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

7th European Conference on Rare Diseases & Orphan Products
8-10 May 2014, Andel's Hotel, Berlin, Germany
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.

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Executive Summary

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

With the Support of

In partnership with

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.

ACKNOWLEDGEMENTS AND CREDITS

We wish to thank the following institutions for their active collaboration.

Conference Organiser: EURORDIS

Co-organised by:

With the Support of

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The responsibility of the content and programme of the 7th European Conference on Rare Diseases lies with the speakers and Programme Committee. The Executive Agency is not responsible for any use that may be made of the information contained therein.
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Project Leader of Support, International Rare Diseases Research Consortium (IRDiRC)
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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 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 patent 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 total attendance. 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 and USA gathered a delegation of 53 participants, representing 6% of total attendance.

By country of origin, Germany had the largest delegation of 129 people, representing 16.6% of participants. Mobilisation of German patient organisations via ACHESE, 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 (46 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, Russian and Italian) was undoubtedly an important factor for registration; overall, delegates originating from countries where these languages dominate represented 64% of all delegates.

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.

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 18% in 2012). The proportion of healthcare professionals and policymakers was more or less stable (56%).

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

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 318 participants downloaded the App, 43% of all participants.

Evolution of the total number of participants to the European Conference on Rare Diseases since 2001
### Thursday, 8 May 2014

<table>
<thead>
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<td>14:30 – 17:00</td>
<td><strong>Tutorial 1</strong>&lt;br&gt;HTA 101 for Rare Diseases&lt;br&gt;<strong>Tutorial 2</strong>&lt;br&gt;RD Connect (an integrated platform connecting registries, biobanks and clinical bioinformatics for Rare Disease research)&lt;br&gt;<strong>Tutorial 3</strong>&lt;br&gt;Supporting the pathway to trials for Rare Diseases: clinical trial design and other considerations&lt;br&gt;<strong>Tutorial 4</strong>&lt;br&gt;How to get the best out of Orphanet data</td>
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### Friday, 9 May 2014

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<td>09:00 – 14:00</td>
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<td>14:00 – 15:30</td>
<td><strong>Theme 01</strong>&lt;br&gt;Improving Healthcare Services&lt;br&gt;<strong>Session 0101</strong>&lt;br&gt;Centres of Expertise – Part 1 (models and practical examples)</td>
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<td>16:30 – 18:00</td>
<td><strong>Theme 02</strong>&lt;br&gt;Knowledge Generation and Dissemination&lt;br&gt;<strong>Session 0102</strong>&lt;br&gt;Centres of Expertise – Part II (designation &amp; evaluation)</td>
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<td>09:00 – 10:30</td>
<td><strong>Session 0103</strong>&lt;br&gt;European Reference Networks (ERNs)</td>
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<td>11:30 – 13:00</td>
<td><strong>Session 0104</strong>&lt;br&gt;Addressing the Challenges of Healthcare Pathways</td>
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<td>14:00 – 15:30</td>
<td><strong>Session 0105</strong>&lt;br&gt;Advances in Diagnostic Possibilities for Undiagnosed Patients</td>
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<td>15:45 – 17:15</td>
<td><strong>Session 0106</strong>&lt;br&gt;Improving the Quality of and Access to Diagnostic Services</td>
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### AT A GLANCE

- 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
- 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
- **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

- **Session 0202** A collaborative model to progress knowledge and research
- **Session 0302** Facts on current patient access challenges to orphan products
- **Session 0402** How to shape a better framework for orphan drug development: EMA/FDA collaboration
- **Session 0502** Different approaches to the social challenges of rare diseases: Social Policy

- **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

- **Session 0104** Delivering Help and Support in a virtual world: what will work best?
- **Session 0204** Breakthroughs in Science
- **Session 0304** Shortages in Authorised Medicines for Rare Diseases
- **Session 0404** Mechanism of Coordinated Access (MOCA) and Transparent Value Framework, Managed Entry Agreements
- **Session 0504** Can people living with a rare disease be independent? Inspiring personal stories

- **Session 0105** Knowledge at the point of care: getting the facts just in time or just in case
- **Session 0205** Pre-competitive tools and resources / public-private partnership in the area of rare diseases
- **Session 0305** Understanding of Orphan Therapies Off-Label Uses and Their New Challenges
- **Session 0405** Can people living with a rare disease be independent? Inspiring solutions by providers

- **Session 0106** Hypo, help or harm? The impact of media promotion of rare diseases
- **Session 0206** Whose data is it?: Stimulating Research and Removing Barriers
- **Session 0306** Rare disease treatments beyond medicinal products
- **Session 0406** 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 ORIDO, 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 (ACRE), Germany
Irene Norstedt, Head of Unit, Personalised Medicine, Directorate for Research and Innovation, European Commission, EU
Lesley Greens, Vice-Chair COMP, Vice-President, CLMB, Co-Chair Programme Committee ECRD 2014, UK

PLENARY SESSION

Friday 9 May 2014
10:15 – 12:00
Session Anchor:
Durhane Wong Rieger, President, COMP, ORDO, 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, ORPHA-NET-INSERM, France
Serge Braun, Chief Scientific Officer, APH (Association Française contre Les Myopathies), France
Kata 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 alliance network), Dutch Federation of Cancer Patient Organisations, The Netherlands
Wills Hughes-Wilson, Chair of Task Force for Rare Diseases & Orphan Drugs of Europallie-EBE, Chief Patient Access Officer, Vice President External Affairs, sobs (Swedish Orphan Biovitrum AB), Sweden
Tvetsa Schyns, European Network for Research on Alternating Hemiplegia (ENRAH), Belgium

