The Rare Disease Puzzle: Bringing the Picture to Life

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

Executive Summary

A conference organised by

EURORDIS
Rare Diseases Europe

Co-organised by

DIA

www.diahomes.org

www.rare-diseases.eu
The European Conferences on Rare Diseases and Orphan Products

- The European Conference on Rare Diseases & Orphan Products is the unique platform/forum across all rare diseases, across all European countries, bringing together all stakeholders - patient representatives, academics, healthcare professionals, industry, payers, regulators and policymakers.

- It is a biennial event, providing the state-of-the-art of the rare disease environment, as well as on monitoring and benchmarking initiatives. It covers research, development of new treatments, healthcare, social care, information, public health and support at European, national and regional levels.

- It is synergistic with national and regional conferences, enhancing efforts of all stakeholders. There is no competition with them, but efforts are complementary, fully respecting initiatives of all.

Find out the latest news about the rare disease community on www.eurordis.org

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Executive Summary - 7th European Conference on Rare Diseases & Orphan Products 08-10 May 2014

ACKNOWLEDGEMENTS AND CREDITS

We wish to thank the following institutions for their active collaboration

Conference Organiser

Co-organised by

EURORDIS

Rare Diseases Europe

With the Support of

In partnership with

Plateforme Maladies

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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.

With the Support of

2016-2014

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.
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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 Barcelona (a 18.4% increase from 2012), including 138 speakers and session chairs. 40 full patient fellowships were awarded. The participants at ECRD 2014 represented 43 countries, including 26 from the EU/EEA. The total number of attendees from the EU countries was 649 (compared to 521 in 2012), corresponding to 82% of the overall attendees. Participants attending from EEA countries corresponded to 82% of the overall number of attendees (629).

Representatives from 15 Eastern European states (Bosnia and Herzegovina, Bulgaria, Croatia, Czech Republic, Georgia, Hungary, Latvia, Republic of Macedonia, Poland, Romania, Russian Federation, Serbia, Slovakia, Slovenia, Ukraine) were present at the conference. In total, participants from Eastern Europe represented 11% of the attendees.

Non-European participation from Australia, Brazil, Canada, India, Israel, Japan, South Africa, Taiwan, and China, as well as a delegation of 55 international participants representing 7% of the total attendance.

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

The other countries with an important presence at the Conference included France (55 participants), Italy (77 participants), Belgium (48 participants), the Netherlands (35 participants), and Switzerland (35 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, Italian, Polish, and Russian) 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 Barcelona differed slightly compared to previous events; the introduction of a new category (medical students and postgraduate trainees) made up 3% of the total figure. There were slightly fewer industry representatives (6% versus 10% in 2012). The proportion of healthcare professionals and policymakers was more or less stable (36%).

The majority of the official Conference partners were represented among the speakers and session chairs, including the DIA, ACHSE, EORD, EnapBio/EBE, EMA/COMP, ESICM, and Orphanet.

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

<table>
<thead>
<tr>
<th>Country</th>
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<tr>
<td>Australia</td>
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<td>Austria</td>
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<td>Belgium</td>
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<td>Brazil</td>
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<td>Romania</td>
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<td>Russian Federation</td>
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<td>Serbia</td>
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<td>South Africa</td>
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<td>United States</td>
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This year we introduced the ECRD Mobile App, enabling participants to access information about 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 353 participants downloaded the App, 43% of all participants.
## Executive Summary - 7th European Conference on Rare Diseases & Orphan Products

### ECRD 2014

#### AT A GLANCE

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<thead>
<tr>
<th>Thursday 8 May 2014</th>
<th>14:30 - 17:00</th>
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<tbody>
<tr>
<td>Tutorial 1</td>
<td>HTA 101 for Rare Diseases</td>
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<tr>
<td>Tutorial 2</td>
<td>RD Connect (an integrated platform connecting registries, biobanks and clinical bioinformatics for Rare Disease research)</td>
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<tr>
<td>Tutorial 3</td>
<td>Supporting the pathway to trials for Rare Diseases: clinical trial design and other considerations</td>
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<td>Tutorial 4</td>
<td>How to get the best out of Orphanet data</td>
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<thead>
<tr>
<th>Friday 9 May 2014</th>
<th>09:00 - 14:00</th>
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<tr>
<td>Opening Session, Plenary Session</td>
<td>Session, Poster Session</td>
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<tr>
<td>Theme 01</td>
<td>Improving Healthcare Services</td>
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<td>Theme 02</td>
<td>Knowledge Generation and Dissemination</td>
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<td>Theme 03</td>
<td>Research from Discovery to Patients</td>
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<td>Theme 04</td>
<td>State of the Art and Innovative Practices in Orphan Products</td>
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<td>Theme 05</td>
<td>Emerging Concepts and Future Policies for Rare Disease Therapies</td>
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<td>Theme 06</td>
<td>Beyond Medical Care</td>
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<th>14:00 – 15:30</th>
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<tbody>
<tr>
<td>Session 0101</td>
<td>Centres of Expertise – Part 1 (models and practical examples)</td>
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<tr>
<td>Session 0201</td>
<td>The role, risks and relevance of registries in shaping therapy development to 2020</td>
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<tr>
<td>Session 0301</td>
<td>Shaping Rare Disease Research Policy</td>
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<td>Session 0401</td>
<td>Current landscape of Policy Development on Orphan Products &amp; Rare Disease Therapies</td>
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<tr>
<td>Session 0501</td>
<td>Early Dialogue and Horizon Scanning of Product development to address unmet medical needs</td>
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<td>Session 0601</td>
<td>Identifying specific social challenges of rare diseases</td>
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<th>16:30 – 18:00</th>
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<tbody>
<tr>
<td>Session 0102</td>
<td>Centres of Expertise – Part II (designation &amp; evaluation)</td>
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<tr>
<td>Session 0202</td>
<td>Session 0202: A collaborative model to progress knowledge and research</td>
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<td>Session 0302</td>
<td>Addressing the Gaps in Research at International Level to Identify Opportunities</td>
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<td>Session 0402</td>
<td>Facts on current patient access challenges to orphan products</td>
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<td>Session 0502</td>
<td>Session 0502: How to shape a better framework for orphan drug development: EMA/FDA collaboration</td>
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<tr>
<td>Session 0602</td>
<td>Different approaches to the social challenges of rare diseases: Social Policy</td>
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<th>Saturday 10 May 2014</th>
<th>09:00 - 10:30</th>
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<tr>
<td>Session 0103</td>
<td>European Reference Networks (ERNs)</td>
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<tr>
<td>Session 0203</td>
<td>Making the invisible visible: The coding of Rare Diseases in Health Information Systems</td>
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<tr>
<td>Session 0303</td>
<td>Incentives to create a favourable eco-system</td>
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<td>Session 0403</td>
<td>EMA / Health Technologies Assessment (HTA) interfacing on rare disease therapies</td>
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<td>Session 0503</td>
<td>Progressive Patient Access Schemes &amp; Patient Involvement in Benefit-Risk Assessment</td>
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<td>Session 0603</td>
<td>Concrete Solutions to Social Challenges: Essential tools for the integration of rare diseases into Social Services</td>
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<td>Session 0104</td>
<td>Addressing the Challenges of Healthcare Pathways</td>
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<tr>
<td>Session 0204</td>
<td>Delivering Help and Support in a virtual world: what will work best?</td>
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<tr>
<td>Session 0304</td>
<td>Breakthroughs in Science</td>
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<td>Session 0404</td>
<td>Shortages in Authorised Medicines for Rare Diseases</td>
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<td>Session 0504</td>
<td>Mechanism of Coordinated Access (MOCA) and Transparent Value Framework, Managed Entry Agreements</td>
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<td>Session 0604</td>
<td>Can people living with a rare disease be independent? Inspiring personal stories</td>
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<tr>
<td>Session 0105</td>
<td>Advances in Diagnostic Possibilities for Undiagnosed Patients</td>
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<tr>
<td>Session 0205</td>
<td>Knowledge at the point of care: getting the facts just in time or just in case</td>
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<tr>
<td>Session 0305</td>
<td>Pre-competitive tools and resources / public-private partnership in the area of rare diseases</td>
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<tr>
<td>Session 0405</td>
<td>Understanding of Orphan Therapies Off-Label Uses and Their New Challenges</td>
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<td>Session 0505</td>
<td>Emerging Ideas for Sustainable Access to Orphan Medicinal Products</td>
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<td>Session 0605</td>
<td>Can people living with a rare disease be independent? Inspiring solutions by providers</td>
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<tr>
<td>Session 0106</td>
<td>Improving the Quality of and Access to Diagnostic Services</td>
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<tr>
<td>Session 0206</td>
<td>Hype, help or harm? The impact of media promotion of rare diseases</td>
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<tr>
<td>Session 0306</td>
<td>Whose data is it?: Stimulating Research and Removing Barriers</td>
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<tr>
<td>Session 0406</td>
<td>Empowering Patient Advocates in Drug Development</td>
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<tr>
<td>Session 0506</td>
<td>Rare disease treatments beyond medicinal products</td>
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<tr>
<td>Session 0606</td>
<td>How Centres of Expertise should/could interface with Social Services</td>
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Friday 9 May 2014
09:00 – 9:45
Welcome address and opening remarks by co-organisers:
Avril Daly, Vice-President EURORDIS, Chair of Rare Diseases
Ireland GRDO, CEO Fighting Blindness, Ireland
Jytte Lyngevi, 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 Health Research at the DG Research & Innovation, European Commission, EU
Lesley Greene, Vice-Chair COMP, Vice-President, CLIMB, Co-Chair Programme Committee ECRD 2014, UK

