methodology is particularly important for rare diseases because the development of most guidelines requires a large amount of peer-reviewed data. Results from the project are expected at the end of the year. Points in discussion suggested that where evidence is limited, a consensus view of best practice is better than no information. It is sometimes important to release information to confirm that some patients with rare diseases require no differences in treatment and management from standard procedures.

Ana Rath, Director, Orphanet, France, described Orphanet's emergency guidelines (covering 50 rare diseases and downloaded 340,000 times) and how they are developed with consultations between patients' organisations, reference centres, and other health professionals. Discussions highlighted the importance of increasing awareness of available material and the importance of translating existing guidelines into other languages.

Tino Muenster, Project Manager, OrphanAnesthesia, Germany, explained how searching for information about anaesthesia for people with a rare disease led to his developing OrphanAnaesthesia - currently with 35 guidelines published but a plan to increase them to over 100. He highlighted how an internet search for a particular condition brought up hundreds of thousands of web pages but it would take a great deal of time and expertise to decide each one's "fitness for use" - not possible in an emergency situation. In the discussion, patient organisation members highlighted how they could disseminate information by including website links for guidelines in their literature and websites.

It was noted that the use of technology is changing how people access health information, particularly for immediate clinical use. The use of apps on smart phones and tablets by staff and patients to present or access information at point of care seems to be gaining in acceptance, although health professionals react better if the information is "badged" as coming from a (national) organisation they trust. Everyone agreed that it is a slow process (usually taking about a year) to ensure consensus and peer review of information, but this is considered to be a vital component of the process.

Jordi Llinares Garcia, EMA, UK; Debra Lewis, FDA, USA; Elizabeth Vroom, The Duchenne Parent Project, The Netherlands; Kris De Boeck, University Gasthuisberg, Belgium

Session 6, entitled "Hype, Help or Harm? The Impact of Media Promotion of Rare Diseases", chaired by Denis Costello, Web Communications Senior Manager & RareConnect Leader, EURORDIS, Spain, featured presentations from Rinke van den Brink, Dutch National Television, who gave an overview of media coverage in the Netherlands in 2012 after the Dutch payer decided to revise the reimbursement criteria for treatments for Fabry, Pompe and Gaucher diseases. Avril Daly, Vice-President, EURORDIS, Chair, Genetic & Rare Disorders Organisation (GRDO), Chief Executive Officer, Fighting Blindness, Ireland, gave an overview of the strategy behind EURORDIS plans to advocate for a European Year for Rare Diseases in 2019. Lastly, Alessia Daturi, patient organisation liaison officer with Telethon Italia shared their experience in communicating about rare disease research to the general public and shared some case studies on issues relating to gene therapy and stem cell research.

Questions and comments reflected that patient groups need to be more discerning and take their power back when it comes to speaking to media. There was a feeling that it is quite easy to be exploited. However, the audience also realised the need for media support to raise crucial awareness. There have been experiences with some very good journalists and successful articles as well as television and radio exposure for rare disease campaigns. It can be very difficult to say "no" to an opportunity to tell our stories but sometimes the timing and other elements must be considered. Media training could be helpful in this area.

THEME SUMMARIES

THEME 3 | RESEARCH FROM DISCOVERY TO PATIENTS

Rare disease research is still too fragmented and compartmentalised. This leads to lack of integration, duplication of efforts, lack of critical mass, thinking in "silos" and waste of resources. It also hinders progress towards better diagnosis and therapy for rare disease patients despite many opportunities offered by new technological developments. To improve the situation and ensure a rapid translation of discoveries into operational diagnostic and therapeutic tools, several initiatives have emerged at local, regional, national and international level, which were reported, along with their outcomes, during the sessions in Theme 3.

Session 1, entitled "Shaping Rare Disease Research Policy", was chaired by Dr Ségolène Aymé, Emeritus Director of Research, Director of International Affairs, ORPHANET-INSERM, France. Rare diseases represent a challenge too big to be mastered alone - collaboration is the key. With over two decades of investment in the area, the European Commission is a major player in funding collaborations for rare disease research. Irene Norstedt, Head of Unit, Personalised Medicine, Health Research Directorate, DG Research and Innovation, European Commission, presented the research programmes funded by the EU. The Seventh Framework Programme (2007-2013) had over €620 million invested in close to 120 collaborative projects (e.g., IRDiRC, RD-Connect). The 2014-2020 programme for research and innovation (around € 80 billion) – known as Horizon 2020 – brings all of the EU's research and innovation funding programmes under a single umbrella. The proposals earmark funding for three key objectives: excellent science, industrial innovation leadership and tackling societal challenges.

Horizon 2020 will continue to support IRDiRC goals, which were presented by Paul Lasko, Chair of the International Rare Diseases Research Consortium (IRDiRC). IRDiRC teams up researchers and organisations investing in rare disease research to stimulate, better coordinate, and maximise output of their efforts around the world. IRDiRC aims to achieve two main objectives by the year 2020, namely to deliver 200 new therapies for rare diseases and the means to diagnose most rare diseases. IRDiRC members include funding agencies, academia, industry, and patient associations. So far, IRDiRC has enlarged the consortium to more than 35 members in 15 countries over four continents (Asia, Australasia, Europe and North America), creating a favorable eco-system to foster international collaborations. Another coordinated European approach (also a member of IRDiRC) aiming to address the needs of the rare disease community is the E-Rare Consortium, which was presented by its coordinator, Daria Julkowska, E-RARE Coordinator, Fondation Maladies Rares, France. The E-Rare-1 project (2006-2010) leveraged funding for rare disease research in countries without specific programmes for rare diseases and thus enabled the participation of researchers in these countries in transnational projects. The E-Rare-2 project (2010-2014) aims at deepening and extending the cooperation among E-Rare-1 and new partners. In the future, the E-Rare-3 will go beyond Europe, from 17 current partners in 13 countries to 27 partners in 17 countries.

Session 2, "Addressing the Gaps in Research at International Level to Identify Opportunities", was chaired by Pr Milan Macek, Jr, Chair of Biology and Medical Genetics, University Hospital Motol, Charles University, Prague, Czech Republic. The International Rare Diseases Research Consortium (IRDiRC) (www.irdirc.org), which launched in 2011, is an umbrella organisation for more than 30 participating global funding organisations and their aligned research projects. The IRDiRC's two main objectives are to deliver 200 new therapies for rare diseases and the means to diagnose most rare diseases by the year 2020. Three scientific committees (Diagnostics, Interdisciplinary and Therapeutics) advise the Executive Committee (comprised of funders) on the best way forward to meet these goals. Dr. Kym Boycott, Investigator, Children's Hospital of Eastern Ontario, Canada, presented the activities of the Diagnostics Scientific Committee focused on enabling the diagnosis for most rare diseases by 2020. Meeting this goal will require an understanding of the mechanism of most rare diseases and the facilitation of clinical translation of genomic sequencing for patients with rare diseases. A number of standards, tools and infrastructures must be put in place and made available to facilitate discovery and collaboration to meet this objective. Most urgent amongst these are a standard ontology to facilitate sharing of rare disease phenotypes and a large-scale platform to share rare disease phenotypes and genotypes to enable the discovery of new disease genes.

