

OPENING SESSION

FRIDAY, 9 MAY 2014

09.00 – 09.45 | Rubin Room (Level -1)

Welcome Remarks by co-organisers:

Avril Daly, Vice-President, EURORDIS, Chair Genetic & Rare Disorders Organisation, Chief Executive Officer, Fighting Blindness, Ireland
Jytte Lyngvig, Director, DIA Europe, Middle East & Africa, Switzerland

Keynote Addresses:

Hermann Gröhe, German Federal Minister of Health, Germany
Christoph Nachtigäller, President, German National Alliance for Chronic Rare Diseases (ACHSE), Germany
Irene Norstedt, Head of Unit, Personalised Medicine, Directorate for Health Research at the DG Research & Innovation, European Commission, EU
Lesley Greene, Vice-Chair COMP, Vice-President, CLIMB, Vice-Chair Committee for Orphan Medicinal Products (COMP), Volunteer Patient Advocate, EURORDIS, France

09.45 -10.15 Coffee break

The Opening Session will be simultaneously interpreted from English into 5 languages:



German
French
Spanish
Polish
Russian

PLENARY SESSION

FRIDAY, 9 MAY 2014

10.15 – 12.00 Rubin Room (Level -1)

Session Anchor:

Durhane Wong Rieger, President, CORD, Canada

Results of the European Commission implementation report on the Council Recommendation on Rare Diseases

John F Ryan, Director Public Health, DG Health & Consumers, European Commission, EU

Round table discussion:

The integration of National Plans within European strategies. What is the challenge of the next 5 years in this area?

Participants:

Ségolène Aymé, Director International Affairs Orphanet, Chair Topic Advisory Group on Rare Diseases, World Health Organization, Project Leader of Support, International Rare Diseases Research Consortium (IRDiRC), Editor in Chief, Orphanet Journal of Rare Diseases (OJRD)

Serge Braun, Chief Scientific Officer, AFM (Association Française contre Les Myopathies), France

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

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

Wills Hughes-Wilson, Chair of Task Force RDs & ODs, EuropaBio-EBE, Vice President Global Public Policy & Government Relations, Swedish Orphan Biovitrum (sobi)

Tsveta Schyns, European Network for Research on Alternating Hemiplegia (ENRAH), Belgium

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

Thomas Wagner, Pneumology/Allergology, Hospital of the Johann Wolfgang Goethe University, Germany

12.00 – 13.00 Lunch

13.00 – 14.00 Dedicated Poster Session

The Opening Session will be simultaneously interpreted from English into 5 languages:



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Russian



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THEME 1 | IMPROVING HEALTHCARE SERVICES

Kate Bushby, Professor of Neuromuscular Genetics, MRC Centre for Neuromuscular Diseases, Institute of Genetic Medicine, International Centre for Life, Newcastle Upon Tyne Hospital, UK
Véronique Héon-Klin, The Federal Ministry of Health, Germany

The development of Centres of Expertise and European Reference Networks in the field of rare diseases is encouraged in the Council Recommendation on an Action in the Field of Rare Diseases and in the Directive on the Application of Patients' Rights in Cross-border Healthcare as a mean of organising care for the thousands of heterogeneous rare conditions affecting scattered patient populations across Europe.

The aim is to link these Centres of Expertise through European Reference Networks (ERNs), in order to gather expertise and improve healthcare for rare disease patients. Theme 1 will look at the experience gained so far in implementing these concepts. In Session 1, three good practical examples of different types of Centres of Expertise will provide the backdrop to a discussion on the organisation of these types of centres.

In Session 2, speakers will focus on the quality criteria that should be considered when evaluating such centres. Session 3 will review where we stand with the establishment of European Reference Networks and what we can expect for rare diseases. Session 4 will explore the challenges of establishing seamless care pathways between the treating physician and the centres of expertise.

In Session 5 and 6, the state of the art concerning advances and breakthroughs in diagnostic possibilities such as new generation sequencing will be presented. The development of innovations in this field is very rapid and the challenge is now to ensure that these innovations benefit the people who need them most.