Friday 9 May 2014
10:15 – 12:00
Session Chair:
Durhane Wong Rieger, President, CORO, Canada
Results of the European Commission implementation report on the Council Recommendation on Rare Diseases
John F. Ryan, Director Public Health, DG Health & Consumers, European Commission, EU

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

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

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

Session 0102
Friday 9 May, 16:30 - 18:00
CENTRES OF EXPERTISE – PART 2
(DESIGNATION & EVALUATION)
Session Chair:
Enrique Terol, Policy Officer, DG SANCQ, 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 anorectal and rare pelvic malformations, Hôpital 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 SANCQ, Health and Consumers Unit, European Commission, EU

Session 0103
Friday 9 May, 09:00 - 10:30
EUROPEAN REFERENCE NETWORKS (ERNs)
Session Chair:
Till Voigtlander, Clinical Institute of Neurology, Medical University of Vienna, Austria
In 2003/2004, the high-level European reflection process on cross-border healthcare initiated, amongst others, a discussion regarding the pan-European establishment and designation of highly specialised medical centres (so-called “Centres of Expertise”) and their interlinking in European Reference Networks (ERN). Ten years later, the preparatory work for this concept has been largely finished with the implementation of the Cross-border Healthcare Directive (in October 2013) and the adoption and entering into force of accompanying legal acts (in May 2014). At the end of this year, the first call for proposals for ERNs will be published. At this transitional state, the session will highlight the current practices in the member states, as well as the details and implementation strategy of the ERN concept, followed by a panel discussion looking at existing experiences with the establishment and management of European networks in the fields of healthcare and research, as well as possible future strategies to ensure sustainability of ERNs once established.

Executive Summary - 7th European Conference on Rare Diseases & Orphan Products | 08-10 May 2014
Session 0105
Saturday 10 May, 14:00 - 15:30

ADVANCES IN DIAGNOSTIC POSSIBILITIES FOR UNDIAGNOSED PATIENTS

Session Chair:
Tijlkie Kiefer, 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
Tijlkie Kiefer, 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
Ois Winther, Associate Professor, DTU Informatics, Technical University of Denmark, Denmark

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

IMPROVING THE QUALITY OF AND ACCESS TO DIAGNOSTIC SERVICES

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

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

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

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

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

THEME 2 | KNOWLEDGE GENERATION AND DISSEMINATION

Session 0201
Friday 9 May, 09:00 - 10:30

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

Session Chair:
Christine Lawry, Chief Executive, Society for Mucopolysacharides, 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 associated with such an approach. Setting the scene – where are we today?

Landscape of Disease Registries in Europe and Challenges at Country Level
Elfrida 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 Hardwars for Therapy Development and Ongoing Matters in the Field of Orphans
Jeremy Manuel, OBE, Chair European Genetic Alliance, European Gaucher Alliance, UK
Carlotta Salti, Professor of Inherited Metabolic Diseases in Adults, Academic Medical Centre, the Netherlands
Michaline Wille, Senior Director Medical Affairs, Shire, Switzerland
Magnus Holmberg, Head of Scientific Advice, Associate Director, NICE, UK

Session 0202
Friday 9 May, 14:00 - 15:30

A COLLABORATIVE MODEL TO PROGRESS KNOWLEDGE AND RESEARCH

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

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

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

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

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

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

Making the Invisible Visible: The Coding of Rare Diseases in Health Information Systems
Interpretation DE + RU

Session Chair:
Ségolène Aymer, Ementus 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. Experiences will be presented during this session.

The clinical spectrum of the diseases should also be recorded with a harmonised nomenclature to ensure the inter-operability of databases.

French and German Agencies’ Experience of Coding Rare Diseases with Orpha Codes in Hospital
Stéfanie 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 help through multiple media. Is there still a place for the traditional phone helpline or are the opportunities offered through social media, the internet and social network sites of greater value and importance? In this session we examine the challenges involved and which sources and deliveries best suit both patient and practitioner.

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

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

Results of the Caller Profile Analysis
Georgi Iakov, 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 emergency situation where the expert may not be involved at the initial assessment. This session examines different approaches to

Overview of RARE-Best Practices EU Project
Domenica Tanuscolo, 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 Anaesthesia, 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 patients and families. It will share perspectives from national alliances, national media and also put forward the merits of a European Year of Rare Diseases in 2019.

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

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

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

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

ADRESSING THE GAPS IN RESEARCH AT INTERNATIONAL LEVEL TO IDENTIFY OPPORTUNITIES
Interpretation DE = RU

Session Chair: Milan Macak, 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.

IRIDIRC road map based on gaps and solutions focusing on three scientific committee areas:
- Facilitating the Diagnosis of Most Rare Diseases 2020: IRIDIRC's path forward
Kym Boycott, Investigator, the Children's Hospital of Eastern Ontario (CHEO), Canada

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

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

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

Session 0303 Friday 10 May, 14:00 - 15:30

SHAPING RARE DISEASE RESEARCH POLICY
Interpretation DE = RU

Session Chair: Ségolène Aymé, Director of Research, Director of International Affairs, ORGANANET, 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 we are closer to the identified needs in the field will be discussed in this session.