Yann Le Cam, Chief Executive Officer, EURORDIS, France, discussed the IRDiRC Therapeutics Scientific Committee, focused on reaching 200 new therapies for rare diseases by 2020. The committee is developing a minimum, though comprehensive, set of recommendations engaging all relevant stakeholders in alternative pathways for medicines development up through approval. These include adaptive clinical trial design and statistical methods, alternatives to animal models, promotion of research on biomarkers and further used on existing ones, and optimal use of sicentific guidance by regulatory agencies. The Committee collaborates closely with the two other scientific committees of IRDiRC for an integrated approach while acting through Working Groups in relevant areas such as Orphan Drugs & Regulatory Affairs, Biomarkers for disease progression and therapy response, Chemically derived products including Repurposing of medicines, and Biotechnology-derived products including cell & gene therapies.

Pr Hanns Lochmüller, Chair of Experimental Myology, Institute of Genetic Medicine, Newcastle University, UK, outlined activities of the Interdisciplinary Scientific Committee which closely collaborates with the other scientific committees and is concerned with cross-cutting themes relevant for rare disease research, in particular ethics & governance, data sharing & bioinformatics, patient registries & natural history studies, and biobanks. Contributions to the IRDiRC road map were proposed, including infrastructural support in these areas to underpin the research and remove bottlenecks towards the ambitious IRDiRC goals.

Session 3, entitled "Incentives to Create a Favourable Eco-System" was chaired by Serge Braun, Chief Scientific Officer, AFM (Association Française Contre les Myopathies), France. Dr. Erik Tambuyzer, Founding Member, BioPontis Alliance Rare Disease Foundation (BArD), Belgium, presented the BioPontis Alliance Rare Disease Foundation as a new philanthropic innovation joint venture model targeting paediatric rare diseases. Because (1) the rare disease field and orphan medicines is precursor (i.e., with much more innovation and less "me-too" products), and (2) patient values may differ from physician and industrial perceptions, the foundation intends to bring the stakeholders - including the patients and clinicians who represent the "real-life" perspective - into the decision process. The mission of BioPontis is to establish a scientific bridge across the innovation gap that will serve as an enabling hub to independent disease foundations, providing a nonprofit partner able to develop drug candidates. The economic model, for instance, replaces upfront IP cost with shared exit IP value. BioPontis organises at the international level innovative project detection and accompanies the inventing scientist in a professional capacity.

Dr Kym Boycott, Investigator, Children's Hospital of Eastern Ontario, Canada, presented the FORGE Canada consortium of 21 Sites, 90 Physicians, and 50 Scientists, which allows rapid identification of genes for rare paediatric single-gene disorders. This network is the result of a public-private partnership covering open access platforms (genome sequencing and data sharing), repurposing, disease pathway interrogation, and preclinical research. It is also linked internationally to other networks such as NeurOmics and RD-Connect.

Virginie Miath, Project and Investment Manager, Conectus, France, presented Alsace Biovalley as an example of a very successful ecosystem based on a new legal framework for research and innovation aimed at fostering the creation of start-ups and the participation of public researchers as shareholders (including the creation of "public incubators" to foster the development of start-ups), a revamped « R&D tax credit » system to attract and develop private R&D investment, and new tech transfer tools. The Alsace region has a unique concentration of research and industrial substrate together with neighbouring German and Swiss regions.

The round table discussion that followed adressed issues such as fragmentation, finding the right connection between local, national and international ecosystems, and building convergence and synergies between the different stakeholders.

In Session 4, "Breakthroughs in Science", chaired by Gert-Jan van Ommen, Department of Human Genetics, Leiden University, Director, Centre for Medical systems Biology, Director BioBanking and Biomolecular Research Infrastructure, Netherlands, an overview of the status of stem cell research in modelling diseases was presented by Christine Mummery, Head of Department, Professor of Developmental Biology, Leiden University Medical Centre, Netherlands, with Hereditary hemorrhagic telangiectasia as a compelling strong example. Pr Mummery showed how stem cell cultures help to assess disease mechanisms and validate new active drugs (e.g., for LQT2 patients) .The discussion addressed the potential of human pluripotent stem cells as disease models, as well as the hypes and hopes of this new technology.

Damian Smedley, Wellcome Trust Sanger Institute UK, highlighted new, publicly downloadable software called EXOMIZER to prioritise putative deleterious mutations for further study (see his Genome Research 2013 paper for further information). Text-mining based phenotype descriptions of human, mouse and zebrafish are interconnected and related to their respective known gene mutations to strengthen evidence of involvement in pathogenesis. This gives a 54x increased power relative to using only in silico pathogenicity predictions. Besides suggesting candidate genes this is an interesting tool for identifying good disease models.

Finally Gert-Jan van Ommen updated the audience on exon skipping drug development, especially in Duchenne muscular dystrophy. After initial phase 2 success, this innovative treatment strategy had a disappointing phase 3 trial, but the results seemed to hold more promise than initially considered. The presentation and discussion highlighted the value of in-depth analysis of trial results and rigorous adherence to phase 2 criteria in phase 3 trials and underscored the need for adapted outcome measures in rare diseases and in particular in neuromuscular diseases.

Unfortunately, not enough time was available for the round table discussion, but the moderator, Serge Braun, Chief Scientific Officer, AFM (Association Française Contre les Myopathies), France, reminded the audience that most fundamentally new therapy developments (such as monoclonals, gene therapy, and possibly antisense, too) show a triphasic N-curve: first a hype phase of (too) high expectations, then a 'sobering' phase with disappointing outcomes, ultimately followed by a slower successful phase based on more realistic insights and approaches.

Session 5 looked at "Pre-Competitive Tools and Resources / Public-Private Partnership in the Area of Rare Diseases (including the Innovative Medicines Initiative)" and was chaired by Nathalie Seigneuret, Senior Scientific Project Manager, Innovative Medicines Initiative (IMI), Executive Office, EU.

The development process of medicines from discovery all the way to patient access is particularly challenging in rare diseases, with models developed outside a clinical context, no access to medicines development advice for academia, little expertise of small pharma and lack of specific rare disease knowledge of large pharma.

In this well-attended session, which started with a brief introduction of the Innovative Medicines Initiatives, one of the largest biomedical research public private partnerships, different experiences of partnerships in rare diseases were presented. Pr Kate Bushby, Institute of Genetic Medicine, Newcastle Upon Tyne Hospital UK, showed how the integration of academia, industry, non-profit, and patient advocacy in the TREAT-NMD Advisory Committee for Therapeutics (TACT) provides multidisciplinary and comprehensive advice which helps academia and industry forward their medicines development in neuromuscular diseases.
This successful model could apply to other rare diseases.

Dr. Lucia Monaco, Chief Scientific Officer, Fondazione Telethon, Italy, explained how partnership with pharma is essential to complete the development and commercialisation of therapies for three diseases which originated from the research funded by Fondazione Telethon. However, forging these partnerships requires determination.