Session 0101
Friday, 9 May, 14:00 – 15:30
Room Saphir 2 & 3



CENTRES OF EXPERTISE - PART 1 (MODELS & PRACTICAL EXAMPLES)
 Interpretation DE + RU

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

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

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

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

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

Session 0102
Friday, 9 May, 16:30 – 18:00
Room Jade

CENTRES OF EXPERTISE - PART 2 (DESIGNATION & EVALUATION)

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

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

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

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

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

Session 0103
Saturday, 10 May, 09:00 - 10:30
Room Jade

EUROPEAN REFERENCE NETWORKS (ERNS)

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

In 2003/2004, the high level European reflection process on cross-border healthcare initiated, amongst others, a discussion regarding the pan-European establishment and designation of highly specialised medical centres (so-called "Centres of Expertise") and their interlinking in European Reference Networks (ERN).

Ten years later, the preparatory work for this concept has been largely finished with the implementation of the Cross-border Healthcare Directive (in October 2013) and the adoption and entering into force of accompanying legal acts (in May 2014). At the end of this year, the first call for proposals for ERNs will be published. At this transitional state, the session will highlight the current practices in the member states, as well as the details and implementation strategy of the ERN concept, followed by a panel discussion looking at existing experiences with the establishment



and management of European networks in the fields of healthcare and research, as well as possible future strategies to ensure sustainability of ERNS once established.

Observatory Study – Building European Reference Networks in healthcare

Willy Palm, Dissemination Development Officer, European Observatory on Health Systems and Policies, Belgium

European Commission Report

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

Round Table: Sustainability – Looking to the future The example of the Austrian Epidermolysis Bullosa (EB) Centre and how it links into a broader network

Gabi Pohla-Gubo, Head of Epidermolysis Bullosa (EB) Academy, General Hospital Salzburg/Salzbürger Landeskrankenhaus (SALK), Paracelsus Medical University Salzburg (PMU), Austria

Past models such as TREAT-Neuromuscular Network (NMD): how this will change moving forward and the funding challenges

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

Session 0104

Saturday, 10 May, 11:30 – 13:00

Room Saphir 2 & 3



ADDRESSING THE CHALLENGES OF HEALTHCARE PATHWAYS
Interpretation DE + RU

Session Chair:

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

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

The Organisation of Healthcare Pathways around the Centres of Expertise: The French model

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

Panel Discussion

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

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

Tsveta Schyns, European Network for Research on Alternating Hemiplegia (ENRAH), Belgium

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

Session 0105

Saturday, 10 May, 14:00 – 15:30

Room Jade

ADVANCES IN DIAGNOSTIC POSSIBILITIES FOR UNDIAGNOSED PATIENTS

Session Chair:

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

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

Sequencing: The Netherlands example

Tjitske Kleefstra, Clinical Geneticist, Radboud University Nijmegen Medical Centre, the Netherlands

UK Example: Deciphering Developmental Disorders (DDD) project and 100,000 Genomes

Wendy Jones, Wellcome Trust Sanger Institute / University of Cambridge, UK

FindZebra – What is currently available as computerised systems for diagnosis, what are their benefits and their limits

Ole Winther, Associate Professor, DTU Informatics, Technical University of Denmark, Denmark

Session 0106

Saturday, 10 May, 15:45 – 17:15

Room Jade

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 the 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 Health Care Systems: Balancing the role of state and private sectors

Milan Macek, Professor, Charles University, Czech Republic

THEME 2 | KNOWLEDGE GENERATION AND DISSEMINATION

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

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

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

The theme also examines the potential challenges and rewards from the perspective of all stakeholders.