EU Horizon 2020: Focus on Research
India Nurrott, 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 Jukowska, e-Rare Coordinator, INSERM, Fondation Maladies Rares, France

International Rare Diseases Research Consortium (IRIDIRC), State of the Art
Paul Lasco, Chair, International Rare Diseases Research Consortium (IRIDIRC), Canada

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

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

BREAKTHROUGHS IN SCIENCE

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

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

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

Use of Animal Models for Exome Prioritisation of Rare Disease Genes
Tijnian Smidte, Wellesse Trust Sanger Institute, UK

Promiss and Status of Exon Skipping in Broad Sense
Gerijn van Ommeren, Leiden University Medical Center, the Netherlands

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

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

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

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

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

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

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

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

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

WHOSE DATA IS IT? STIMULATING RESEARCH AND REMOVING BARRIERS

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

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

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

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

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

Session 0307
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
Stina 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-Brisrat, Quality Assurance, Regulatory Affairs, Head International Regulatory Strategy, Shire, Switzerland

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

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
François Houyvet, Treatment Information and Access Director, EURORDIS, France

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

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

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

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

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

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

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

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

Panel Discussion: Scientific and procedural hurdles for orphans
Lezza Ciupenko, 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 Allience, UK

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

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

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

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

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

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

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

Session Chair:
André Lhoir, Member of Committee for Orphan Medicinal Products (COMP), 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 Domen, 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, AKU Society, UK

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

Education & Training Initiatives in Drug Development for Patients
Marc Hardie, Director Therapeutic Development, EURORDIS, France

Executive Summary - 7th European Conference on Rare Diseases & Orphan Products 08-10 May 2014
Understanding and Using Health Technology Assessment to Make a Case for Better Patient Care

Elena Nicolò, 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

Ad Schuurman, Head of the Business Contact Centre and International Affairs of the National Healthcare Institute, the Netherlands

Jan Geissler, European Patients’ Academy on Therapeutic Innovation (EUPATI), Germany

Christine Mayer-Nicolai, Merck KgaA, Germany

Session 0502

Friday 9 May, 16:30 - 18:00

HOW TO SHAPE A BETTER FRAMEWORK FOR ORPHAN DRUG DEVELOPMENT: EMA/FDA COLLABORATION

Interpretation DE + RU

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

Common or Coordinated Guidelines – Why do we need collaboration and how could it be achieved?

Patricia Hurter, Senior Vice President, Global Pharmaceutical Development, Vertex, USA

How Trans-Atlantic Collaboration can Speed up Efficient Drug Development: Case Study of Fibrosis (CP) Clinical Trial Networks

Kris Da Boeck, Paediatric Pulmonology, University Ghent, Belgium

Case Study: Duchenne Muscular Dystrophy

The European Perspective: Elizabeth Vroom, Parent Project, the Netherlands

The US Perspective: Pat Furlong, Parent Project Muscular Dystrophy, USA

Panel Discussion

Spinos Vamvakas, Head of Scientific Advice, Human Medicines Development, Vertex, USA

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

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

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

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

Are we ready? Is what is missing and what is needed? A regulator’s perspective?

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

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

EMERGING IDEAS FOR SUSTAINABLE ACCESS TO ORPHAN MEDICINAL PRODUCTS

Interpretation DE + RU

Session Chair:
Kerstin Westermark, Senior Expert, Committee for Orphan Medicinal Products (COMP) Membars, European Medicines Agency, Sweden

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

Differential Pricing – A way to demonstrate social solidarity to alleviate the impact of the financial crisis?

Flaminia Macchia, Director European Public Affairs, EURORDIS, Belgium

Licensing, Orphan Status and Reimbursement - Can we harmonize the evidence required?

Brigitte Bloch-Daum, Associate Professor and Deputy Head of the Department of Clinical Pharmacology at the Medical University of Vienna; Austrian Delegate to the COMP, Vice-Chair of the Committee of Orphan Medicinal Products (COMP Member), Volunteer Patient Advocate, EURORDIS, Austria

Panel Discussion

Yann Le Cam, Chief Executive Officer, EURORDIS, France

Jordi Llinares Garcia, Head, Product Development Scientific Support Department, European Medicines Agency, EU

Pauline Evers, EGAN (European genetic alliances network / Dutch Federation of Cancer Patient Organisations), the Netherlands

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

RARE DISEASE TREATMENTS BEYOND MEDICINAL PRODUCTS

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

Advances in technology, design and computer programming all support more effective and targeted medical devices to deliver diagnostics and treatments for patients with rare diseases. These medical devices can play a vital role in optimising health and quality of life, by enabling quicker and more accurate diagnosis. Some medical devices are designed for more targeted, more effective or more convenient delivery of a therapy. This session examines these issues from the point of view of the different stakeholders.

Round table discussion

Radostow Kaczmarek, Member of the EHC Steering Committee, EuropeanHaemophilia Committee (EHC), Belgium

Peter Rutherford, Medical Director – Europe, Middle East and Africa, Baxter Healthcare, Switzerland

John Wilkinson OBE, Director of Devices, Medicines and Healthcare Products Regulatory Agency (MHRA), UK

Kerstin Westermark, Senior Expert, Committee for Orphan Medicinal Products (COMP Membars), Medical Products Agency, Sweden

Small Hadj-Rabia, Department of Dermatology, Hôpital Necker - Enfants Malades, France

Jordi Llinares Garcia, Head, Product Development Scientific Support Department, European Medicines Agency, EU

Paolo Siviero, Head of Economic Strategy and Pharmaceutical Policy, Italian Medicine Agency (AIFA), Italy

Complementing Theme 4, Theme 5 will look to the future at the availability of therapeutic interventions.

Participating in the exchange of views on how early dialogue is working in practice and how we can ensure a continuous dialogue throughout the lifecycle of a drug, what level of cooperation between the EMA and the FDA is possible or desirable to speed up drug development; how we can ensure that the voice of the patient is heard throughout all aspects of the processes; whether we are making the best use of the current regulatory framework; and whether patient access can be improved through increased collaboration at all stages.

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

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

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

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

Early and continuous dialogue: what is it; what does it mean; when do we need it?

Panel Discussion

Jordi Llinares Garcia, Head, Product Development Scientific Support Department, European Medicines Agency, EU

Francois Meyer, Advisor to HAS’ President, International Affairs, French National Authority for Health, Haute Autorité de Santé (HAS), France
### Session 0601
**Friday 9 May, 14:00 - 15:30**

#### IDENTIFYING SPECIFIC SOCIAL CHALLENGES OF RARE DISEASES

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

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

**Current Challenges and Issues**
- Dorica Dan, President, Romania Prader Willi Association, Romanian National Alliance for Rare Diseases, Romania
- How National Plans can Address These Issues
  - Simona Bellagambi, National Alliance for Rare Diseases, Romania

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

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

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

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

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

**Orphanet Disability Projects**
- Myriam de Chalendar, Orphanet, France

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

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

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

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

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

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

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

**Panel Discussion**
- Terry Dignan, Serious Fun Network, Ireland
- Norbert Höidebeck-Stuntebeck, Charitable Foundation for People with Disabilities, Prader-Willi Syndrome, Germany
- Lisen Julie Mohr, Frimbu, Norway
- Anders Olason, Ägrena, Sweden
The 7th European Conference on Rare Diseases and Orphan Products (ECRD 2014) was held in Brussels, Belgium. These conferences are held biennially, and ECRD 2014 was supported by EURORDIS (European Reference Networks). The conference provided a platform for patients, patient organizations, researchers, and funders to share their expertise and experiences.

