7th European Conference on Rare Diseases & Orphan Products

The Rare Disease Puzzle: Bringing the Picture to Life

08-10 May 2014
Andel’s Hotel, Berlin, Germany

Programme

A conference organised by EURORDIS

Co-organised by DIA

www.rare-diseases.eu
MESSAGE FROM CO-CHAIRS

On behalf of the European Organisation for Rare Diseases (EURODIS) and DIA Europe, it gives us great pleasure to welcome all stakeholders involved in the rare disease environment to the 7th European Conference on Rare Diseases & Orphan Products (ECrD 2014 Berlin).

ECrD is the foremost meeting of the rare disease community and a unique platform across all rare diseases, across all European countries, bringing together all stakeholders - patients, caregivers and patients’ representatives; academics, scientists and researchers; payers and regulators; health care professionals, industry, policy makers and representatives of the Member States. The first ECrD was held in Copenhagen in 2001, bringing together over 300 stakeholders from all interested parties in the rare disease community. Such a positive impact was achieved by this groundbreaking initiative that there has been an ECrD held in different member states at two year intervals since then, monitoring and benchmarking relevant initiatives and empowering the RD community.

ECrD 2014 aims at sustaining the dynamic of exchange of an increasing number of good practices, in a much broader spectrum of activities. The anticipated attendance at ECrD 2014 is 700 participants representing over 40 countries from around Europe and beyond.

Today, an estimated 30 million people are affected by rare diseases in Europe. Due to the low prevalence, national policy makers find it a very real challenge to fully meet the needs of people with rare diseases. However, there is a need to ensure continuity of action and prevention of duplication of efforts. Indeed, this thematic approach is designed to allow less advanced regions in this field to benefit from experience sharing with other areas in Europe.

With its plenary and parallel sessions addressing specific issues, knowledge-sharing and debate are encouraged, the exchange of real experiences and best practices are integrated into the programme, cooperation and networking are stimulated and awareness is increased while ensuring continuity of action and prevention of duplication of efforts. Indeed, this thematic approach is designed to allow less advanced regions in this field to benefit from experience sharing with other areas in Europe.

The Opening & Plenary Sessions are interpreted from English into five languages: French, Spanish, German, Polish and Russian. The Poster Committee has been impressed by the high quality of poster presentations submitted to the conference. The 100 posters per day that have been selected are showcased in the poster area located on the first floor of the conference venue. We encourage you to look at them. A Patient Advocates Fellowship Programme has been made available to support the participation of patients’ advocates.

Lastly, and by no means least, thanks to our host country, participants will enjoy ample opportunities to network and visit the vibrant and cultured city of Berlin.
PROGRAMME CO-CHAIRS

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)

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

Lesley Greene
Vice-President, Children Living with Inherited Metabolic Diseases (CLIMB), Vice-Chair, Committee for Orphan Medicinal Products (COMP), Volunteer Patient Advocate, EURORDIS, UK

PROGRAMME COMMITTEE

John Dart
Chief Operating Officer, Debra International, EURORDIS Board of Directors, UK

Loïc Guillemin
Chairman of European Federation of Internal Medicine (EFIM) Research Working Group Hôpital Cochin - Service de Médecine Interne, France

Véronique Héon-Klin
Federal Ministry of Health, Germany

Thierry Laugel
Managing Partner, KLS, France

Yann Le Cam
Chief Executive Officer, EURORDIS, France

Birka Lehmann
German Federal Institute for Drugs and Medical Devices, Ad-hoc member of the DIA Advisory Council, Alternate member of the Paediatric Committee (PDCO), Germany

Kevin Loth
Vice-Chair Task Force Rare Diseases & Orphan Drugs of EuropaBio-EBE, Celgene, UK

Milan Macek
Former Chairman, European Society for Human Genetics, Czech Republic

Christoph Nachtigaele
President, German National Alliance for Chronic Rare Diseases (ACsHE), Germany

Gabor Pogany
President, Rare Diseases Hungary, Hungary

Peter Saltonstall
President and Chief Executive Officer, National Organization for Rare Disorders (NORD), USA

Bruno Sepodes
Chair COMP, National Competent Authority, University of Lisbon, Portugal

Till Voigtländer
Clinical Institute of Neurology, Medical University of Vienna, Austria

Durhane Wong Rieger
President, Canadian Organization for Rare Disorders (CORD), Canada

ORGANISING COMMITTEE

Sharon Ashton
Senior Event Manager, EURORDIS, France

Mary Dunkle
Vice-President of Communications, National Organization for Rare Disorders (NORD), USA

Anja Helm
Senior Manager of Relations with Patient Organisations, EURORDIS, France

François Houyëz
Treatment Information and Access Director, Health Policy Advisor, EURORDIS, France

Lene Jensen
Director, Rare Diseases Denmark, Denmark

Tamara Kohler
Team Leader, Planning and Development, DIA Europe, Switzerland

Jytte Lyngvig
Director, DIA Europe, Switzerland

Mirjam Mann
Director, German National Alliance for Chronic Rare Diseases (ACsHE), Germany

Maria Mavris
Therapeutic Development Director, EURORDIS, France

Esther Neiditsch-Prigioni
General Secretary, ProRaris (Swiss National Alliance for Rare Diseases), Switzerland

Cor Oosterwijk
Director, VSO (Dutch National Alliance for Rare Diseases), The Netherlands