To ensure successful translational research, a rare disease focused infrastructure is needed. A proposal for a pilot of a pre-competitive informatics platform engaging with health systems and patient organisations was presented by Dr Justin Paschall, Team leader, Variation, European Bioinformatics Institute, UK.

The discussion highlighted how these collaborations are essential to address the translational gap in the research for rare diseases. There are clear challenges in building and maintaining these partnerships, in particular the willingness and commitment to collaborate in order to accomplish shared research goals; trust amongst partners; and transparency with sharing of data and results. However, these collaborations create a new ecosystem by aligning public and private interests and bringing together complementary expertise, and foster the development of new treatments for rare diseases for the benefit of patients.

Session 6 provided opportunities to hear how new ways of working and new technologies help stimulate research for diseases that are often marginalised, whilst also highlighting some of the legal and ethical barriers that have to be overcome.

"Whose Data Is It?: Stimulating Research and Removing Barriers " was chaired by Kay Parkinson, Chief Executive, Alström Syndrome, UK.

Outcomes of the session – improved understanding of the research benefits of disease specific work was illustrated by Phil Beales, Professor of Medical Genetics, UCl, UK, who outlined his thoughts for a Ciliopathy Service based on the successful Bardet-Biedl and Alström multi-disciplinary services funded by NHS England, both of which involve the patients' groups as equal partners in service delivery. Pr Beales stressed the benefits of having the patient groups involved.

The presentation of Pr Hans Scheffer, Associate Professor Clinical Molecular Genetics, Radboud University Nijmegen Medical Centre, Netherlands, on the responsible use and indication criteria for next generation sequencing (NGS) diagnostics in clinical practice

raised many issues: whether patients should be informed of other diseases they may have a propensity for beside the disease which was originally being researched; the complexity of the consent forms that patients are asked to sign; whether patients and families fully understand what they are signing; and the great strides forward in disease understanding that NGS will bring.
Gauthier Chassang, Lawyer in EU and International Law, INSERM, France, offered a comprehensive look at the EU Data Protection law reform and scientific research. He concentrated mainly on the new elements of the reform, the reinforcement of data controller's obligations, new rules for personal health data processing for scientific research, reinforcement of prior consent and personal health data for scientific research.

This productive, well attended session gave many different angles on developing research and overcoming barriers.

THEME 4 | STATE OF THE ART AND INNOVATIVE PRACTICES IN ORPHAN PRODUCTS

Fourteen years have passed since the adoption of the European Regulation on Orphan Medicinal Products. Success in the stimulation of the research, development and bringing to the market of appropriate medications for orphan diseases has been achieved beyond expectation. However, work remains to continue to improve the legislative framework, to ensure patients have access to these treatments. The aim of Theme 4 is to examine the latest initiatives and discuss innovative practices in orphan medicinal products at all stages of the development chain. It also addresses the main challenges being faced in accessing and securing the availability of rare disease treatments to patients. The theme includes presentations and panel discussions, enriched with the experience and expertise of the mainstakeholders shaping the orphan landscape today.

Session 1, "Current Landscape of Policy Development on Orphan Products & Rare Disease Therapies" was chaired by Bruno Sepodes, Chair, Committee for Orphan Medicinal Products (COMP), European Medicines Agency. In this session, regulators aimed to provide the latest status of current and emerging regulatory tools to support orphan therapies development and approval processes. The session had the participation of exceptional and well-known experts in the field from the European Medicines Agency (EMA), the Food and Drug Administration (FDA) and industry. The panel was enriched by the presence of Marlene Haffner, Chief Executive Officer, Haffner Associates, USA, who for over 30 years had an immeasurable impact upon the development of orphan medicines and was responsible for the administration of the US Orphan Drug Act, the first legislation of this type in the world. A presentation on the state of play in Europe was followed by perspectives from across of the Atlantic and finished with a broader perspective of countries where an Orphan Regulation is now coming of age and those where intentions and draft regulations are being sought. The discussion between speakers, panelists and audience highlighted: (i) the importance of the parallel work held by EMA and FDA; (ii) the high interest in designations and the importance of seeking scientific advice and of early dialogue; (iii) the existence of many rare diseases still with no therapeutic development; (iv) the development of the 'Rare Pediatric Disease Review Voucher' by the FDA; (v) the importance of having a patient focused medicines development plan; (vi) the 'Breakthrough Designation' and 'Adaptive Licensing'; (vii) the need of collaboration with all stakeholders and consortiums reaching toward a common goal; and (viii) how the success of all these initiatives can be hindered if access to orphan medicines remains a hurdle.

Session 2, "Facts on Current Patient Access Challenges to Orphan Products" was chaired by Thomas Heynisch, Deputy Head of Unit, Food & Healthcare Industries Biotechnology, DG Enterprise, European Commission, EU.This session provided an overview of the various cost-containment measures in the EU and included presentations from François Houÿez, Treatment Information and Access Director, EURORDIS, France; Paolo Siviero, Head of Economic Strategy and PharmaceuticalPolicy, Italian Medicines Agency (AIFA), Italy; and Heidi Wagner, Senior Vice President, Global Government Affairs, Alexion Pharmaceuticals, USA.

The issue of EU reference pricing was raised, and in particular the potentially damaging effects it could endanger around generating a sufficient level of revenues needed to finance investments in medicines, particularly orphan products.

The point was made that patients' dissatisfaction with regard to access to treatment existed before the economic crisis of 2008; however discontent seems to have increased since. Industry restated its interest in maintaining a high degree of cooperation. Competent authorities responsible for pricing/reimbursement made clear that they were facing multiple challenges. The so-called "blockbuster drug model" seems to becoming outdated while more focused medicines tailored to the needs of individual patients or clearly defined patient groups find more widespread use. This more focused therapeutic approach has implications for national healthcare budgets since more individualised medicines tend to be superior in their therapeutic properties while at the same time often more expensive.

Public authorities have to guarantee patients' access to state-of-the-art medical treatment. Fair payment mechanisms which ensure sustainable and predictable public expenditures are required to guarantee access, while sufficient rewards for innovation are crucial to foster innovation. The legal framework for orphan medicines allows them to charge premium prices. Hence the phenomenon of extending the scope of indications leads to severe problems for public health budgets. Member State authorities consider this a prime challenge as it undermines the assumption of classifying orphan medicines as "niche" products whose costs are predictable.

Session 3, "EMA-Health technology Assessment (HTA) Interface on rare disease therapies" was chaired by Spiros Vamvakas, Head of Scientific Advice, Human Medicines Special Areas, European Medicines Agency, EU. Growing interfacing and early dialogue between the European Medicines Agency (EMA) and Health Technology Assessment (HTA) with the involvement of patient representatives is an opportunity for developers to receive simultaneous feedback from both sides on their development plans and reduce the time of orphan therapies availability to patients. During this session, available regulatory options, practical experiences gained so far on parallel protocol assistance with HTA bodies, as well as procedural trends for the future were presented. EMA has provided these opportunities since 2010, but so far developers have used them mainly for medicines for common conditions and one of the goals of this session was to raise awareness for the usefulness of these procedures in the course of medicines development.