Session 0201
Friday, 9 May, 14:00 – 15:30
Room Amethyst

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

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

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

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

Industry Perspective: Advantages and disadvantages of disease registries vs. drug registries
Daniel Rosenberg, Senior Director, Head Epidemiology & Observational Studies, Actelion

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

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

Session 0202
Friday, 9 May, 16:30 – 18:00
Room Amethyst

A COLLABORATIVE MODEL TO PROGRESS KNOWLEDGE AND RESEARCH

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

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

Ispra Platform of Services for Rare Disease Registries
Ciaran Nicholl, Ispra Sector Head, Internal and External Communications Unit, Programmes and Stakeholders Relations Directorate, Joint Research Centre, European Commission, EU

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

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

Session 0203
Saturday, 10 May, 09:00 – 10:30
Room Saphir 1



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

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

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

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



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

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

Session 0204
Saturday, 10 May, 11:30 – 13:00
Room Saphir 1

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

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

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

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

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

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

Session 0205
Saturday, 10 May, 14:00 – 15:30
Room Bernstein

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

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

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

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

Emergency Guidelines and Emergency Cards – Practical experience for clinician and patient
Odile Kremp, Director, Orphanet, France

Saturday Night at a University Hospital – How to handle a patient with a rare disease
Tino Münster, Project Manager, OrphanAnesthesia, 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
Room Bernstein

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

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

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

Success Criteria for Attractive Rare Disease Day Activities
Evanina Morcillo Makow, Spanish Federation of Rare Diseases representative & Chief Executive Officer, DEBRA, Spain

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

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

THEME 3 | RESEARCH FROM DISCOVERY TO PATIENTS

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

Rare disease research is still too fragmented and compartmentalised. This leads to lack of integration, duplication of efforts, lack of critical mass, thinking in “silos” and waste of resources.

It also hinders progress towards better diagnosis and therapy for rare disease patients despite many opportunities offered by new technological developments. To improve the situation and ensure a rapid translation of discoveries into operational diagnostic and therapeutic tools, several initiatives have emerged at local, regional, national and international level which will be reported along with their outcomes during the sessions in this Theme.

Session 0301
Friday, 9 May, 14:00 – 15:30
Room Saphir 1



SHAPING RARE DISEASE RESEARCH POLICY
 Interpretation DE + RU

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

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

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

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

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

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

Session 0302
Friday, 9 May, 16:30 – 18:00
Room Saphir 1



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

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

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

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

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

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

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

Session 0303
Saturday, 10 May, 09:00 – 10:30
Room Opal

INCENTIVES TO CREATE A FAVOURABLE ECO-SYSTEM

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

Win-win situations are possible through partnerships and initiatives between industry, public institutions and charity organisations, which open a new model to develop advanced therapeutic medicinal products.

Examples will be given of successful ecosystems that contributed to the marketing of innovative treatments of rare diseases. This includes successful private fund raising for translational research centres involving all stakeholders; local and transnational bioclusters, as well as new infrastructure models for drug development.

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

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

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

Session 0304
Saturday, 10 May, 11:30 – 13:00
Room Opal

BREAKTHROUGHS IN SCIENCE

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

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

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

Use of Animal Models for Exome Prioritisation of Rare Disease Genes
Damian Smedley, Wellcome Trust Sanger Institute, UK
Promises and Status of Exon Skipping In Broad Sense
Gertjan van Ommen, Leiden University Medical Center, the Netherlands



Session 0305
Saturday, 10 May, 14:00 – 15:30
Room Opal

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

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

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

Supporting the Drug Development Pathway for Rare Diseases – The experience of the-Neuromuscular Network (NMD) advisory committee for therapeutics (TACT)

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

European Bioinformatics Institute (EBI) Initiative

Justin Paschall, Team Leader, Variation, European Bioinformatics Institute, UK

The Experience of a Charity in Translating the Results of Basic Research to Therapies for Patients

Lucia Monaco, Chief Scientific Officer, Fondazione Telethon, Italy

Session 0306
Saturday, 10 May, 15:45 – 17:15
Room Opal

WHOSE DATA IS IT?: STIMULATING RESEARCH AND REMOVING BARRIERS

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

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

Stimulating Research and Monitoring Patients

Phil Beales, Professor of Medical Genetics, UCL, UK

The Responsible Use and Indication Criteria for Next Generation Sequencing Diagnostics in Clinical Practice

Hans Scheffer, Associate Professor Clinical Molecular Genetics, Radboud University Nijmegen Medical Centre, the Netherlands

The EU Data Protection Law Reform and Scientific Research: What's new?