**SUMMARY OF OPENING SESSION**

Princess Beatrix of the Netherlands opened the conference, and on behalf of the EU Commission and EURORDIS welcomed everyone. She noted that EURORDIS is the European Patient Voice, representing patients to the EU, and that it plays a lead role in the development of the European Reference Networks. She recalled the role of the EU Council Recommendation on an action plan for rare diseases (2006). Since 2006, EURORDIS has been advocating for the adoption of the German National Plan of Action for Rare Diseases, which had been announced by the German Federal Minister of Health.

**The Hand of the European Research Council (ERC)**

Chrispich Nychtigall, President of ACARE (Alliance Chronic Rare Disease Europe), began the opening session with a keynote speech. The ERC keeps the most important network of Research Units for Rare Diseases (E-RARE) and is involved in the development of the ERCs. Since 2006, the ERCs have contributed to the development of the European Reference Networks. The ERCs have identified rare diseases and supported the development of new diagnostic and therapeutic tools.

**The ECRIN-IA Ecosystem**

Mr. Nachtigall focused on the role of the ERCs in the development of the European Reference Networks. He mentioned the importance of the ERCs in the development of the European Reference Networks, as well as the development of the ERCs in the European Union. He highlighted the role of the ERCs in the development of the European Reference Networks, as well as the development of the ERCs in the European Union.

**Europe and the German National Plan of Action**

The German National Plan of Action for Rare Diseases is an example of what can be achieved when governments, patients, and researchers work together. The German National Plan of Action was developed by EURORDIS and the German National Alliance, partner of EURORDIS. The plan was supported by EURORDIS and the German National Alliance, partner of EURORDIS. The plan was supported by EURORDIS and the German National Alliance, partner of EURORDIS.

**The Future of Rare Disease Research**

The future of rare disease research is a focus of the event, which is to ensure access to therapies in a timely manner. The future of rare disease research is a focus of the event, which is to ensure access to therapies in a timely manner. The future of rare disease research is a focus of the event, which is to ensure access to therapies in a timely manner.
The Plenary Session and roundtable discussion were chaired by Durhane Wong-Rieger, President of the Canadian Organization for Rare Disorders (CORD) and one of the speakers for the ECRD 2014 in Berlin, remarking that it offers a tremendous opportunity to take stock of what the EU has accomplished in the field of rare diseases. While the establishment of “orphan drugs” onto the horizon, the EU made rare diseases a reality and the conference states that it has come in a short period of time. Not long ago, stakeholders in the field were invited to a consultation on the need for national plans and strategies and the value of Centres of Expertise. Today programmes and discussions are focused on the implementation of plans.

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

John Ryan, Director for Public Health at the European Commission, DG Health and Consumers (DG SANCO), evoked the Commission Communication of 2008 and the Council Recommendation that followed the year after. Pulling limited resources, the European Commission focused on the recognition and visibility of rare diseases with the aim of supporting policies in Member States and coordination among them. This led to the adoption of a “soft law” on the definition of rare diseases, codification, European Reference Networks (ERNs) and for those healthcare providers joining the networks. The EUCERD adopted an Opinion on the landscape across Europe. The EUCERD adopted an Opinion on the IRDiRC, the codification of rare diseases, the development of European Reference Networks, the identification of e-health solutions in the area of rare diseases, as well as the creation of a platform for rare disease registration, an initiative that will demonstrate leadership at the global level.

Rare Disease Day is an example of effective mobilisation and the Commission would be happy to find a way to support this initiative.

The process of concentration of expertise has been taking place for some time and it is now time to build networks to make sure that patients have a better future for patients, families, and ultimately for all those people who love is affected by a rare disease, their life changes and from that day forward they spend vast amounts of time and energy looking for information, targeted research, care, treatments and cures. The burden of rare diseases, from the traditional healthcare, research diagnosis, therapy, palliative and social care, regulatory affairs and policy. Yet all the pieces still need to be brought together, which explains the theme of ECRD 2014 Berlin:

The Rare Disease Puzzle - Bringing the Picture to Life. ECRD 2014 Berlin aims to present all the pieces of that puzzle in six related healthcare, research diagnosis, therapy, palliative and social care, started in 1980 when her daughter was diagnosed with cystinosis. Speaking on behalf of the rare disease patient communities, Ms Greene also took a moment to remind the audience of how frequently chronic and untreatable rare diseases and the related long-term care needs require a different approach in order for patients to live a better future for patients, families, and ultimately for all those communities who are present and build on the active contributions of all participants in putting those pieces together into a coherent whole.

The 800 stakeholders attending ECRD 2014 Berlin also represent a cross-section of stakeholders and groups for a solution to the same jigsaw. Patients and families are invited to join this search: the day they receive the news that they or someone they love is affected by a rare disease, their life changes and from that day forward they spend vast amounts of time and energy looking for information, targeted research, care, treatments and cures. The burden of rare diseases, from the traditional healthcare, research diagnosis, therapy, palliative and social care, regulatory affairs and policy. Yet all the pieces still need to be brought together, which explains the theme of ECRD 2014 Berlin: Rare Disease Puzzle - Bringing the Picture to Life. ECRD 2014 Berlin aims to present all the pieces of that puzzle in six related healthcare, research diagnosis, therapy, palliative and social care, started in 1980 when her daughter was diagnosed with cystinosis. Speaking on behalf of the rare disease patient communities, Ms Greene also took a moment to remind the audience of how frequently chronic and untreatable rare diseases and the related long-term care needs require a different approach in order for patients to live a better future for patients, families, and ultimately for all those communities who are present and build on the active contributions of all participants in putting those pieces together into a coherent whole.

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There is a strong case for improving care closer to patients: expert centres still need to develop these guidelines, which should take into account input from patients and families. A representative of an Italian patient group shared their experience in mapping centres for their patients, as those officially designated do not always correspond to the “real” ones, i.e., those offering care. In order to make a difference, Pr Bushby stated that patients know where centres are and the task for the years to come is to make the development of guidelines a reality for the whole of Europe.

Serge Braun, Chief Scientific Officer, AFM (Association Française contre Les Myopathies), France, mentioned the close cooperation established between patients and Centres in France that are sometimes involved in clinical research to rethink the economic basis for centres that need to stay as close to patients as possible. The European Reference Networks (ERN) are a good example of all experts in one place instead of travelling across the country.

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

In order to speed up the discovery of natural history and pathophysiology of diseases, while it is not possible to set up registries for all diseases, initiatives for this purpose should be created and dialogue should be made possible. These solutions are expensive however and require specific decisions to be taken.

Finally, Dr Aymé called for a cultural change that consists of the acknowledgement of the need for academics and industry to work together. In isolation, academic research is not as successful as hoped and industry struggles to develop research in all fields. Hence the need for a common culture, as there is no common way: progress is to be expected in 2016.

For the 1st session, chaired by Enrique Terol, Policy Officer, DG SANCO, Health and Consumers Unit, European Commission, was introduced the audience to the criteria which will be applied to select the Centres of Expertise constituting the future ERNs: 6 sets of criteria for networks, 5 sets of general criteria for centres, and 2 sets of specific criteria for networks. The assessment will be performed by an independent body.