THEME 1 | IMPROVING HEALTHCARE SERVICES

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

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 Section 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 1 (MODELS AND PRACTICAL EXAMPLES) Interpretation DE + RU

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

This session looks at the interpretation of the concept 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 will 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 bulbosa (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) – Perspectives 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 within the healthcare system / how they interact with local networks
Pierre Sarda, Department de Genétique Médicale, Hôpital Arnaud de Villeneuve, France

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

CENTRES OF EXPERTISE - PART 2 (DEVELOPMENT & 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 Samacks, Coordinator of expert centre on aortic and rare pelvic malformations, Hospital 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 stage, 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.
Session 0105
Saturday 10 May, 14:00 - 15:30

ADVANCES IN DIAGNOSTIC POSSIBILITIES FOR UNDIAGNOSED PATIENTS

Session Chair:
Tijltske 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 clinical care will be provided.

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

UK Example: Deciphering Developmental Disorders (DDD) project and 100,000 Genomes
Wendy Jones, Welcome 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 financially benefit from 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 Tubingen, 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 Macák, Professor, Charles University, Czech Republic

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 Mucopolysaccharides 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 posed by such an approach. Setting the scene – where are we 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 Hardills for Therapy Development and Ongoing Matters in the Field of Orphans
Jeremy Manuel, OBE, Chair European Genetic Alliance, European Gaucher Alliance, UK
Carla Hollik, Professor of Inherited Metabolic Diseases in Adults, Academic Medical Centre, the Netherlands
Michaline Wille, Senior Director Medical Affairs, Shire, Switzerland
László Héljetenko, Head of Scientific Advice, Associate Director, NICE, UK

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

ADRESSING THE CHALLENGES OF HEALTHCARE PATHWAYS Interpretation DE + RU

Session Chair:
Sabine Samacki, 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 Samacki, 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 Genetics, MRC Centre for Neuromuscular Diseases, Institute of Genetic Medicine, International Centre for Life, Newcastle Upon Tyne Hospital, UK
Helena Kääbräinen, Research Professor, National Institute for Health and Welfare, Finland
Tsveta Schyns, Coordinator of EuroGentest, Laboratory for Quality and Affordability − Organisation of exome sequencing, an example from Belgium
Tjitske Kleefstra, Clinical Geneticist, Radboud University Nijmegen Medical Centre, the Netherlands

Theda Wessel, Berlin Center for Rare Diseases, Charité – University Medicine Berlin, Germany

Session 0103
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 Aymer, Eminent 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 list of relevant information very quickly and at minimal cost. Experience 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 and Health Information Systems
Stefanie Weber, Head of the Medical Classifications Unit, German Institute of Medical Documentation and Information (DIMDI), 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 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 with the sources and delivers best suit both patient and practitioner.

The Challenges for Helplines, the 116 Number and Why We Are Advocating It
Dorica Dan, Patient Organisation Liaison Officer, Telethon Italia, Blindness, Ireland

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 Isakov, 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 and 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 diseases and the challenges of low numbers of 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 (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 Datari, Patient Organisation Liaison Officer, Telethon Italia, Italy

Theme 3: Research from Discovery to Patients
Kay Parkinson, Chief Executive, Alström Syndrome, UK

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, national and international level which will be reported along with their outcomes during the sessions in this Theme.

Panel Chair:
Ralph Schuster, Chief Executive, Alström Syndrome, UK

Panel Discussion
Daria Julkowska, The E-Rare perspective, EURORDIS, France
Paul Lasko, Chair, International Rare Diseases Research Consortium (IRDiRC), Canada

SHAPING RARE DISEASE RESEARCH POLICY
Interpretation DE + RU

Session Chair:
Sérgio Aymé, Director, 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 there is enough funding to find the identified needs in the field will be discussed in this session.

EU Horizon 2020: Focus on Research
Luca Di Nia, 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 Laslo, 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 Macák, 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
Hans Loechmüller, Chair of Experimental Myology, Institute of Genetic Medicine Newcastle University, UK

Panel Debates: Current progress and ways to utilise synergies between committee activities
**THEMES**

**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 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

Stína 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

Marlène 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

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

**Cost Containment Measures for Medicines in the European Economic Crisis**

Francois Housse, Treatment Information and Access Director, EURORDIS, France

Greek Health Crisis: from austerity to denialism

Alexander Kentikelenis, Research Associate, Department of Social Medicine, University of Cambridge, UK

Panel Discussion: Viewpoint and engagement of different stakeholders involved

Paolo Silviero, 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

Spios 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 European Disease Point of View

Samuel Rigourd, Global Program Regulatory Director, Novartis Pharma, Switzerland

Panel Discussion: Scientific and procedural hurdles for orphans

Lezla Cipjenko, 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.