In this session Leeza Osipenko, Senior Scientific Advisor, NICE, UK, discussed the elements an HTA body needs to decide on the added-value of a new medicinal product and the possibilities for NICE to be

involved in providing Scientific Advice on the development of new medicines. She underlined the usefulness for sponsors to engage early in dialogue together with both regulators and HTA bodies during development. Dr Helma Gusseck, a patient representative from Pro-Retina, Germany, presented the positive experience she had as a patient representative participating as expert in a Protocol Assistance procedure in the EMA and highlighted opportunities for further improvement such as involvement of more than one patient representative per procedure. Finally, Dr Samuel Rigourd, Global Program Regulatory Director, Novartis, presented the experience of being one of the first companies to apply and receive parallel EMA and HTA Protocol Assistance on a rare ophthalmological condition. Spiros Vamvakas provided an overview of the procedures and therapeutic areas covered already by this approach. There was a lively discussion amongst session participants and many questions on practicalities of how to simultaneously approach Regulators and HTA bodies during the development of an orphan medicine.

Session 4 "Shortages in authorised medicines for rare diseases", chaired by Jeremy Manuel, OBE, Chair European Genetic Alliance, UK, offered the different perspectives of the clinician, the regulator and industry. Pr Carla Hollak, Professor of Inherited Metabolic Diseases in Adults, Academic Medical Centre, Netherlands, discussed the virus that in 2009 affected the USA-based Genzyme plant producing the authorised treatments for Gaucher disease (Cerezyme) and Fabry disease (Fabrazyme). Pr Hollak detailed the approach taken by the Gaucher community and described a meeting held in Bad Honnef (Germany) under the leadership of the medical/scientific group European Working Group on Gaucher Disease (EWGGD) and the patient group European Gaucher Alliance (EGA). She explained how clinicians from around Europe described the extent of the problem and the companies – including two researching potential therapies undergoing phase 3 clinical trials – then detailed the availability of therapy and whether a further supply of medicine (some before final marketing approval) could be made available. A joint strategy was developed and a paper published, co-authored by leading clinicians and patient leaders, which captured the strategy to manage the shortage and set out an agreed priority of the allocation of available treatment. Pr Hollak stressed that the collaboration between all stakeholders was the primary reason for the ultimately successful navigation of the supply shortage.

Brendan Cuddy, Scientific Administrator, European Medicines Agency, EU, described the actions taken by the EMA on product shortages and the obligation of manufacturers. He detailed the various reasons for shortages (economic, business and manufacturing/supply chain) and described the legal framework and statutory obligations of the manufacturers. He discussed the work of the EMA in the development of an implementation plan to help develop a proactive risk-assessment in an effort to prevent shortage problems developing, rather than a reactive policy after they had already occurred. In the belief that the public felt that patient safety is linked to the supply chain, the EMA is taking initiatives to address the issue and is particularly keen to ensure there is more effective communication between the industry and the regulators.

The final presentation was from Dr Carlo Incerti, Senior Vice President, Head of Genzyme Global Medical Affairs Italy. Dr Incerti described general reasons that shortages occur (economic and technical) and showed the complex manufacturing process for biological products. He then detailed the company's handling of the crisis from discovery of the virus and the consequences, expressly discussing the obligations of the company towards regulators, clinicians and patient groups, as well as the shareholders. He evoked the balance between the genuine desire to keep patients and clinicians informed and legal obligations regarding reporting to regulators and shareholders. Dr Incerti emphasised that long-term planning is central to any business continuity, as well as engaging with all the parties so that they work together

The speakers were joined by Francis Houÿez, Treatment Information and Access Director, EURORDIS, France, for the discussions which followed. In essence, the conclusions of the Session were that: (i) Collaboration is essential between all stakeholders (including patients) and is the best way to create a unified approach to shortage when it arises. (ii) Engaging regulators at all points in the process and seeking a proactive approach to risk assessment (and having plans in place to cover the potential risks) is also a vital element. (iii) Constant levels of communication with all the stakeholders when a situation arises (having regard to legal restrictions that may be necessary) is essential to ensure the most effective management of a shortage.

Session 5 "Understanding of Orphan Therapies Off-Label Uses and Their New Challenges" chaired by André Lhoir, Member of the European Medicines Agency Committee for Orphan Medicinal Products (COMP), Belgium.

Because there are thousands of rare disorders and only a few treatments (79 authorised orphan drugs in Europe at the time of the conference) off-label use is a common practice in the treatment of rare diseases, which companies are not allowed to mention, payers hesitate to reimburse and prescribers are responsible for this off-label use.

Marc Dooms, Senior Orphan Drug Specialist, University Hospitals, Leuven, Belgium, opened the session with a general introduction to the concept of off-label as well as unlicensed use and repurposing. Good off-label use should be limited to life-threatening conditions, standard treatment failure or non-availability and unmet medical need. Collecting clinical evidence is an important issue: maybe a database with positive as well as negative results could be helpful. Greet Musch, Director General Pre-Authorisation, Federal Agency for Medicinal and Health Products, Belgium, represented the Belgian agency point of view, and explored several issues of unmet medical need in the areas of safety and efficacy. She gave an overview of the local actions taken in order to provide patients with new medicinal products as soon as possible, in early collaboration with the reimbursement body, including joint scientific advice, and early reimbursement.

For Yves Juillet, Secretary General, Academy of Medicine Foundation, France, the objective is to allow patients to retain the benefit of off-label used products without having the prescribers put at risk due to the lack of official recognition of the claim and safety issues. Challenges were identified: A complete MA dossier will be difficult to gather; a new specific scheme should be identified allowing products to be agreed upon based on the provision of sufficiently demonstrated evidence; once these products are recognised they should be reimbursed. The risk, if authorities don't go in this direction, is to lose definitively products that can help specific groups of patients who may have no other treatment options. The session closed with questions and answers from a large audience.

Session 6 on "Empowering Patient Advocates in Drug Development" chaired by Nick Sireau, Chairman, AKU Society, and Co-founder of Findacure, had speakers looking at three key issues. Maria Mavris, Director of Therapeutic Development at EURORDIS, discussed drug development education and training initiatives for patients. She presented the EURORDIS Summer School, which takes place in June every year in Barcelona, Spain, and which provides patient advocates with an in-depth and accessible series of lectures and workshops on the regulatory process for orphan drugs. Maria also presented the new European Patients Academy on Therapeutic Innovation (EUPATI) programme that teaches patient groups about clinical trials.

The second speaker, Elena Nicod, is Research Officer at the London School of Economics' Health and Social Policy Department. She gave an overview of how health technology assessment is used to evaluate patient care and looked at models of drug pricing. Few patient advocates have a good understanding of what happens after marketing authorisation, and Elena explained the challenges involved in obtaining reimbursement across European countries.