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

Two legal Acts were recently published to define what is expected of the ERN and to define the designation procedure. The main criterion to be applied will be: clear and solid eligibility criteria of the network members, a key role for Member States endorsement and approval; voluntary participation and a commitment to the rules; a transparent and efficient network planning organisation across the EU and major pieces of the future puzzle: the ENRAH, the ENR-NET, the ERN and the patient experience. For each centre, a small number of recommendations with specific decisions to be taken are needed to facilitate the transition from care to research.

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

During the second plan, the evaluation process was simplified to include an annual activity report and an external visit after 4 years only if problems are detected.

The 2nd session, chaired by Enriqueta Terol, Policy Officer, DG SANCO, Health and Consumers Unit, European Commission, was devoted to the designation and evaluation processes for Centres of Expertise. The creation of the Centre of Expertise and the main components of the new process were introduced. For each centre, a small number of recommendations have been prepared and an independent technical assessment. The first call for proposals is expected in 2015. As an illustration of what an ERN could be in the future, the experience of the Centre of Expertise for rare diseases of the Limburg (surgical specialty) can be seen as a model to follow, this is unlikely to happen as the ERN have no funding anticipated to support their networking activities, while TREAT-NMD had the necessary budget to carry out its activities.

In Session 4, Pr Sabine Sarnacki, Coordinator of the expert centre on anorectal and rare pelvic malformations, Hôpital Necker Enfants Malades, Paris, France, presented the new organisation of the ENRAH. This is inspired by the documents produced by the Rare Disease Task Force and the EUCERD, and is a strong independent technical assessment. The first call for proposals is expected in 2015. As an illustration of what an ERN could be in the future, the experience of the Centre of Expertise for rare diseases of the Limburg (surgical specialty) can be seen as a model to follow, this is unlikely to happen as the ERN have no funding anticipated to support their networking activities, while TREAT-NMD had the necessary budget to carry out its activities.

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

The definition of a healthcare pathway (HP) was clarified to mean the combination of the multiple medical or paramedical specialists, interdisciplinary care and the planning and well-being of the patient and their family. Pr Kata Bushby, Institute of Genetic Medicine, Newcastle Upon Tyne Hospital, UK, presented and discussed the pathway organisation for neuromuscular diseases and Theda Wessel (Berlin Centre for Rare Diseases, Charité- University Medicine Berlin) reported a simple method they used to simplify the healthcare pathway in their rare disease centre, by obtaining from the hospital direction one single medical file for each patient gathering all specialty reports. The experience of the healthcare pathway referent or complex cases manager of the ‘Trouver un Traitement’ (France) is shared and will be applied to all rare diseases generating complex medico-social situations. The implementation of such a professionalisation of care should be adapted to the rare disease policy and the financial constraints of each country.

In the 5th session, chaired by Dr Tjitske Kleefstra, Clinical Geneticist, Radboud University Medical Center Nijmegen, Netherlands, examples of novel diagnostic possibilities were presented. Dr Kleefstra discussed how implementation of whole exome sequencing (all protein coding regions of the human genome) was introduced in the diagnostic process of heterogeneous disorders as hearing loss, vision loss, movement disorders and intellectual disability in the Department of Human Genetics of Radboud UMC Nijmegen. First a geneticist identified a gene where the whole exome is sequenced in all conditions. This is followed by a platform setting up a diagnostic report. The diagnostic yield has increased considerably for all conditions. The number of disorders for which whole exome sequencing can offer diagnostic certainty is growing and after other 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 well evaluated thus far.

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 (UPD) technologies as array CGH and whole exome sequencing. Pr Milan Macak, Jr. Chair of Biology and Medical Genetics, University Hospital Motol, Charles University, Prague, Czech Republic, presented rare genetic disorders, presented case studies of inappropriate use of patients’ and (grand)parents’ confidence in the healthcare system and implementing some action. It is important to apply both a top down (regulatory) and a bottom up (informing patient groups) approach to this issue, which will improve the genetic testing offer.

Dr Ola Winther, Associate Professor, DTU Informatics, Technical University of Denmark presented the development of the search engine FindZebra. The internet has become a primary information resource about illnesses and treatments for both medical and non-medical users. Standard web search is by far the most common resource about illnesses and treatments for both medical and non-medical users. Standard web search is by far the most common resource about illnesses and treatments for both medical and non-medical users. However, there are important questions to address as: “Can we really benefit from the expertised rare diseases community, in particular patients, if not maintained by industry?” and “How could we best implement these new technologies to share data repositories?” There was a robust panel discussion with members, including Jeremy Manuel, European Gaucher Alliance UK; Carla Hollak, Academic Medical Centre, Amsterdam; Micheline Wille, Shire, Switzerland; and Loeza Osipenko, Nice UK.

The overwhelming conclusion of the workshop was that industry, clinicians, payers, regulators and patient organisations should look for a way to maximise and expand the value of registries, to publicity through media. The theme also examines the relationship between registries, to publicity through media. The theme also examines whether registries better serve the rare disease community, in particular patients, if not maintained by industry?” and “How could we best implement these new technologies to share data repositories?”

Theme 2: Knowledge Generation and Dissemination

Without accurate, effective information on rare diseases, diagnosis and appropriate care are delayed. Without accurate, effective information on rare diseases, diagnosis and appropriate care are delayed. There was a call for the European Medicines Agency’s (EMA) to develop a database of rare diseases to support the EMA’s Committee for Medicinal Products for Human Use (CHMP) applicants in the development of their application. The EMA’s Committee for Medicinal Products for Human Use (CHMP) has always had to address the EMA’s Committee for Medicinal Products for Human Use (CHMP) has always had to address this very important issue, especially with the introduction of novel diagnostic possibilities. The theme also examines the relationship between registries, to publicity through media. The theme also examines the relationship between registries, to publicity through media. The theme also examines whether registries better serve the rare disease community, in particular patients, if not maintained by industry?” and “How could we best implement these new technologies to share data repositories?”

In Session 2, chaired by Stefan Schreck, Head of Unit, Health Informatics and Medical Devices, European Commission, and presentation sessions, posters and panel discussions in this session on the expansion of the ‘onDiag’ programme to collect and exchange data across Europe. The ‘onDiag’ project is an international initiative for the exchange of information on the management of rare diseases. The aim of the project is to create a platform that will allow for the exchange of data on the management of rare diseases. The ‘onDiag’ project is an international initiative for the exchange of information on the management of rare diseases. The aim of the project is to create a platform that will allow for the exchange of data on the management of rare diseases. The ‘onDiag’ project is an international initiative for the exchange of information on the management of rare diseases. The aim of the project is to create a platform that will allow for the exchange of data on the management of rare diseases.

In Session 4, chaired by Lesley Greene, Vice-President, CLIMB, Vice-President, CLIMB, Vice-President, CLIMB, Vice-President, CLIMB, Vice-President, CLIMB, and Voluntary Patient Advocate, EURORDIS, sought to answer the question of if a physician can ask ‘My Disease Will Work Best?’ European Network of Rare Disease HelpLines’ Call to Action. The presentation was aimed to identify challenges and solutions to early identification and effective communication of rare disease diagnoses. The presentation was aimed to identify challenges and solutions to early identification and effective communication of rare disease diagnoses.
Tine Muenter, Project Manager, OrphanAnaesthesia, Germany, explained how searching for information about anaesthesia in the field of rare diseases can contribute 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 understand each one’s “fitness for use” and not possible in an emergency situation. In the discussion, patient organisation members highlighted how they could disseminate information by including website links for guidelines in their literature and websites.