**Session 0405**

Saturday 10 May, 14:00 - 15:30

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

Session Chair:

André Lhote, Member of Committee for Orphan Medicinal Products (COMP), Portugal

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 Jullet, Secretary General, Academy of Medicine Foundation, France

Off-label Use: Good and bad practices

Marc Dömen, Senior Orphan Drug Specialist, University Hospitals Leuven, Belgium

National Institute of Health (NIH) Guidelines that Include Off-label Use

Great 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 Generation

**Session 0406**

Saturday 10 May, 15:45 - 17:15

**EMPOWERING PATIENT ADVOCATES IN DRUG DEVELOPMENT Interpretation DE + RU**

Session Chair:

Nick Sireau, Chairman, AXU 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 Lavaris, Director Therapeutic Development, EURORDIS, France
THEMES

Understanding and Using Health Technology Assessment to Make a Case for Better Patient Care
Elena Nicoli, 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

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
Paolo Siviero, Head of Economic Strategy & Development Process
London School of Economics, UK
Elena Nicoli, a Case for Better Patient Care
London School of Economics, UK

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

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

Case Study: Duchenne Muscular Dystrophy
The European Perspective: Elisabeth Vroom, Parent Project, the Netherlands
The US: Perspective: Pat Furlong, Parent Project Muscular Dystrophy, USA

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

CEO PATIENTS’ VOICES ON AFRICAN, ASIAN, AND SOUTH AMERICAN PATIENT ACCESS
Interpretation DE + RU

Session Chairs:
Kerstin Westermarck, Senior Expert, Committee for Orphan Medicinal Products (COMP Member), Medical Products Agency, Sweden
Small Hadi-Rabia, Department of Dermatology, Hôpital Necker - Enfants Malades, France

President
John Wilkinson OBE, Director of Devices, Medicines and Healthcare Social Securities Association, Austria

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

Panel 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

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 Support 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

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
Rit de Riddler, 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 Chairs:
Kerstin Westermarck, Senior Expert, Committee for Orphan Medicinal Products (COMP Member), Medical Products Agency, Sweden

A round table discussion
Round table discussion
Radostow 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 Westermarck, Senior Expert, Committee for Orphan Medicinal Products (COMP Member), Medical Products Agency, Sweden
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Session 0506
Saturday 10 May, 15:45 - 17:15

RARE DISEASE TREATMENTS BEYOND MEDICINAL PRODUCTS
**Session 0601**
**Friday 9 May, 14:00 - 15:30**
**THEME 6 | BEYOND MEDICAL CARE**

**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 Bellagamba, UNIAMO, Italy
- Case Study: The experience of France
- Christel Nourissier, Alliance Maladies Rares, France

**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 Höidebeck-Stuntebeck, Charitable Foundation for People with Disabilities, Prader-Willi Syndrome, Germany
- Lisen Julie Mohr, Froimbu, Norway
- Anders Olason, Ágrenska, Sweden

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

**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 Meijer, Mother of a young adult, living with a degenerative rare disease, the Netherlands

**My Story**

Piotr Ryan, Fighting Blindness, Dublin, Ireland

**Let to Fly – Independent Life camps for rare disease youth with intellectual disabilities**

Beata Boncz, Hungarian Williams Syndrome Association, Hungary

**Krisztina Pogany, Living with Williams Syndrome, Hungary**

**Panel Discussion and Lively Debate**

- Francesco Falai, CIBERER, Spain
- Annette Gritter-Kisslich, Medical Director of the Charité Center for Women and Child Health and Human Genetics, Germany
The 7th European Conference on Rare Diseases and Orphan Products (ECRD 2014 Berlin) was opened by EURORDIS Vice President, Annette Widmann-Mauz. The event, which has grown since the first ECRD was held in 2001 in Copenhagen, is the largest of its kind. 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 (Drug Information Association), and in close collaboration with the European Commission, the European Medicines Agency (EMA), the European Federation of National Organisations for Rare Diseases (NORD), the Canadian Organization for Rare Disorders (CORD), the German National Alliance for Chronic Rare Diseases (ACHRE, CORPHANET), the European Society for Human Genetic & ESHG, the industry through the EBE-Europolio, as well as members of the Programme Committee.

Ms Daly reminded the audience that the ECRD provides a multi-national and multi-stakeholder platform for 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 these conditions.

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, another is the active participation of patients in a treatment in their lifetime; hence, ECRD 2014 Berlin will equally focus on a patient-centred 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 the Regions’ summer initiative to mobilise representatives of the ECRD advocated within the Committee on behalf of the rare disease community. The EURORDIS advocated for action on rare diseases: one of EURORDIS’ recommendations is to provide
delicated funding to support National Rare Disease 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 implementation of the EU Directive 2009/22/EC – the ‘EORTD’s, the European Organisation for Rare Diseases’ (EORTD, the ‘EORTD’s)’). By supporting and coordinating the organisation of 20 National Conferences in the EU and in 4 European countries. They included recommendations on: Quality Criteria and Best Practices for Centres of Expertise for Rare Diseases in Member States; Improving Informed Decisions based on the Clinical Added Value of Orphan Medicinal Products (CASPOM) Information Flow; European Reference Networks for Rare Diseases; Rare Disease Progress in the FP6 and FP7 Framework Programmes; Rare Disease National Plans/Strategies, and Opinion on Potential Areas for European Collaboration in the Field of Newborn Screening.

Advisement 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, 2014–2020, which aims to support the research and development of new, innovative medicines and medical devices, as well as rare disease research within the European Union (EU).

Ms Daly concluded 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 level. 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; national and international federations, 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 Europe.