The third speaker was Tony Hall, Co-founder of Findacure, a new UK charity that helps build the capacity of patient groups. Tony explained how Findacure is running a series of workshops to train emerging patient groups in skills such as fundraising and medicines development. He gave the example of the work of the AKU Society, which was instrumental in putting together a consortium to drive the development of the drug nitisinone for AKU.

Nick Sireau concluded by highlighting the crucial role that patients play in medicines development and why they need to be at the centre of the process.

THEME 5 | EMERGING CONCEPTS AND FUTURE POLICIES FOR RARE DISEASE THERAPIES

Exploring practical policies being developed to facilitate access to treatments for rare diseases, in Theme 5 participants heard the perspectives of a range of stakeholders, including patient representatives, regulators, HTA bodies, payers and industry. Exchanges of views took place on how early dialogue is working in practice and how a continuous dialogue throughout the life-cycle of a drug can be ensured; what level of cooperation between the EMA and the FDA is possible or desirable to speed up drug development; how the voice of the patient can be heard throughout all aspects of the processes; whether the best use is made of the current regulatory framework; and whether patient access can be improved through increased collaboration at all stages. To conclude, the theme examined the external influencing factors that can either support or threaten the continued availability of rare disease treatments and what the future holds in terms of potential alternatives in the development and availability of therapeutic interventions.

Session 1, "Early dialogue and Horizon Scanning of product development to address unmet medical needs", was chaired by Paolo Siviero, Chair, MEDEV and Head of Economic Strategy & Pharmaceutical Policy, AIFA, Italy. In the last few years, Health Technology Assessment (HTA) bodies have taken a new approach toward Scientific Advice (early dialogue) activities. Scientific Advice is a tool for regulators and medicines developers to increase the chance of successful development and, therefore, the availability of high-quality effective and safe medicines for the benefit of patients.

Horizon Scanning is the systematic examination of potential threats, opportunities and potential future developments in order to identify treatments likely to become available that may have significant implications for clinical practice, service design, finance and economic sustainability. Scientific Advice offers industries information on the development of new medicines, such as phase III clinical trial design, according to sample size, primary and secondary endpoints, choice of comparators, time duration of treatment, inclusion/exclusion criteria, etc. Horizon Scanning analyses the kinds of medicines that are still arriving on the market. This tool helps payers to be ready in allocating resources on time. Early Dialogue and Horizon Scanning are necessary to reduce uncertainty in the efficacy and safety profile in order to accelerate the process of medicines development, in particular when there are unmet clinical needs. The main key points to address are: (i) Facilitating patient access to orphan medicines for unmet medical needs; (ii) Economic sustainability; (iii) How is this new approach changing the company business model?; and (iv) What is the patients' role?

In the Panel Discussion that followed, participants brought up many pertinent points:
Jan Geissler, European Patients' Academy on Therapeutic Innovation (EUPATI), Germany, described his own disease experience. It is very important to take into consideration the perspective of the patient from the beginning of the development of the medicine. Christine Mayer-Nicolai, Merck KgaA, Germany, pointed out that most failures in drug development are due to imperfect study design and the lack of expected results. Early Dialogue is needed to determine the best design for the development of new drugs. Industry acknowledges the unmet medical needs and wonders which direction to take - how to include patient perspectives, as this is one of the main points currently under discussion. There is a need for continuous cooperation between all stakeholders (regulators, HTA bodies, payers, companies and patients). Jordi Llinares Garcia, Head, Product Development Scientific Support Department, European Medicines Agency, made the point that dialogue needs to begin at an early stage in order to foresee upcoming problems. It should involve all stakeholders, including patients, and should contain concrete proposals and include the HTA perspective.

Francois Meyer, Advisor to HAS President, International Affairs, French National Authority for Health, Haute Autorité de Santé (HAS), France, evoked three approaches toward HTA: National HTA scientific advice; Parallel HTA-EMA scientific advice (EMA and HTA bodies); and Multi-HTA scientific advice (cooperative advice from EU HTA bodies). This a voluntary activity of HTA bodies, sponsored by the EU Commission- no fee for companies. The EUnetHTA (28 Member States – total budget € 9.428.550) had 2 pilots in 2012 and 8 in 2013. The outcomes show that it has been a successful experience. Three more pilots are being planned. SEED (Shaping European Early Dialogues for health technologies) is an international project financed by the European Commission for 22 months (October 2013 – August 2015). The SEED Consortium, led by HAS, is composed of 14 European agencies specialised in the field of Health Technology Assessment. The objective of SEED is to reduce the risk of production of data that would be inadequate to support the company's future reimbursement request. Ad Schuurman, Head of the Business Contact Centre and International Affairs of the National Healthcare Institute, Netherlands, explained that payers struggle to allocate limited resources. Due to the increase in costs of new therapies, will payers be able to sustain pharmaceutical expenditure in the long run? Payers' aims include planning responsibility and priority allocation. It is important to find new models for collaboration with companies. If we are clear on what we need, companies know how to respond. There is a need for medium- and long-term planning (at least 3 years) to avoid unwanted surprises at the end of the process. At the same time, we have to speed up the process as we are in an emergency situation, especially when it comes to unmet medical needs.

In Session 2: "How to Shape a Better Framework for Orphan Drug Development: EMA/FDA Collaboration" chaired by Jordi Llinares Garcia, Head of Product Development Scientific Support Department, European Medicines Agency, EU, and Debra Lewis, Deputy Director of Office of Orphan Products Development, FDA, USA, perspectives and case studies to improve collaboration between patients, researchers and regulators to benefit drug development and patient access were described. Improving global collaboration is key to rare disease therapy development. EMA and FDA regulators described collaborations addressing orphan designation and other orphan drug development issues and joint workshops facilitating application submissions. Parallel scientific advice could improve to maximize the programme potential.

Patricia Hurter, Senior Vice President, Global Pharmaceutical Development, Vertex, USA, provided an industry view on successful collaborative approaches on cystic fibrosis therapeutic development to bring cystic fibrosis products to market, including regulatory considerations. Pr Kris DeBoeck, Paediatric Pulmonology, University Gasthuisberg, Belgium, described a master plan to improve outcomes for cystic fibrosis, including understanding and targeting the basic defect and assessing efficacy in vivo. The Clinical Trial Network was stressed to optimise treatment development collaborations with the therapeutic development network. Regulatory issues included the need for a common modern view on outcome measures and regulatory expectations.

Elizabeth Vroom, Parent Project, Netherlands, and Pat Furlong, Parent Project Muscular Dystrophy, USA, provided a joint case study in Duchenne muscular dystrophy collaboration in Europe and the USA including their transatlantic collaboration since 1994. They described collaboration at all levels (e.g., funding, studies, regulation, etc.) triggered by patient representatives. Issues included collaboration needs on outcome measures, natural history, biomarkers, guidelines, etc. They described new approaches from patient representatives to barriers and a Regulatory Strategy, i.e., regular meetings with the regulators, interviews with industry, a white paper – putting patients first, a benefit/risk pilot, and draft guidance. Spiros Vamvakas, Head of Scientific Advice, Human Medicines Special Areas, European Medicines Agency, discussed EMA Scientific Advice, its network and procedures. The audience/panel discussion included how to apply these collaborative approaches to groups earlier in development targeting well-designed natural history studies to establish meaningful endpoints as critical. Experienced groups also mentor new patient organisations in effective collaborative practices.