It was noted that the use of technology is changing how people access health information, particularly for immediate clinical care. Yet there are barriers to accessing up-to-date information and there are issues related to the quality of the information available, such as the lack of standardisation.

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

The 5th session, entitled “Knowledge at the point of care: getting the facts just in time or just in case”, chaired by Peter Fandem CBE, Director, National Health Service, National Genetics and Genomics Education Centre, UK, highlighted the importance of rare diseases and their clinicians need information and knowledge - not only for immediate treatment 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 must ensure the patient’s needs can be met at any time, through their pre-registration training, but once in practice immediate access to information and knowledge to deal with unexpected situations is required. Four experts shared their experiences in practice and how they engage Rare Disease service participants engaged fully in the discussions which followed.

Domenico Taruscio, Director, National Centre for Rare Diseases, Italy, highlighted a project to identify the procedures for the European Reference Networks, Senior Manager & Research Project Leader, EURORDIS, Spain, featured presentations from Rijke van den Brink, Dutch National Television, who gave an overview of media coverage in the Netherlands in 2012 after the Dutch navy detected a leak in a facility attempting to store 100,000 live animals for Fabry, Pomaee and Gaucher diseases. Avril Daly, Vice-President, EURORDIS, Chair, Genetic & Rare Disorders Organisation (GRDO), Chief Executive Officer, Fighting Blindness, Ireland, gave an overview of the strategy behind EURORDIS plans to advocate for a European Year for Rare Diseases in 2019. Lastly, Alessia Datari, patient organisation liaison officer with Telethon Italia shared their experience in communicating about rare disease research to the general public and shared some case studies on issues relating to gene therapy and stem cell research.

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

Timo Lijó Larrea, Garcia, EMA, UK, soda Lewis, FDA, USA, Elizabeth Vroom, The Duchenne Foundation Project, The Netherlands, Wil De Boer, University Hamburg, Belgium

Session 6, entitled “Hypha, Help or Harm? The Impact of Media Promotion of Rare Diseases”, chaired by Denis Costello, Coordinator, Rare Disease Collaboration for Rare Disease Research, EURORDIS, France, was an opportunity for researchers and clinicians to highlight the importance of media in promoting research on rare diseases and to train professionals to use them. A particular methodology is particularly important for rare diseases, and to train professionals to how they are developed with consultations. Such a methodology is particularly important for rare diseases, and to train professionals to use them. A particular methodology is particularly important for rare diseases, and to train professionals to how they are developed with consultations. Such a methodology is particularly important for rare diseases, and to train professionals to use them. A particular methodology is particularly important for rare diseases, and to train professionals to how they are developed with consultations. Such a methodology is particularly important for rare diseases, and to train professionals to use them. A particular methodology is particularly important for rare diseases, and to train professionals to how they are developed with consultations. Such a methodology is particularly important for rare diseases, and to train professionals to use them. A particular methodology is particularly important for rare diseases, and to train professionals to how they are developed with consultations. Such a methodology is particularly important for rare diseases, and to train professionals to use them. A particular methodology is particularly important for rare diseases, and to train professionals to how they are developed with consultations. Such a methodology is particularly important for rare diseases, and to train professionals to use them. A particular methodology is particularly important for rare diseases, and to train professionals to how they are developed with consultations.
Dr. Gert-Jan van Omman, Department of Human Genetics, Leiden University, Director, Centre for Medical systems Biology, Director Biobanking and Biomolecular Research Infrastructure, Netherlands, an overview of the status of stem cell research in modelling diseases amongst partners; and transparency with sharing of data and results. However, these collaborations create a new ecosystem by aligning public and private interests and bringing together complementary expertise, and foster the development of new treatments for rare diseases for the benefit of patients.

Session 6 provided opportunities to hear how new ways of working and new technologies help stimulate research for diseases that are often under-researched. This is the case for Duchenne muscular dystrophy. After initial phase 2 success, this innovative treatment strategy had a disappointing phase 3 trial, but the trial seemed to hold more promise than initially considered. The presentation and discussion highlighted the value of in-depth analysis of trial results and the importance to this 2 criteria in phases 1 and 2, and underscored the need for adapted outcome measures in rare diseases and in particular in neuromuscular diseases.

Unfortunately, not enough time was available for the round table discussion that followed addressed issues such as fragmentation, the right connection between local, national and international systems, and building convergence and synergies between the different stakeholders. In Session 4, “Breakthroughs in Science”, chaired by Gert-Jan van Omman, Associate Professor Clinical Molecular Genetics, UK, a number of key presentations featuring successful and promising work were presented.

The presentation of Pr Hans Scheffer, Associate Professor Clinical Molecular Genetics, UK, who outlined his thoughts on the benefits of disease specific work was illustrated by Phil Beales, Professor of Medical Genetics, UCL, UK. Pr Beales stressed the benefits of having the patient perspective and the high interest in designations and the importance of seeking support for the development of the ‘Rare Pediatric Disease Review Voucher’ for drug development. Pr Beales introduced the concept of an ‘Innovation Medicines Initiative’ (IMI) and the point was made that patients’ dissatisfaction with regard to access to and costs of orphan medicines is one of the main reasons why patients are dissatisfied with their treatment. The experience and expertise of the main stakeholders shaping the orphan landscape today.

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involved in providing Scientific Advice on the development of new medicines. She underlined the usefulness for sponsors to engage with the EU bodies, as the EMA recently increased the number of challenger companies involved in the evaluation of more than one patient representative per procedure. Finally, Dr. Samuel Ripigro, Global Program Regulatory Director, Novartis, presented the use of clinical trials and the importance of patient engagement.

Session 5 - Understanding of Orphan Therapies Off-Label Uses and Their New Challenges

Chair: André Lhoir, Member of the Management Board, Agency for Orphan Medicinal Products (COMP), Belgium.

Because there are thousands of rare disorders and only a few treatments (79 authorised orphan drugs in Europe at the time of the conference), off-label usage is a common practice in the treatment of rare diseases, which companies are not allowed to mention, payers hesitate to reimburse and physicians are responsible for this decision. The EMA has set out an agreed priority of the allocation of available treatment.

Marc Dooms, Senior Orphan Drug Specialist, University Hospitals, Leuven, Belgium, opened the session with a general introduction to the concept of off-label use as well as unlicensed use and repurposing. Good clinical use should be limited to life-threatening conditions: standard treatment failure or non-availability and unmet medical needs. The EMA's perspective is that the company and the patient are the ones that should have a database positive as well as negative results could be helpful. Great Musch, Director General Pre-Authorisation, Federal Agency for Medicinal Products, Belgium, described the Belgian agency point of view, and explored several issues of unmet medical need. She also pointed out that registration was a key stage in determining 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 Schuurman, Head of Health Technology Assessment (HTA) bodies; and Multi-HTA scientific advice (cooperative advice from EU HTA bodies). This a voluntary activity of HTA bodies, sponsored by the EU Commission- no fee for companies. The first speaker was Tony Hall, Co-founder of Findacure, a new patient alliance (EGA). She explained how clinicians from around Europe work together in putting together a consortium to drive the development of new orphan medicines. The second speaker, Elena Nicod, is Research Officer at the London School of Economics’ Health and Social Policy Department. She gave an overview of the procedures and the therapeutic areas covered so far. There was a lively discussion amongst session participants about how to simultaneously approach Regulators and HTA bodies during the development of an orphan medicine.