Within the EU, the new Research Framework Programme (FP7) has established a number 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 about 40% of the scientific discussions at these meetings, as part of the assignment of the committees 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).

The Drug Information, Transparency and Access task force (DITA) is a number of volunteer patient representatives of EURORDIS. ECRD Summer School, advising patient representatives in EMA Committees and international consortia, has continued in 2014. In collaboration with 200+ representatives from 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. EURORDIS has also expanded its collection of online e-learning tools while patients’ involvement has continued in the EMA’s DITA and School, advising patient representatives in EMA Committees and international consortia, has continued in 2014. In collaboration with

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Lesley Greene, Vice-Chair of the EMA Committee for Orphan Products, spoke for the Rare Disease Patient Communities, noting that the Rare Disease Puzzle is a big challenge, and that it is one that requires the input of all stakeholders to solve.

Speaking on behalf of the rare disease patient communities, Ms Greene recalled how, since the first Rare Disease Day was held in 2008, the rare disease movement started in 1980 when her daughter was diagnosed with cystinosis. At that time there was no specific treatment in place. The medical community, healthcare, research diagnosis, therapy, palliative care and social care, regulatory authority 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 brought together key players and stakeholders to present the latest developments 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 represented basic research, academia, industry, patient advocates, NGOs and the organisation. A platform to improve quality, comparability and usability of rare disease registries has been created in cooperation with the Joint Research Centre (JRC). The platform’s main objectives will be to provide a central access point to registries worldwide, to support the harmonisation of new and existing registries, to develop their interoperability by developing computer protocols and to organise training sessions. The EC hopes that the initiative will lead to a substantial quality improvement in the management of rare diseases across Europe.

Concerning Centres of Expertise, Directive 2011/24/EU on Patients’ Rights in Cross-border Healthcare, that ensured rulings of all Member States are in place, the preparatory report by the European Parliament noted that patient rights to seek treatment in other EU Member States. The Directive clarifies patients’ rights to seek treatment abroad and above all that patients are at the heart of the decision. It is the first time that collaboration between centres is laid out systematically and, as a result of the directive, the EU Commission adopted the Delegated and Implementing Acts that set out the criteria and the conditions for European Reference Networks (ERNs). Centres of Expertise are key to the success of the initiative, as they are considered to be centres of excellence, the hubs of a network.

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.

Katya Bushby, Professor of Neuromuscular Genetics at the Institute of Genetic Medicine of the University of Newcastle, UK, outlined three key challenges for the time ahead: the harmonisation of standards of care; the way expertise, advice and services are supported; and the way services appear in some countries and excluded in others. A “European glue money” is vital to secure "the glue money" for the networks: this is especially important where services are less developed or for areas/diseases that are less developed.

The round table included representatives from the plenary speakers, patient organisations and the European Medicines Agency. The plenary speakers were asked to set out the key challenges the network faced and how they addressed them. The panelists then had the opportunity to discuss the main challenges that remain in the areas of organisation of healthcare, research, and the development and availability of orphan medicinal products in Europe.
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 her experience in mapping centres for their patients, so that officially registered centres do not always correspond to the “real” ones, i.e., those offering the best care. Ms Schyns recalled the experience of AHC (alternating hemiplegia of childhood): when the pathophysiology was discovered, everything changed. The designation process required in that first phase, in which patients were also involved. AHC patients and parents self-organised “on their own 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 medicinal products. Ms Wills-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 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 dance in the same 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 no time to lose. In each global health initiative, the right component is on its way: progress is to be expected in 2016.

Finally, Dr Ayém called for a cultural change that consists of the acknowledgement of the need for academic and industry to work together and to look at things in a different way. In order to speed up the discovery of natural history and pathophysiology of diseases, while it is not possible to set up a single comprehensive network, 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 products development costs can amount to 200 million. It is fundamental to be exhaustive in registration and make sure that the risk of patients being exposed to happens; this self-report, should this happen in a curated and managed way.

IRDIHC objectives imply that we still need some additional 100 OMPS on the market in the next six years. Many developers struggle through the development of new sequencing technologies, the discovery of new variants and genes. If patients are not fully involved in the discovery, the results may be far from the information on the research they are concerned about, and, in many cases, to which they have contributed. It is therefore essential to ensure that any application has an impact on the healthcare and the effective use of therapies. At the first session, Dr Ayém and colleagues then framed the need for an application to have an impact on the healthcare and the effective use of therapies. At the second session, Tsveta Schyns recalled the experience of AHC: (alternating hemiplegia of childhood): when the pathophysiology was discovered, everything changed. The designation process required in that first phase, in which patients were also involved. AHC patients and parents self-organised “on their own 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 medicinal products. Ms Wills-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 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.

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for coordination activities. Inter-regional bodies dedicated to relations with the educational, medical and social sectors will collaborate with 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 services required for the diagnosis and well-being of the patient and their family. Dr Rita Bushby, Institute of Genetic Medicine, Newcastle Upon Tyne Hospital, UK, presented findings of a healthcare pathway organisation for neuromuscular diseases and Theda Wessell (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 specialty reports. The experience of the healthcare pathway referent or complex cases manager of the French national organisation (AFM) is cited and should be ideally extended 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 Tizkia Kleeastra, Clinical Geneticist, Radboud University Medical Centre Nijmegen, Netherlands, examples of novel diagnostic possibilities were presented. Dr Kleeastra 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 geneticist is applied where the whole exome is sequenced in all conditions. This is followed by a HP which is applicable in the diagnostic process of heterogeneous conditions. The diagnostic yield has increased considerably for all conditions. The number of disorders for which whole exome sequencing can be offered is expanding. The paper described 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 and discussed.