Gauthier Chassang, Lawyer EU and International Law, INSERM, France

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

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

Emmanuelle Lecomte-Brisset, Quality Assurance, Regulatory Affairs, Head International Regulatory Strategy, Shire, Switzerland

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

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

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

Session 0401
Friday, 9 May, 14:00 – 15:30
Room Opal

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

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

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

Current Landscape – Overview by European Medicines Agency

Stiina Aarum, Acting Head of Orphan Medicines, European Medicines Agency, EU

State of Play from the US Perspective

Debra Lewis, Deputy Director of Office of Orphan Products Development, FDA, USA

Development of International Orphan Drug Policies

Emmanuelle Lecomte-Brisset, Quality Assurance, Regulatory Affairs, Head International Regulatory Strategy, Shire, Switzerland

Panel Discussion

Marlene Haffner, Chief Executive Officer, Haffner Associates, USA

Session 0402
Friday, 9 May, 16:30 – 18:00
Room Opal

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 is increasing in times of austerity. Viewpoints and engagement of main stakeholders involved in the process at country and European level will be shared in this session.

Cost Containment Measures for Medicines in the European Economic Crisis

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

Panel Discussion: Viewpoint and engagement of different stakeholders involved

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

Heidi Wagner, Senior Vice President, Global Government Affairs, Alexion Pharmaceuticals, USA

Session 0403

Saturday, 10 May, 09:00 – 10:30

Room Saphir 2 & 3



EMA-HEALTH TECHNOLOGY ASSESSMENT (HTA) INTERFACING ON RARE DISEASE THERAPIES
Interpretation DE + RU

Session Chair:

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

Growing interfacing and early dialogue between the European Medicines Agency (EMA) and Health Technology Assessment (HTA) is an opportunity for developers to receive simultaneous feedback from both sides on their development plans and reduce the time of orphan therapy availability to patients.

During this session, available regulatory options, practical experiences gained so far on parallel protocol assistance with HTA bodies, as well as procedural trends for the future will be presented.

Experience to date on interfacing in rare diseases, status with protocol assistance and where are we going – future directions

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

Experience of the Parallel European Medicines Agency (EMA) Health Technology Assessment-(HTA) Scientific Advice (SA) from an Orphan Disease Point of View

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

Panel Discussion: Scientific and procedural hurdles for orphans

Leeza Osipenko, Senior Scientific Advisor, NICE, UK
Helma Gusseck, Pro-Retina Germany, Germany

Session 0404

Saturday, 10 May, 11:30 – 13:00

Room Jade

SHORTAGES IN AUTHORISED MEDICINES FOR RARE DISEASES

Session Chair:

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

Experience with Fabry Shortage

Carla Hollak, Professor of Inherited Metabolic Diseases in Adults, Academic Medical Center, the Netherlands

Regulatory Perspective

Brendan Cuddy, Scientific Administrator, European Medicines Agency, EU

Genzyme, a Sanofi Company – How a company should/could respond

Carlo Incerti, Senior Vice President, Head Genzyme Global Medical Affairs, Italy

Panel Discussion

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

Session 0405

Saturday, 10 May, 14:00 – 15:30

Room Amethyst

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

Session Chair:

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

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

Challenges Associated with Healthcare System: Reimbursement

Yves Juillet, Secretary General, Academy of Medicine Foundation, France

Off-label Use: Good and bad practices

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

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

Greet Musch, Director General DG PRE – authorisation, Federal Agency for Medicinal and Health Products, Belgium