Session 3, "Progressive Patient Access Schemes and Patient Involvement in Benefit-Risk Assessment", was chaired by Jaroslaw Waligora, Policy Officer, Directorate-General for Health and Consumers, European Commission, EU and included presentations by Luca Pani, Director General, Italian Medicines Agency (AIFA), and Pauline Evers, EGAN (European Genetic Alliances Network/ Dutch Federation of Cancer Patient Organisations), Netherlands. The session started with a presentation of the recent transformation of the knowledge cycle in the therapeutic innovation cycle. Cutting-edge discoveries are challenging the adaptation of the regulation and appraisal processes. Currently, products can be accessed early via conditional approval (15 products in Europe so far) and by registration under exceptional circumstances (28 products in Europe so far). Data cannot be supplied when the indication is very rare, scientific knowledge is insufficient or collecting data is unethical. The third option for early access is via compassionate use, available in some countries. These possibilities are considered as not adequate in responding to the needs of patients and should be replaced by adaptive licensing. The basic principles of adaptive approaches are facilitating early access by approving medicines early, with acknowleged uncertainty about the favorable and unfavorable effects. The Progressive Patient Access Scheme would make fuller use of all sources of information to update regulatory and treatment decisions. The EMA is inviting companies to participate in a pilot project to explore adaptive licensing with products in development. The Progressive Patient Access model requires a significant coordination among stakeholders but is worth being fully explored as a potential solution for medicines approval. The development process of Ataluren to treat Duchenne muscular dystrophy patients was presented as an example of the difficulties specific to the field of rare diseases. Mark Rothera, Chief Commercial Officer, PTC Therapeutics, USA, and Yann Le Cam, Chief Executive Officer, EURORDIS, took part in the panel discussion that followed.

In Session 4, "Mechanism of Coordinated Access (MOCA) and Transparent Value Framework, Managed Entry Agreements", the presentations and subsequent discussions dealt with finding a balance between the issue of access to affordable medicines and the long-term viability of the European pharmaceutical industry. Chaired by Thomas Heynisch, Deputy Head of Unit, Unit Food & Healthcare Industries Biotechnology, DG Enterprise, European Commission, EU, and featuring presentations from Wills Hughes-Wilson, Chief Patient Access Officer & Vice President External Affairs, Sobi, Sweden, and Luca Pani, Director General, Italian Medicines Agency (AIFA), the interactions between different Member States (MS) and European policies and diverging priorities among or even within MS (with regard to the different relevant public authorities/ministries concerned) were evoked. The main objectives of the Orphan Drugs Working Group of the Process of Corporate Responsibility in the Field of Pharmaceuticals were discussed along with its deliverables, i.e., how to improve access to orphan medicines for patients with rare diseases. Two outcomes were referred to in more detail: Recommendations and the Transparent Value Framework (TVF).

The coordinated mechanism between volunteering MS and other relevant parties should support the exchange of information leading to informed decisions on pricing & reimbursement. One major advantage of the ongoing MoCA project is the possibility to keep its structure flexible and informal while speeding up overall access by bringing together the decision-makers in the area of pricing/reimbursement at an early stage, as opposed to a legally binding formalised process.

With regard to the topic of managed entry agreements the efforts of the Working Group established under the Process of Corporate Responsibility was described. The way in which these agreements are designed was elucidated by the Italian experience. Furthermore the pros and cons of this instrument were addressed. In addition, the need to have reliable quality data on which the agreements can be based was highlighted.

Session 5, "Emerging Ideas for Sustainable Access to Orphan Medicinal Products" was chaired by Kerstin Westermark, Senior Expert, Committee for Orphan Medicinal Products (COMP Member), Medical Products Agency, Sweden. What are the environmental threats to the continued sustainable availability of treatment for rare diseases and how can we navigate them? The current situation in Europe was reflected on, particularly the economic crisis adding to the difficulties already faced by patients with rare diseases and unmet medical needs.

The successful EU collaboration in orphan medicinal product designation and authorisation was presented as a model. It was emphasised that marketing authorisation is decided following benefit/risk assessment, i.e., strictly on scientific grounds, while pricing and reimbursement are politically - value - based. However, a common scientific ground (documentation) should be possible to obtain, which could potentially reduce/close the gap between the requirements for EU, MA and national reimbursement. The parallel

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EMA/HTA scientific advice, focusing on the needs for regulators for Benefit-Risk assessment as well as for Health Technology Assessment (HTA), could potentially reduce the need for further questions from HTA bodies and a timely access for patients.

It was acknowledged that the economic crisis has affected Member States differently and that the same price for all countries would not be realistic. A 'differential pricing' system around clusters of countries with similar GDP and average income p.c. (one price per cluster) is currently being tested. The EU could play a supportive role here while respecting national competences. The importance of a transparent, continuing dialogue, building confidence between producers and payers was emphasised whereby new innovative medicines for rare diseases could be rewarded while securing a pricing system that would guarantee access to treatment for patients based on sound economic/value based processes that take into account the diversity of the EU Member States.

Session 6: "Rare Disease Treatments beyond Medicinal Products", was chaired by Lesley Greene, Vice-President, CLIMB, Vice-Chair Committee for Orphan Medicinal Products (COMP), Volunteer Patient Advocate, EURORDIS.

Discussions in this Round Table session were guided by several pre-selected topics, beginning with the distinction between a medicinal product and a medical device. This highlighted the different regulatory requirements, also pointing out how we routinely use devices such as spectacles or walking sticks assessed as "low risk", while a dialysis machine requires a "high risk" assessment. Protective gloves illustrate devices that require no specific skill for use while others require training for patient or carer. Special camps that teach children how to self-infuse in a fun environment were described by haemophilia patient expert Radoslaw Kaczmarek, member of the Steering Committee, European Haemophilia Consortium (EHC), Belgium.
An increased engagement between the Medical Devices regulators, clinicians, industry and consumers with complex and rare diseases would ensure appropriate "userbility" development of devices and more accurate needs assessment.

Compelling examples of the challenges to provide sun blocking devices for patients with Xeroderma Pigmentosum and to heal skin in those with Epidermolysis Bullosa were provided by Dr. Smail Hadj Rabia, Department of Dermatolgy, Hôpital Necker - Enfants Malades, Paris; MAGEC - French Expertise Center on Rare Skin Diseases, Genodermatoses Network, France.