Dr Wendy Jones, University of Cambridge, UK, presented data from the 2020 Nijmegen conference about uniparental disomy disorders. Ethical aspects with regard to incidental findings are evaluated and discussed.

In Session 6, chaired by Prof 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. His explained features such as ‘diagnostic routing’, ‘gene core lists’ and proposed a scoring system for NGS tests, which would allow clinicians, 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 NGS. 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 Macak, Jr. Chair of Biological and Medical Genetics, University Hospital Motol, Charles University, Prague, Czech Republic, presented the insights from his GeneClinics database. He presented case studies of inappropriate use of patients’ and (grand)parents’ confidence in the healthcare system and inability to find a disease causing gene after that offer genetic tests directly to primary care physicians and the public. These offers fall into the category of ‘genetic horror’. It is important to apply both a top down (regulatory) and a bottom up (informing patient groups) approach to this issue, which would help to avoid public misuse of genetic testing.

Dr Wendy Jones, University of Cambridge, UK, presented data on the ‘Deciphering Developmental Disorders’ project. Within this project the aim is to establish diagnosis by applying current technologies as array CGH and whole exome sequencing, and systematically looking for the presence of uniparental disomy in all cases. Data from over 10,000 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 system FindDzebra. 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 method for the retrieval of 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 relatively under-studied. In the presentation, the need for specialised search engines with simple 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 FindDzebra search engine is available at http://www.finddzebra.com/.

The new ‘Massive Parallel Sequencing’ or ‘Next Generation Sequencing’ (NGS) tests 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. His explained features such as ‘diagnostic routing’, ‘gene core lists’ and proposed a scoring system for NGS tests, which would allow clinicians, 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 NGS. 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.

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Tine Muenster, Project Manager, OrphanAnaesthesia, Germany, explained how searching for information about anaesthesia for people with rare diseases led to his developing OrphanAnaesthesia - currently with 35 guidelines, published but a plan to increase them to over 100. He highlighted how the internet search for a particular condition brought up hundreds of thousands of web pages but it would take a great deal of time and effort to translate these into knowledge that is not possible in an emergency situation. In the discussion, patient organisation members highlighted how they could disseminate information, 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 situations. The development of apps on smart phones and tablets by staff and patients to present or access information at point of care seems to be the need of the day and expected to be a very fast process. It is 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.

Domenico Taruscio, Director, National Centre for Rare Diseases, Italy, highlighted the importance of health professionals working together, through their peer-regisstration 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 human contact, 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 suffer 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 Fandom CBE, Director, National Health Service, National and Genomics Education Centre, UK, identified that patients and their clinicians need information and knowledge - not only for the management of their illness 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 need to be able to access the necessary medical information and expert advice in clinical situations. However, all participants agreed 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.

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Dr. Gert-Jan van Omme, Department of Human Genetics, Leiden University, Amsterdam, gave a presentation on the importance of in-depth analysis of trial outcomes, ultimately followed by a slower successful phase based on in silico-based phenotype descriptions of human, mouse and zebrafish. Genome Research 2013 paper for further information. Text-mining new, publicly downloadable software called EXOMIZER to explore genome-wide associations for a Ciliopathy Service based on the successful Bardet–Biedl and Nephronphthisis experiences of partnerships in rare diseases were presented. Pro Kata Buzdygh, Institute of Genetic Medicine, Newcastle Upon Tyne Hospital, showed how these are achieved in a non-profit, and patient advocacy in the TREAT-NMD Advisory Committee for Therapies (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 these 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 pre-competitive informatics platform engaging with health systems and patient organisations was presented by Dr. Kym Boycott, Investigator, Children’s Hospital of Eastern Ontario, Canada. This network consists of genes for rare paediatric single-gene disorders. This network seeks to identify 500 rare genes for rare diseases, avoids overlapping gene searches, and provides opportunities to hear how new ways of working can be done in the future. EU.

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 place of the scaling up of the orphans in the European Union is now coming of age and their importance is increasing. Industry, however, seems to have increased since. The point was made that patients’ dissatisfaction with regard to access to new orphan medicines is one of the challenges. The so-called orphan medicines are the medicines which are more focused medicines tailored to the needs of individual patients and more costly. The orphan medicines are more costly than the orphan medicines. The so-called orphan medicines are the medicines which are more costly than the orphan medicines. The so-called orphan medicines are the medicines which are more costly than the orphan medicines.

The theme of the session was “The ENMC European Network for the Coordination of Rare Diseases and Orphan Drugs” and was chaired by Nathalie Seigneuret, Senior Access Director, EURORDIS, France; Paolo Siviero, Head of Access, Association France Orphelines; and Nathalie Seigneuret, Senior Access Director, EURORDIS, France; Paolo Siviero, Head of Access, Association France Orphelines.

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involved in providing Scientific Advice on the development of new medicines. She underlined the usefulness for sponsors to engage with all the parties so that they work together.