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

Session 0406

Saturday, 10 May, 15:45 – 17:15

Room Saphir 1



EMPOWERING PATIENT ADVOCATES IN DRUG DEVELOPMENT
Interpretation DE + RU

Session Chair:

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

Education & Training Initiatives in Drug Development for Patients

Maria Mavris, Director Therapeutic Development, EURORDIS, France



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

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

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

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

Complementing Theme 4, Theme 5 will look to the future at what practical policies are being developed to facilitate access to treatments for rare diseases. Participants will hear the perspectives of a range of stakeholders, including patient representatives, regulators, HTA bodies, payers and industry. The theme will include exchanges of views on how early dialogue is working in practice and how we can ensure a continuous dialogue throughout the life-cycle of a drug; what level of cooperation between the EMA and the FDA is possible or desirable to speed up drug development; how we can ensure that the voice of the patient is heard throughout all aspects of the processes; whether we are making the best use of the current regulatory framework; and whether patient access can be improved through increased collaboration at all stages. To conclude, the theme will examine the external influencing factors that can either support or threaten the continued availability of rare disease treatments and what the future holds in terms of potential alternatives in the development and availability of therapeutic interventions.

Session 0501
Friday, 9 May, 14:00 – 15:30
Room Jade

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

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

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

Panel Discussion
Jordi Llinares Garcia, Head, Product Development Scientific Support Department, European Medicines Agency, EU
François Meyer, Advisor to HAS' President, International Affairs, French National Authority for Health, Haute Autorité de Santé (HAS), France
Ad Schuurman, Head of the Business Contact Centre and International Affairs of the National Health Care 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
Room Saphir 2 & 3



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

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

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

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

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

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

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

Session 0503
Saturday, 10 May, 09:00 – 10:30
Room Amethyst

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

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

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

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

Patient View on Progressive Patient Access Schemes

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

Panel Discussion

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

Session 0504

Saturday, 10 May, 11:30 – 13:00

Room Amethyst

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

Session Chair:

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

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

Concepts & MOCA Pilots (feedback from the process around the first pilots)

Wills Hughes-Wilson, Chief Patient Access Officer & Vice President External Affairs, Sobi, Sweden

Managed Entry Agreements

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

Panel Discussion

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

Session 0505

Saturday, 10 May, 14:00 – 15:30

Room Saphir 1

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

Session Chair:

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

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

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

Flaminia Macchia, Director European Public Affairs, EURORDIS, Belgium

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

Brigitte Bloechl-Daum, Associate Professor and Deputy Head of the Department of Clinical Pharmacology at the Medical University of Vienna; Austrian Delegate to the COMP, Vice-Chair of the Committee on Reimbursement of Drugs of the Austrian Social Securities Association, Austria

Panel Discussion

Adam Heathfield, Senior Director, Worldwide Policy, Pfizer, UK

Rembert Elbers, Member of the Committee of Orphan Medicinal Products (COMP), Member of the Scientific Advice Working Party, Former Head of Federal Institute for Drugs and Medical Devices (BfArM) Oncology Unit, Germany

Session 0506

Saturday, 10 May, 15:45 – 17:15

Room Amethyst

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RARE DISEASE TREATMENTS BEYOND MEDICINAL PRODUCTS

Session Chair:

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

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

Round table discussion

Radoslaw Kaczmarek, Member of the EHC Steering Committee. European Haemophilia Consortium (EHC), Belgium

Maria Pascual, Vice President Medical and Clinical Operations & Regulatory Affairs, TiGenix, Spain

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

THEME 6 | BEYOND MEDICAL CARE

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

Whilst high quality, accessible clinical care is essential for people with rare diseases, we are far more than just patients. This Theme aims to explore social and other non-medical issues that impact on quality of life and access to full citizenship, to identify innovative solutions and programmes that address these issues and to make the case for embedding best practice in these areas into European and national policies and provisions.

The approach will be participative, with platform speakers and delegates sharing knowledge, experience and aspirations, feeding into EURORDIS' future strategy on specialised social services.