The lack of small protective gloves for children or devices to apply cream to one's own back were simple but good examples of how the proper device can make life simpler. Special software for diagnosis and remote monitoring of health was also discussed. The USA has a Human Use Device system for designation of medical devices intended for rare disease populations. It would be very valuable to include the FDA in discussions with a view to developing a similar system in Europe. This would improve the current ad hoc situation where there are no incentives for the development of medical devices for rare diseases as compared to medicinal products covered by the Orphan Regulation 141/2000.
In conclusion, it was unanimously agreed that a multi-stakeholder platform is needed to develop these issues in a more formal setting, having demonstrated how medical devices play a vital role in allowing the patient improved delivery of their medicinal product, swifter diagnostics, and remote monitoring offers greater independence and an improved quality of life.

THEME 6 | BEYOND MEDICAL CARE

While high quality, accessible clinical care is essential for people with rare diseases, we are far more than just patients, Theme 6 explored other social and other non-medical issues that also impact quality of life and access to full citizenship, identifying innovative solutions and programmes that address these issues and making the case for embedding best practices in these areas in European and national policies and provisions.The approach was participative, with platform speakers and delegates sharing knowledge, experience and aspirations, feeding into EURORDIS' future strategy on specialised social services. The need for National Plans and Centres of Expertise to recognise and include non-medical care was emphasised and the guiding principles and experience gained so far were evaluated. Innovative schemes that may serve as models for promoting independent living were described, together with personal experiences of service users.

In Session 1, "Identifying Specific Social Challenges of Rare Diseases", chaired by Stein Are Aksnes, Leader of the Norwegian Advisory Unit on RareDisorders, Oslo University Hospital, Norway, a presentation by Dorica Dan, President, Romania Prader Willi Association, Romanian National Alliance for Rare Diseases, Romania, gave a comprehensive and experience-based overview of the different relevant aspects of social challenges people living with rare diseases and their families often face. This was followed by a presentation from Simona Bellagambi, UNIAMO, Italy, on how national plans can address these issues. Social services are highlighted in several national plans, while other countries to a lesser degree mention these issues in their plan. The different national plans can be of great inspiration for other countries when establishing, implementing and evaluating their own plans. The experience of France was presented by Christel Nourissier, Alliance Maladies Rares, France, who was involved in creating the

Avril Daly, GRDO, Ireland and Lesley Greene, COMP, UK

French plan. Questions from the audience were taken after each presentation. The session ended with a short discussion on the importance of holistic view on these issues, and how the different disciplines can work together to meet the needs of people with rare diseases and their families.

Session 2, "Different Approaches to the Social Challenges of Rare Diseases: Social Policy", chaired by Helena Kääriäinen, Research Professor, National Institute for Health and Welfare, Finland, approached the social challenges in the lives of individuals with rare diseases and offered practical solutions to resolve them. Patient's experiences must be listened to and used as the starting point when searching for solutions.

Pedro Oliveira, University of Lisbon, introduced the Patient Innovation platform, a nonprofit social network for patients and caregivers to share their innovations. A considerable share of innovations in the field of patient care has started from ideas of individual patients.

Presentations on different approaches to identify the disability and social needs of patients with rare diseases followed. Myriam de Chalendar, Orphanet, France, presented the Orphanet disability projects to add disability information to the Encyclopaedia for patients, create disability factsheets for professionals (15 completed to date) and document the disabilities associated with each rare disease (857 diseases already indexed) with the terms of the International Classification of Functioning in order to build a database. Data is collected from medical experts, disability professionals and patient organisations. The aims are to increase knowledge on the daily difficulties experienced by patients and help social agencies in distributing appropriate disability compensation measures.

Birthe Holm, Rare Diseases Denmark, presented the Social Profiles project of Rare Diseases Denmark, which has been developed through a state funded project involving rare disease patient societies and government authorities. Profiles exist for 25 rare diagnoses so far. The profiles present the social needs in different phases of the disease and at different ages. They aim at upgrading skills for patient society advisers and providing a virtual tool kit for social and healthcare.

Session 3 "Concrete Solutions to Social Challenges: Essential Tools for the Integration of Rare Diseases into Social Services" was chaired by Lene Jensen, Chief Executive Officer, Rare Diseases Denmark. Although systems for social support differ amongst European countries, rare disease patients' needs for specialised social services are similar. Dorica Dan, President, Romania Prader Willi Association, Romanian National Alliance for Rare Diseases, Romania, presented documents from EUCERD workshops on principles for specialised social services and on training for providers. Both papers and four fact sheets were distributed to the participants.

Following this presentation, four panelists spoke briefly of their own practice and experience as advisors and directors for specialised social services. A number of questions and comments were put forward, touching upon several important issues, such as: (i) The goal of specialised social services – a continuity of care, a matter of quality of life; (ii) How to define specialised social services: respite care is not only a matter of giving a break to the care givers, it is also about giving the patients opportunities to meet others; (iii) A holistic approach for each patient is extremely important; (iv) We must work for access to specialised social services for all rare disease patients throughout Europe; (v) We need to share knowledge and to secure the quality of knowledge shared; (vi) We need to work together across borders when it comes to the training of the providers – to exchange experience and to open up the training programmes across borders; (vii) Undiagnosed rare disease patients suffers from many of the same problems and should, if possible, be included; (viii) Training of caregivers could be organised into two steps: basic and specialised training.

In Session 4 "Can People Living with a Rare Disease Be Independent? Inspiring Personal Stories" chaired by Gabor Pogany, President, Rare Diseases Hungary, three personal accounts described the daily life challenges for people living with rare diseases. The audience was impressed by the different life strategies employed for attaining a more autonomous life.

The creative and innovative solutions, together with the courage of the speakers, strongly affected the audience: participants laughed, cried, and ultimately left the room with a profound admiration for each of the speakers and on a note of hope and courage to deal with their own challenges.

Hanka Meutgeert started the session by introducing the audience to her family and particularly Rick, her 26 year old son who lives with the metabolic disease aspartylglucosaminuria. Rick lives in adapted housing facilities. He has been living partially away from home since he was 12 years old. Hanka shared with the participants the difficulties the family went through during the process of arranging Rick's partial accommodation away from home:
"Parents who need to work leave their children with other carers. Why should it be different for those with rare diseases?" Hanka argued, recalling the difficulty of family and close friends in accepting the partial delegation of Rick's care to the adapted housing services. Hanka finished her presentation by showing a video of Rick and his current and future accommodations, as Rick will move into a new, bigger apartment soon.

Peter Ryan presented what he called the "total war" between his old and new life, in a touching presentation in which he described his struggle after his first symptoms manifested. Peter, now in his mid twenties, was diagnosed with Leber Hereditary Optic Neuropathy (LHON) when he was 19 years old. LHON is a rare genetic disorder with symptoms that usually begin with a sudden loss of central vision. Symptoms struck Peter unexpectedly, leading to a loss of capacities that affected most of his daily routine and obliged him to re-think his professional and social life as well as his passion for sports. Peter went through periods of frustration, isolation, denial and grief until he got to what he described as "rock bottom".

Fortunately, he went? through that phase and ended up starting a "new life", one in which he embraces his current capacities, making the best of them. Peter has recently gone back to practice sports. He is a member of the Para Cycling Development Squad and is going to the 2016 Paralympic Games in Rio.