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

Marc Dooms, Senior Orphan Drug Specialist, University Hospitals, Leuven, Belgium, opened the session with a general introduction to the concept of off-label use as well as unlicensed use and repurposing. Good clinical use should be limited to life-threatening conditions: standard treatment failure or non-availability and unmet medical need. The EMA’s perspective is that the company and the patient are the ones that should have a database with positive as well as negative results could be helpful. Great Musch, Director General Pre-Authorisation, Federal Agency for Medicinal Products, Belgium, described the Belgian agency point of view, and explored several issues of unmet medical need. She also pointed out that registration was a key stage in determining 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 Schuurman, Head of Health Technology Assessment (HTA) bodies; and Multi-HTA scientific advice (cooperative advice from EU HTA bodies). This a voluntary activity of HTA bodies, sponsored by the EU Commission- no fee for companies. The first speaker was Tony Hall, Co-founder of Findacure, a new patient alliance (EGA). She explained how clinicians from around Europe work together in putting together a consortium to drive the development of new orphan medicines.

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

Nick Sisau reviewed by highlighting the crucial role that patients play in medicines development and why it is so important to be at the centre of the process.
In Session 2: “How to Shape a Better Framework for Orphan Drug Development: EMA/FDA Collaboration” chaired by Jordi Pujol Ferrusola, European Federation of Pharmaceutical Industries and Associations (EFPIA), Belgium, and Tessa DeYoung, Pharmaceutical und Lebensmittel Industries (P&L), Germany, participants were asked to provide feedback on how to improve collaboration between stakeholders to ensure that patients receive timely access to medicines. The feedback was divided into sessions: "Patient Organizations in Action: The Critical Role of Patient Organizations in the Outcome Chain" and "Regulatory Considerations: What Can Be Done by the EU/US?”

In Session 4: "Clinical Trials in Rare Diseases: A Global Perspective on Harmonisation and Streamlining" chaired by Håkan Carlsson, Secretariat of the Rare Diseases Global Alliance, Sweden, and Patricia Cloonan, Global Head of Clinical Development, Shire, USA, the focus was on the need for global harmonisation in clinical trials. The discussion highlighted the need for streamlined processes to improve patient access to medicines and accelerate drug development.

The coordinated mechanism between volunteered MSs and other relevant stakeholders could support the exchange of information on rare diseases, leading to informed decisions on pricing & reimbursement. One major obstacle is the lack of incentives for early-stage projects, as opposed to a legally binding formalisation process.

In Session 5: "Emerging Ideas for Sustainable Access to Orphan Medicinal Products" chaired by Kerstin Westermark, Senior Expert, the European Medicines Agency (EMA), Sweden, the discussion focused on the need for sustainable access to medicines for rare diseases. The session highlighted the importance of ongoing dialogue with stakeholders to identify innovative solutions and programmes that address the needs of rare disease patients.

In Session 6: "Rare Disease Treatments beyond Medicinal Products" chaired by Leslie Greens, Vice-President, CLIMB, Vice-Chair of Orphan Medicinal Products (COMP), Volunteer Advocates, EURORDIS, the discussion focused on the need for innovative solutions beyond medicinal products, such as devices and digital health technologies, to improve patient access to rare disease treatments.

In Session 7: "Beyond Medical Care" chaired by Janice Wolter, Policy Officer, Directorate-General for Health and Consumers, European Commission, the discussion focused on the need for a comprehensive approach to rare disease care, including social and psychological support, and the role of non-medical care providers in improving patient outcomes.

This session was dedicated to the importance of non-medical care in rare disease management and the need for greater recognition and support for non-medical care providers. The discussion highlighted the need for improved coordination and collaboration between different stakeholders to ensure that patients receive the best possible care.

In Session 8: "The European Medicines Agency's Role in Rare Disease Innovation" chaired by Tomasz Grywacz, Director General, the European Medicines Agency (EMA), the discussion focused on the role of the EMA in fostering innovation in the development of rare disease medicines. The session highlighted the importance of the EMA's commitment to rare disease medicines, including its role in supporting the development of innovative therapies and the need for greater collaboration between the EMA, pharmaceutical companies, and patient organizations to ensure timely access to medicines for rare diseases.

In Session 9: "International Approach to Rare Disease Care" chaired by Martin Hutter, Health Care Delivery, the discussion focused on the need for a coordinated international approach to rare disease care, including the importance of collaboration between different countries and the role of international organizations in supporting rare disease research and development.

In Session 10: "The Role of Stakeholders in the Development of Rare Disease Medicines" chaired by James Lachman, CEO, the discussion focused on the important role of stakeholders in the development of rare disease medicines, including patient organizations, pharmaceutical companies, and regulatory agencies. The session highlighted the need for greater collaboration and coordination between stakeholders to ensure timely access to medicines for rare diseases.
Session 2, “Different Approaches to the Social Challenges of Rare Diseases: Social Policy”, chaired by Helena Käräjänkari, Research Professor, National Institute for Health and Welfare, Finland, presented the social challenges in the lives of individuals with rare diseases and offered practical solutions to resolve them. The panel was concluded and used as the starting point when searching for solutions.

Pedro Oliveira, University of Lisbon, introduced the Patient Innovation platform, a non-profit social network for patients and caregivers to share experiences. Creation of innovations in the field of patient care has started from ideas of individual patients.

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

Birth Holm, Rare Diseases Denmark, presented the Social Profiles project of Rare Diseases Denmark, which has been developed through a state funded project. The project is an innovative social care service for rare disease patients and society, which aims to develop and accompany patients on the journey from diagnosis to adulthood and independence. Social Profiles exist for 25 rare diseases, and the project is now being extended to include 40 or more diseases. The presentations outlined the structure of Social Profiles, including a contact list of local professionals, a personal development plan and life education plan, a self-care plan, a life education plan, and a care plan.

In Session 4 “Can People Living with a Rare Disease Be Independent? Inspiring Solutions by Providers” was chaired by Christoph Münch, Head of the Unit for Rare Diseases (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 Ancorchea, Spanish Federation of Rare Diseases (FEDER), Spain. The presentation highlighted the need for online psychological support for rare disease patients, as living with a rare disease can lead to a lot of emotional stress, leading to depression and anxiety and leaving affected people in apathy and isolation. Psychological support reaches out to meet the needs of the patient and family, can re-establish emotional stability, introduce coping strategies to daily living and encourage self-help. 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 advice services can bridge existing gaps in healthcare services.