The need for National Plans and Centres of Expertise to recognise and include non-medical care will be emphasised and the guiding principles and experience gained so far evaluated.

Innovative schemes that may serve as models for promoting independent living will be described, together with personal experiences of service users.

Session 0601

Friday, 9 May, 14:00 – 15:30

Room Bernstein

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 and demonstrate how National Plans and legislation can be a powerful national advocacy tool that can lead to change and improvement in patients' holistic care.

Current Challenges and Issues

Dorica Dan, President, Romania Prader Willi Association, Romanian National Alliance for Rare Diseases, Romania

How National Plans can Address These Issues

Simona Bellagambi, UNIAMO, Italy

Case Study: The experience of France

Christel Nourissier, Alliance Maladies Rares, France

Session 0602

Friday, 9 May, 16:30 – 18:00

Room Bernstein

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

Session Chair:

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

Social services face challenges when dealing with individuals with rare diseases. Depending on the type of the service and the life situation of the individual, adaptation of the available services is often needed. For that, information on the rare disease, specifically

tailored to the needs of social services, is needed, including experiences from the individuals who have provided or received such services. This session gives examples of different approaches to these challenges.

Orphanet Disability Projects

Myriam de Chalendar, Orphanet, France

Social Profiles Project by Rare Diseases Denmark

Birthe Holm, Rare Diseases Denmark, Denmark

Online Platform for Patients to Share Innovative Solutions/ Discoveries

Pedro Oliveira, University of Lisbon, Portugal

Session 0603

Saturday, 10 May, 09:00 – 10:30

Room Bernstein

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 health care professionals is encouraged.

Guiding Principles for Specialised Social Services and Guidelines for the Training of Social Service Providers

Dorica Dan, President, Romania Prader Willi Association, Romanian National Alliance for Rare Diseases, Romania

Panel Discussion

Terry Dignan, Serious Fun Network, Ireland

Norbert Hödebeck-Stuntebeck, Charitable Foundation for People with Disabilities, Prader-Willi Syndrome, Germany

Lisen Julie Mohr, Frambu, Norway

Anders Olauson, Ågrenska, Sweden

Session 0604

Saturday, 10 May, 11:30 – 13:00

Room Bernstein

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

Session Chair:

Gabor Pogany, President Rare Diseases Hungary, Hungary

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

Case studies and initiatives:

The Balance between Needing Care and Living Independently for a Young Man with a Degenerative Rare Disease

Hanka Meutgeert, Mother of a young adult, living with a degenerative rare disease, the Netherlands

My Story

Peter Ryan, Fighting Blindness, Dublin, Ireland

Let to Fly – Independent life camps for rare disease youths with intellectual disabilities

Beata Boncz, Hungarian Williams Syndrome Association, Hungary
Krisztina Pogany, Living with Williams Syndrome, Hungary

Session 0605

Saturday, 10 May, 14:00 – 15:30

Room Saphir 2 & 3



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

Session Chair:

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

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

Online Psychological Support for people living with a rare disease

Alba Ancochea, The Spanish Federation of Rare Diseases (FEDER), Spain

Ristoro Fantasia – Creating employment for people living with a rare disease

Renza Barbon Galluppi, President of Italian Federation of Rare Diseases (UNIAMO), Italy

Empowerment Weekends for Young Adults with Anorectal Malformations

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

Panel Discussion

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

Session 0606

Saturday, 10 May, 15:45 – 17:15

Room Saphir 2 & 3



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

Session Chair:

John Dart, Chief Operating Officer, DEBRA International, UK

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

The Proposed Role of Centres of Expertise Based on European Union Committee of Experts on Rare Diseases (EUCERD) Recommendations

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

Speaker invited

Panel Discussion and Lively Debate

Francesc Palau, CIBERER, Spain

Annette Grütters-Kieslich, Medical Director of the Charité Center for Women and Child Health and Human Genetics, Germany