The speakers were joined by Francis Houzou, 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 patient representatives, regulators, HTA bodies, payers and industry) in order to evaluate patient care and learn at models of drug pricing. Few patient advocates have a good understanding of what happens after patients are approved for a drug. The process involves patient groups 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 drug development 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 a new drug.

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

**THEME 6: CONCEPTS AND FUTURE POLICIES FOR RARE DISEASE THERAPIES**

Exploiting 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 advocacy is required to get involved in the collaborative stages. To conclude, the theme examined the external influencing factors that can either support or threaten the continued involvement of patient groups in the planning and implementation 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 Paul Coulie, Chief Scientific Officer, Economic Strategic, Pharmacology, Policy, AIFA, Italy. In the last few years, Health Technology Assessment (HTA) bodies have taken a new approach to assessing medicines. Scientific Advice is a tool for regulators and medicines developers to improve the design of the clinical development plan and, therefore, the ability 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 phases of development, expected outcomes, and aims to improve EU regulatory and HTA bodies' understanding of the medicinal products and therapies in their portfolios. This helps payers to be ready in allocating resources on time. Early dialogue and horizon scanning allows the identification of uncertainties in the efficacy and safety profile in order to accelerate the approval process. The presentation focuses on unmet medical needs and revalidation (i.e., re-evaluation of already authorized medicinal products, which are already approved and therefore are no longer considered innovations). The goal is to change the way things are done and, therefore, changing the company business model; and (v) What is the impact of Horizon Scanning on the regulatory landscape?

In the Panel Discussion that followed, participants brought up many policy points. The final speaker was Jan Gelisels, 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 patient involvement is crucial; the concept of off-label use needs to be understood; and there is a need for continuous cooperation between all stakeholders (eg, EMA, patient groups, and HTAs) in order to establish a common understanding. The value of early dialogue needs to begin at an early stage in order to foresee upcoming problems. It should involve all stakeholders, including payers, and should contain concrete proposals and include the HTA perspective.

Francois Meyer, Advisor to HAS President, International Affairs, French National Authority for Health, Haut 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 and commercial from EU HTA bodies). This a voluntary activity of HTA bodies, sponsored by the EU Commission—no fee for companies. The EU HTA (28 Member States – total budget € 9.428.550) had 2 pilots in 2012 and 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 (March 2010 – August 2015). The SEED Consortium, led by HAS, is composed of 14 European agencies specialized in the field of HTA and medicines development. The project was designed to reduce the risk of production of data that would be inadequate for regulatory purposes. The main objective of SEED was to help design and implement a new planning framework that takes into account the need for a medium- and long-term planning (at least 3 years) to avoid unexpected results. It was also known that, at this stage, the time we have to speed up the process as we are in an emergency situation, especially when it comes to unmet medical needs.
The basic approaches to study the indication is very rare, scientific knowledge is insufficient or (28 products in Europe so far). Data cannot be supplied when practices. 

The coordinated mechanism between volunteering MS and other relevant stakeholders should support the exchange of information, leading to informed decisions on pricing & reimbursement. One major outcome of the Orphan Drugs Working Group of the Process of Corporate Responsibility in the Field of Pharmacueticals were discussed along with its deliverables, i.e. how to improve access to orphan medicines, how to support patients and how to improve the exchange of information, attractiveness of the new treatments and the need to have reliable data on which the agreements were based is enhanced.

The success EU collaboration in orphan medicinal product designation and authorisation was presented as a model. It was 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. Sissis Vamvakas, Head of Scientific Advice, Human Medicines Special Areas, European Medicines Agency, discussed EMA Scientific Advice, its network and procedures. Elizabeth Vroom, Parent Project, Netherlands, and Pat Furlong, Parent Project Muscular Dystrophy, USA, provided a joint case study in Duchenne muscular dystrophy collaboration with the therapeutic development network. Regulatory issues included the need for a common modern view on outcome measures and regulatory expectations.

The Session 5, “Emerging Ideas for Sustainable Access to Orphan Medicinal Products” was chaired by Kerstin Westmarkar, Senior Expert, Department of Health Policy, the European Medicines Agency (EMA), Brussels. The session started with a presentation on the recent transformation of the knowledge cycle in the therapeutic innovation cycle. Cutting-edge discoveries are shaping the adaptation of the regulatory and appraisal processes. Currently, products can be developed across EU countries in parallel, leading to more accurate needs assessment. The current situation in Europe was reflected on, particularly the currently accessible 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 importance is not adequately addressed. The need for collecting data is unstructural. The third option for early access is via parallel authorization (PA) or use the innovative medicinal products covered by the Orphan Regulation 141/2000. For rare diseases, we are far more than just patients, Theme 6 discussed the importance of how rare medical conditions 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 deliberations leading to a draft proposal on how to apply these collaborative approaches to groups earlier in development targeting new natural history studies to establish meaningful endpoints as critical. Experienced groups also mentor new patient organisations in effective collaborative practices.

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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 Kårörn, Research Professor, National Institute for Health and Welfare, Finland, discussed the social challenges in the lives of individuals with rare diseases and offered practical solutions to resolve them. Several case studies were presented 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 case studies and innovative tools in the field of patient care that has started from ideas of individual patients.