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

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

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Peter Ryan presented what he called the “total war” between his old and new life, in a touching presentation in which he described his struggle after his first symptoms manifested. Peter, now in his mid twenties, was diagnosed with Leber Hereditary Optic Neuropathy (LHON) when he was 19 years old. LHON is a rare genetic disorder with symptoms that usually begin with a sudden loss of central vision. Symptoms struck Peter unexpectedly, leading to a loss of capacities that affected most of his daily routine and obliged him to re-think his professional and personal life. Fortunately, he won’t: 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, rare disease patients’ needs for specialised social services are similar. Dorica Dan, President, Romania Prader Willi Alliance, Romanian National Alliance for Rare Diseases, Romania, presented documents from EUCERD workshops on social services are similar. Dorica Dan, President, Romania Prader Willi Alliance, Romanian National Alliance for Rare Diseases, Romania, presented documents from EUCERD workshops on
THEME 1 - IMPROVING HEALTHCARE SERVICES

P 1 Haemoglobinopathies in Europe: Health & Migration Policy Perspectives
Patricia Aguilar Martinez, Michael Angastiniotis, Androulla Ellerthunien, Beatrice Gubba, Maria del Mar Marfà Pereira, Roumyna Petrea-Benedict, Joan-Lluís Vives Corrons

P 2 se-atlas: Cartographic Representation of Experts on Rare Diseases
Holger Storf, Tobias Hartz, Liu Pfaffl, Kathrin Rommel, Mareike Derks, Elisabeth Nyykyngä, Jörg Schmidtke, Holm Groaeser, 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 Polko-Guba, Elisabeth Möhringer, Rainer Riedt, 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. Mundis, C. Schoedl, T. Rabie, 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 Visionà Dalla Pozza, Michela Biasio, Ema Toti, Paola Facchin

P 6 The first year of activity of Students’ Scientific Circle on Rare Diseases in Pomeranian Medical University in Szczecin, Poland
Michał Skoczylas, Jackie Rudnicki, Marcin Sawicki, Anna Walecka

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

P 9 A Standard of Care in Huntington's Disease
D Rae, A Hamilton, Z Miedzybyrdzka 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, D Rae, S McCann, Z Miedzybyrdzka on behalf of the EHDN Standards of Care Working Group

P 11 The Projects 12 months 12 Therapeutic Groups and Everyone adds in the Community of Madrid
Isabel Fernandez, Isabel Motero Alba Ancochea

P 12 Only the strong survive
Birtha Byskov Holm, Lena Haugen, Kjersti Vardbakk

P 13 Rare Disease Centre at Hannover Medical School: experiences of two years work

P 14 Healthcare transition in rare diseases patients: results from a population-based Registry
Monica Mazzocchi, Cinzia Mininella, Martina Bua, Paola Facchin

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

P 16 Treatment of patients with rare diseases: Individual treatments or protocols?
M. Raul, G. Ferla, E. Sroczynska, E. Di Ruzzi

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
Pisana Ferrari, Iain Armstrong, Rino Alighetti, Luke Howard, Henrik Ryffenius, Arlye Fischer, Sandra Lombardo, Sean Studler, Lolo Guillevin

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 Chemaly, Rima Nabbout, Thomas Wagner, Frank Ückert

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

P 21 EUCERD Joint Action (EJA). WP7: “Quality of care/Centres of Expertise”
Africa Vitanuova, Richard Woolley, Virginia Corrochano, Beatrice Gómez, Jordi Molas, José David Barberá, Francesc Palau

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 Ouelflette, Brigitte Belanger

P 24 Improving healthcare in adult patients with rare diseases in Poland
Joanna Sulików, Joanna Pera, Magdalena Strach, Janslaw Krzycki, Izabella Kierkowska, Agnieszka Slowik, Tomasz Grotzicki

P 25 Newborn Screening For Inherited Metabolic Diseases: the Network Of Emilia-Romagna Region
M. Viola, E. De Ruzzi, C. Cassis, G. Basucci, P. Pigliatti, M.O. Bal, F. Barone, I. Bettocchi, E. D. Ruzzi

P 26 Newborn Screening For Inherited Metabolic Diseases: the Network Of Emilia-Romagna Region
M. Viola, E. De Ruzzi, C. Cassis, G. Basucci, P. Pigliatti, M.O. Bal, F. Barone, I. Bettocchi, E. D. Ruzzi

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

P 28 Quality analysis of healthcare network through patients’ and families’ judgment
Silvia Marçal, Sara Barbieri, Miriam De Lorenzi, Paola Facchin

P 29 Road to a cure for Dravet syndrome
Ana Mingorance-Le Meur, Marius Montaldo, Julian Isla, Luis Miguel Aras

P 30 Gene Panel Diagnostics for Disorders with Abnormal Bone Mass
Uwe Kernka, Björn Fischer, Ralf Ohaim, Peter Krawitz, Tomazz Ziemetof, Michael Arling, Stefan Mundlos, Peter N. Robinson

P 31 Establishment of narcolepsy-centres in Germany
Ulf Karthwe, Erbert Mauer, Herbert Dahmen, Volker Westdickenberg, Christine Pitzen, Claudia Schütto

P 32 Assessing healthcare utilization and healthcare needs in HD patients in South East health region, Norway
M. v. Walsem, E. Howe, J.C. Frich, N. Andellic

P 33 The Orphanet Rare Diseases Ontology (ORDO): a reference tool integrating clinical and genetic data
Pauline McCormack, Anna Kole

P 34 Assessing the potential of a European Reference Network for Neuromuscular Diseases: outcomes of an exploratory study
Teresinha Evangelista

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

P 36 Rare professionals for rare diseases
Piergiorgio Mottolino, Giulia Marianni

P 37 Multidisciplinary clinic for patients with Gorlin syndrome in South Korea
E. Cho, D. Chua, J. Song, Y. Yoo

P 38 Boys with sex chromosome aneuploidy (SCA) compared to a clinical sample
K. Færmestad, S. Stokke

P 39 Improving healthcare and social services for patients with Neuro-muscular diseases in the Southeast healthcare region in Sweden
Rebekka Posthor, Charlotte Lilja, Olaf Danielsson, Cecilia Gunnarsson

THEME 2 - KNOWLEDGE GENERATION 1 DISSEMINATION

P 41 Mapping the differences in care for 5000 Spinal Muscular Atrophy patients, a survey of 24 national registries in North America, Australasia and Europe
Catherine L. Blanden, Hanns Lochmuller

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

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 Hyry
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 under the trade name Esbriet® and is not approved for marketing by InterMune in the EU and Canada under the trade name Esbriet® and is not approved for marketing in the United States. InterMune’s research programs are focused on the discovery of targeted, small-molecule therapeutics and biomarkers to treat and monitor serious pulmonary and fibrotic diseases.

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Mapi is the global leader in patient focused research services including Post-Marketing, Registries, Linguistic Validation, Health Economics, Strategic Market Access and through the Mapi Trust; the largest library of Patient Reported Outcomes Assessment tools. Mapi is the only clinical research services provider with patient focused expertise that spans the entire clinical trials continuum from protocol development to Post Marketing value consulting. Visit http://www.mapigroup.com/ for more information about Mapi.

COMETIC

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

COTÉ ORPHAN CONSULTING

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

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

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Reguliance specializes in providing US FDA regulatory consulting services to small- and medium-size firms. Expertise includes: US Agent services for Orphan Drug Designation, Drug Master Files, and IND/NDA/BLA/ANDA applications; and due diligence for licensing and acquisition. We work with our EU affiliates to coordinate US and EMA filings.

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Quintiles is the world’s largest provider of biopharmaceutical development and commercial outsourcing services with a network of more than 27,000 employees conducting business in approximately 100 countries. We have helped develop or commercialize all of the top-50, best-selling drugs on the market. Quintiles applies the breadth and depth of our service offerings along with extensive therapeutic, scientific and analytics expertise to help our customers navigate an increasingly complex healthcare environment as they seek to improve efficiency and effectiveness in the delivery of better healthcare outcomes.

EXHIBITING COMPANIES

BLUEPRINT GENETICS

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

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