Krisztina Pogany lives with Williams Syndrome, a rare genetic developmental disorder associated with a cardiac malformation. Supported by Beata Boncz, Krisztina presented the Independent Life Camps for Rare Disease Youth with Intellectual Disabilities, organised in Hungary. The programme is run on a farm, where participants get in contact with nature, animals, including horses, and work in the countryside. Krisztina enthusiastically presented how these camps helped her to gain experience with daily life activities, work, adapted housing, handling money and other "adult life" experiences. All this, while having fun in a "take it easy but seriously" environment. Krisztina concluded by sharing some lessons learned in the camp, proudly concluding that "getting over our fears is not impossible".

The general opinion of the participants was that more sessions like this would be helpful.

Session 5 "Can People Living with a Rare Disease Be Independent? Inspiring Solutions by Providers" was chaired by Christoph Nachtigäller, President, ACHSE (German National Alliance for Chronic Rare Diseases), Germany.

The topic of online psychological support for people living with a rare disease was presented by Alba Ancochea, Spanish Federation of Rare Diseases (FEDER). Living with a rare disease can generate a lot of emotional stress, leading to depression and anxiety and leaving affected people in apathy and isolation. Psychological support reaches out to meet the needs of the patient and family, can re-establish emotional stability, introduce coping strategies to develop habits of self-care, and encourage social networking. As an online offer it is not restricted by geographical matters, dependence or care. It offers immediate, individualised and confidential care. Since more and more people have access to computers and the Internet today, qualified online advisory services can bridge existing gaps in healthcare services.

Ristoro Fantasia was presented by Renza Barbon Galluppi, President of the Italian Federation of Rare Diseases (UNIAMO), Italy). The restaurant "Ristoro Fantasia", and "Apartments Fantasia" are social projects in Venice that support young adults affected by rare diseases who have difficulties finding appropriate training or regular work due to their disabilities. These young people are trained in various chores of the kitchen and restaurant and integrated in accordance with their abilities. The objective is to show that putting disabled people with the right training on the right job at the right time works wonders. The project not only provides a job, it offers young people with disabilities a place to socialise, test abilities, and gain awareness of their autonomy and productive capacity.

Empowerment Weekends for Young Adults with Anorectal Malformations was presented by Annette Lemli, Vice-President, SoMA (patient organisation for people with anorectal malformations). Anorectal malformations are hereditary and most affected children need plastic surgery shortly after birth and professional medical attendance further on. The outcome of the disease depends on early diagnosis followed by proper treatment. Even under good conditions the transition from childhood to young adult poses a challenge. At some stage the affected youngsters have to take responsibility for their disease management. The Empowerment Weekends project offered by SoMA for teenagers (age range 14-25) covers various topics linked to anorectal malformations, offers individual guidance, including the composition of personal medical results and supporting the young adults in accepting and learning to manage their disease. The project seeks to boost participants' self-esteem in order to help them to lead an independent live.

The discussions following the presentations covered many topics: Does the implementation of the projects pose problems? For the first project, online contact is very helpful for patients with certain limitations and a useful tool to start with, but transition to face-to-face contact is preferred for further proceedings. For the second project, funding is essential. For the third project, as it addresses personal matters at a very sensitive phase of life – why would young people want to participate and share with others? By taking care of affected people from an early age, patient organisations are able to establish enough trust and confidence for young people to take part in such programmes. Whether the projects could be applied to other contexts was also discussed. The audience agreed that all projects could be used as "models". The recruiting process for the restaurant was of specific interest, as were possibilities for funding. In Germany, for example, the government offers specific funding for social integration projects.

Session 6, "How Centres of Expertise Could/Should Cooperate with Social Services", chaired by John Dart, Chief Operating Officer, DEBRA International, UK, focused on the key role that Centres of Expertise in rare diseases can play in coordinating and facilitating non-clinical care and support for affected individuals and families. Examples were given from the UK and French experiences, supplemented by perspectives from Spain and Germany in the panel session. The emphasis placed on the inclusion of social care in the guidelines for Centres of Expertise was welcomed.

The way in which social care is organised varies greatly from country to country (and even within a single country) and it is not possible to be overly prescriptive in defining the precise mechanisms used to promote coordination. However, in most countries the challenge lies in the local or regional provision and management of social care services which require Centres of Expertise to interact with multiple partners. In most cases, people living with a rare disease will use generalist care provision, rather than services specifically for their condition.

The inclusion of staff within the Centre of Expertise team with particular responsibility for supporting patients to access social care within their own locality was seen as particularly important. These case managers/social workers can both make the expertise of the centre available to local care providers and support patients in advocating for adaptations to services tailored to their needs.

Josep Torrent i Farnell, Autonomous University Barcelona, Spain and Pauline Evers, EGAN (European Genetic Alliances Network), Dutch Federation of Cancer Patient Organisations, The Netherlands.

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EXHIBITING COMPANIES

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We provide NGS-based genetic diagnostics of rare diseases. Our patented targeted sequencing method, OS-Seq™, enables us to provide high quality service with competitive prices. Within 21 days, we provide a full service with sequencing, bioinformatic analysis, Sanger confirmation and a comprehensive statement made by our geneticists and clinicians.

CENTOGENE
CENTOGENE is a leading laboratory in genetic testing for rare disorders. We support medical professionals and patients worldwide with advanced genetic testing services, providing high quality reports to make the right treatment decisions. We have implemented a prestigious quality control scheme, holding multiple accreditations (ISO, CAP, CLIA).

COMRADIS
Comradis is a global healthcare communications agency dedicated to rare diseases. Comradis, part of AMICULUM, helps stakeholders to raise disease awareness, develop and market new therapies and improve access to treatments for rare diseases. We deliver effective global or regional communication and commercialization strategies.

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Coté Orphan Consulting specializes in orphan regulatory affairs, risk evaluation, and streamlined project management for emerging treatments for the world's rarest diseases. Our experienced team can guide you through the orphan product development process, from conception to trials to the marketplace. Regardless of company size and capabilities, we bolster your in-house regulatory capacity to help you comply with FDA regulations.

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DNA Genotek provides high-quality biological sample collection, stabilization and preparation products for human genetics, microbiology and animal genetics. The company's products protect and stabilize multiple sample types for long-term storage at ambient temperature to ensure the highest quality results for genetic analysis and testing. The products' reliability and ease-of-use have resulted in rapid adoption by thousands of academic, biotechnology, diagnostic, agriculture, and other leading institutions around the globe.

INTERMUNE
InterMune is a biotechnology company focused on the research, development and commercialization of innovative therapies in pulmonology and orphan fibrotic diseases. In pulmonology, the company is focused on therapies for the treatment of idiopathic pulmonary fibrosis (IPF), a progressive, irreversible, unpredictable and ultimately fatal lung disease. Pirfenidone is approved for marketing by InterMune in the EU and Canada under the trade name Esbriet® and is not approved for marketing in the United States. InterMune's research programs are focused on the discovery of targeted, small-molecule therapeutics and biomarkers to treat and monitor serious pulmonary and fibrotic diseases.

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OPEN APP
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