Presentations on different approaches to identify the disability and social needs of patients with rare diseases followed. Myram 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 aim is 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 in which rare disease patient organisations and government authorities. Profiles exist for 25 rare diseases and patients can present themselves in different phases of the disease and at different ages. They aim at upgrading skills for patient society advisors 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 Ireland. The session focused on concrete actions. Presentation by Birthe Holm, Rare Diseases Denmark, presented how both Rare Diseases Denmark and Ireland distributed papers and four fact sheets were distributed to European countries, rare disease patients’ needs for specialised compensation measures.

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 progressed over time, and the patient experienced visual problems and difficulties with everyday activities. Peter went through periods of frustration, isolation, denial and eventually acceptance of his condition. 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 European country’s most affected children need plastic surgery shortly after birth.

Kristina Pogány lives with Williams Syndrome, a rare genetic developmental disorder associated with a cardiac malformation. Supplementary care and support for both children and adults was provided by the Centre of Expertise for Williams Syndrome in Hungary. The project aims to improve children’s quality of life, to reduce barriers and ensure that all children with Williams Syndrome get in contact with nature, animals, including horses, and work in the countryside. Kristina enthusiastically presented how she has been able to improve her quality of life through family and community involvement, with daily life activities, work, adapted housing, handling money and other “adult-like” skills. “I think that it’s about being ‘at home’ and feeling self-confident.” Kristina 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 4 “Can People Living with a Rare Disease Be Independent? Inspiration Solutions by Providers” was chaired by Christopher Hall, Rare Disease Organisation (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 Ancócechea, Spanish Federation of Cancer Patients (FEPAC), Spain. Psychological support reaches out to meet the needs of the patient and family. Can re-establish emotional stability, introduce coping strategies to reduce depression and anxiety, and encourage social inclusion. 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 Barbolini Gallup, President of the Italian Federation of Rare Diseases (UNAMO), 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 it, offers young people with disabilities a place to socialise, test abilities, and gain awareness of their autonomy and productive capacity.

Empowerment Workshops for Young Adults with Anorectal Malformations was presented by Annette Lamik, Vice-President, SoMA (patient organisation for people with anorectal malformations). Anorectal malformations are hereditary and most affected children need plastic surgery shortly after birth. Young people have to take responsibility for their medical management. The Empowerment Workshops 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 young adult’s acceptance and ability to adapt. The project seeks to boost participants’ self-esteem in order to help them to lead an independent life.

The discussion following the presentations covered many topics on the projects and how they can improve patients’ quality of life. The projects focus on social support, self-esteem and autonomy. In Germany, the project offers specific funding for social integration projects.
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P 1 Haemoglobinopathies in Europe: Health & Migration Policy Perspectives
Patricia Aguilar Martinez, Michael Angastiniotis, Androulla Effthimionou, Beatrice Gubbi, Maria del Mar Miquel Pereira, Roumyana Petrova-Benedict, Joan-Lluís Vives Corrons

P 2 se-atlas: Cartographic Representation of Experts on Rare Diseases
Holger Storf, Tobias Hartz, Wulf Pfleiffer, Kathrin Rommel, Mareike Derks, Elisabeth Nyyonuu, Jörg Schmidtke, Holm Graesen, Mirjam Knoss, Thomas Wagner, Frank Ückert

P 3 EB House Austria and EB-CLINET: A Centre of Expertise (CE) and a model for establishing a European Reference Network (ERN) for Genodermatoses
Gabriela Polia-Guba, Elisabeth Mühringer, Rainer Riedl, Johann W. Bauer, Helmut Hintner

P 4 A correct and timely diagnosis for patients with a suspected rare disease is most important for an adequate disease management. In many patients a correct diagnosis is delayed despite multiple visits at different medical specialists. Information about the diagnostic pathway is still scarce. To better understand and improve the diagnostic pathway sufficient data is required
S. Mendiola, C. Scholl, T. Rabe, M. Stuhmann, M. Engel, C. Ziedler

P 5 The impact of rare diseases on the healthcare system: linking the Veneto Region Rare Diseases Registry with health current statistics
Laura Visiona Dalla Pozza, Michela Biasio, Elsa Tota, Paola Facchin

P 7 The first year of activity of Students’ Scientific Circle on Rare Diseases in Pomeranian Medical University in Szczecin, Poland
Michal Slowiaczyk, Jacek Rudnicki, Marta Sawicka, Anna Walecka

P 8 From Life Stories to the Healthcare System: Narrative Medicine and Rare Diseases
Amalia Egle Gente, Maria De Santis, Carlo Donati, Emanuela Mollo, Agata Polizzi, Domenica Tarascio

P 9 A Standard of Care in Huntington’s Disease
D Rae, A Hamilton, Z Miedzybrodzka on behalf of the EHDN Standards of Care Working Group

P 10 A Survey on the Centres of Expertise for Rare Diseases in Italy
R. Mingarelli, S. Ciampa, R. Ruotolo, M. Di Giacinto, M.O. Bal, F. Baronio, I. Bettocchi, E. Di Ruscio

P 11 The Projects 12 months 12 Therapeutic Groups and Everyone adds in the Community of Madrid

P 12 Only the strong survive
Birthe Byskov Holm, Lena Jansen

P 13 Prenatal therapy in developmental disorders: drug targeting via intra-amniotic injection to treat X-linked hypohidrotic ectodermal dysplasia
Katharina Herrmann, Pascal Schneider, Peter Krieg, AnhThu Dang, Kenneth Huttner, Holm Schneider

P 14 Healthcare transition in rare-generations patients: results from a population-based Registry
Monica Mazzaucato, Cinzia Minichella, Martina Bua, Paola Facchin

P 15 Assessing the potential of a European Reference Network for Neuromuscular Diseases: outcomes of an Italian workshop
Teresinha Evangelista

P 16 Treatment of patients with rare diseases: individual treatments or protocols?
M. Viatte, M. Vizelio, E. Cazzador, E. Ruzzo, E. Di Ruscio

P 17 Clinical Utility Gene Cards and the next-generation sequencing (NGS) database
Anna Dierking, Jörg Schmidtke

P 18 Impact of pulmonary arterial hypertension (PAH) on the lives of patients and carers

P 19 Comparative assessment of family’s experience of patients with Dravet Syndrome on the use of rectal diazepam and buccal midazolam
Nathalie Couque, Nicole Chennay, Rima Nabbout

P 20 The European Huntington’s Disease Network - Young Adults Working Group
Michael Orth, Jamie Leavy, David Drain, Michaela Grein

P 21 ERCURD Joint Action (EJA). WP7: “Quality of care/ Centres of Expertise”
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P 22 A Clinical advisory board for a Rare disease (Prader-Willi Syndrome)
Susanne Blichfeldt, Stense Farholt

P 23 Survey of people affected by rare diseases in Quebec, Canada: their experience from diagnosis to treatment and with their entourage
Gail Ouellette, Brigette Belanger

P 24 Improving healthcare in adult patients with rare diseases in Poland
Joanna Sulikka, Joanna Pera, Magdalena Strach, Janskiew Krzysztof, Izabela Kierczewska, Agnieszka Slowik, Tomasz Grabicki

P 25 Newborn Screening For Inherited Metabolic Diseases: the Network Of Emilia-Romagna Region

P 26 Newborn Screening For Inherited Metabolic Diseases: the Network Of Emilia-Romagna Region

P 27 Understanding the cost of hereditary angioedema in England
M. Hebert, T. Holbrook, A. MacCulloch, A. Mannon

P 28 Quality analysis of healthcare network through patients’ and families’ judgment
Silvia Maraca, Sara Barbieri, Miriam De Lorenzo, Patrizia Benzi, Paola Facchin

P 29 Road to a cure for Dravet syndrome
Ana Mingorance-Le Meur, Marisol Montolio, Julian Ila, Luis Miguel Aras

P 30 Gene Panel Diagnostics for Disorders with Abnormal Bone Mass
Uwe Kornack, Björn Fischer, Ralf Ohaim, Peter Krawitz, Tomasz Ziemotek, Michael Ameling, Stefan Mundlos, Peter N. Robinson

P 31 Establishment of narcolepsy-centres in Germany
Ulf Kaliewitz, Eike Mayer, Herbert Dahmen, Volker Westkicken, Christine Pitzen, Claudia Schmitt

P 32 An appropriate way of organizing services for rare disorders
L. Kula, A. Craxi, S. Zaun, L. Lorenz, L. Mladen, L. Hansgen, J. Krauskopp

P 33 Assessing healthcare utilization and healthcare needs in HD patients in South East health region, Norway
M.R. v. Wallenst, E. Howe, J.C. Frich, N. Andelic

P 34 Genodermatoses Network - Towards A Rare And Severe Skin Diseases European Reference Network sharing experience and knowledge with Mediterranean and Middle-Eastern experts and centres
Christine Bodeime, Johann Bauer

P 35 Understanding the healthcare experiences and needs of people living with Huntington’s Disease (HD): an exploratory study
D Raa, S McCann, Z Miedzybrodzka on behalf of the EHDN Standards of Care Working Group

P 36 Rare professionals for rare diseases
Paigorgiottio Mottotto, Giulia Mariani

P 37 Multidisciplinary clinic for patients with Gorlin syndrome in Italy
Charlotte von der Lippe, Laja Schulz

P 38 Boys with sex chromosome aneuploidies (SCA) compared to a clinical sample
K. Fajerst, K. Makkonen, L. Makkonen

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Rebecca Posthoff, Charlotte Lilja, Olof Danielsson, Cecilia Gunnarsson

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P 42 Setting up strategies: patient inclusion in biobank and genomics research in Europe
Pauline McCormack, Anna Kole

P 43 The Orphanet Rare Diseases Ontology (ORDO): a reference tool integrating clinical and genetic data

P 44 Compassionate use* refers to a manufacturer providing its drug, often for free, to patients on a temporary basis
Hanna Hany
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P 142 Shining a light in the black box of orphan drug pricing
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P 143 Building an orphan drug market database to empower decision making by patient organizations and pharma-biotech companies
Philippe Gorry

P 144 Parallel Submissions for Orphan Designation with the FDA, EMA and MHLW/PMDA
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P 145 Multi-criteria Decision Analysis (MCDA): Testing a proposed MCDA framework for orphan Drugs
C. Schey, M. Connolly

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Velia Maria Lapaduta, Giulia Mariani

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Danica Dorina, Maria Puli

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P 167 A Project to Take the Next Generation of Leaders Nationally and Internationally
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P 168 Funding a Program, the autonomy, and the social integration of the people affected by Alternating Hemiplegia of Childhood (AHC) in Italy, and for the empowerment of their families
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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.