Patrice Régnier
Finance & Support Services Unit Director, EURORDIS, France

Rainer Riedl
President, Pro Rare Austria, Austria

Mirosław Zielinski
Chairman, Polish National Forum on the Treatment of Orphan Diseases (ORPHAN), Poland
Thursday, 8 May 2014

12.00 – 18.00  Registration open for ECRD 2014
14.30 – 17.00  Pre-conference Tutorials
18.00 – 19.00  Patient Groups Welcome Reception

Friday, 9 May 2014

08.00 – 18.00  Registration open
09.00 - 09:45  Opening Session
09:45 - 10:15  Coffee break
10:15 - 12:00  Plenary Session
12:00 – 13:00  Lunch and exhibition
13:00 - 14:00  Poster Session
14:00 - 15:30  Session 1 - choose from 6 parallel sessions
15:30 - 16:30  Coffee break, posters and exhibition
16:30 - 18:00  Session 2 - choose from 6 parallel sessions
18:00 – 19:00  Networking Reception on-site

Saturday, 10 May 2014

09:00 - 10.30  Session 3 - choose from 6 parallel sessions
10:30 - 11:30  Coffee break, posters and exhibition
11:30 - 13:00  Session 4 - choose from 6 parallel sessions
13:00 - 14:00  Lunch, posters and exhibition
14:00 - 15:30  Session 5 - choose from 6 parallel sessions
15:45 - 17:15  Session 6 - choose from 6 parallel sessions
17:15 - 17:45  Informal farewell coffee

GENERAL INFORMATION

Conference Languages

Simultaneous interpretation
All pre-conference workshops and tutorials will be conducted in English only.

Opening & Plenary Sessions
The Opening & Plenary Sessions taking place on the morning of Friday, 9 May will be simultaneously interpreted from English into 5 languages: German, French, Spanish, Polish and Russian.

Other Sessions
A selection of other sessions will be simultaneously translated into German and Russian. Please refer to the programme for more specific details. All other sessions will be conducted in English only.

Registration Opening Times

The registration desks are located on the ground floor of the conference venue. A separate Speakers registration desk will be set-up and open during the following times:

Thursday, 8 May 12.00 – 18.00
Friday, 9 May 08.00 – 18.00
Saturday, 10 May 08.30 – 14.00

On-site Speaker Room (Speaker Preview)
The Speakers Room is located on the 1st floor of the conference venue in the Aquamarin room. On-site welcome staff will direct you.

ACKNOWLEDGMENTS AND CREDITS

Credits, supports and legal information
We wish to thank the following institutions for their active collaboration

Conference Organiser
The 7th European Conference on Rare Diseases & Orphan Products (ECRD 2014) is a conference organised by EURORDIS

Co-organised by
DIA Europe

With the Support of
• ACHSe (German National Alliance for Chronic Rare Diseases)
• AFM Télénôhon
• CORD (Canadian Organization for Rare Disorders)
• EFIM (European Federation of Internal Medicine)
• EuropaBio-EBE
• European Commission, DG Health and Consumers’ Protection
• European Medicines Agency
• European Society of Human Genetics
• NORD (National Organization for Rare Disorders), USA
• Orphanet

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 and certificates are available on request from the DIA registration desk.
PATIENT GROUPS WELCOME RECEPTION

Thursday, 8 May 2014
18.00 – 19.00
Andel’s Hotel, Berlin, Germany

A welcome reception will be held for registered patients and patients’ advocates on Thursday, 8 May 2014 from 18.00 to 19.00 at the ECRD 2014 conference venue (Andel’s Hotel, Berlin, Germany). Drinks and snacks will be served.

NETWORKING RECEPTION

Friday, 9 May 2014
18.00 – 19.00
Andel’s Hotel, Berlin, Germany

All registered participants are invited to attend this convivial networking reception taking place from 18.00 to 19.00 at the ECRD 2014 conference venue (Andel’s Hotel, Berlin, Germany).

The winners of the Posters will be announced and presented with their certificates at this networking event. Enjoy drinks and finger food while mingling with your fellow participants.

FAREWELL COFFEE

Saturday, 10 May 2014
17.15 – 17.45
Andel’s Hotel, Berlin, Germany

All registered participants are invited to attend this informal farewell coffee break. This will be the occasion to say goodbye to fellow participants before leaving Berlin or enjoying an extended stay in the city.
### Thursday 8 May 2014

<table>
<thead>
<tr>
<th>Time</th>
<th>Tutorial 1</th>
<th>Tutorial 2</th>
<th>Tutorial 3</th>
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<tr>
<td>14:30 - 17:00</td>
<td>Diamant HTA 101 for Rare Diseases</td>
<td>Quartz RD Connect (an integrated platform connecting registries, biobanks and clinical bioinformatic for Rare Disease research)</td>
<td>Topas 1 Supporting the pathway to trials for Rare Diseases: clinical trial design and other considerations</td>
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### Friday 9 May 2014

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<tr>
<th>Time</th>
<th>Event</th>
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| 09:00 - 09:45 | Opening Session - Plenary Room Rubin (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 |
| 09:45 - 10:15 | Coffee Break                                                    |
| 10:15 - 12:00 | Plenary Session - Plenary Room Rubin (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 |
| 12:00-13:00 | Lunch and Exhibition                                               |
| 13:00 - 14:00 | Poster Session                                                     |
| 14:00 - 15:30 |海报 Session 0101 - Saphir 2 & 3  
Centres of Expertise - Part 1 (Models & Practical Examples)  
Poster Session 0201 - Amethyst  
The Role, Risks and Relevance of registries in shaping therapy development to 2020  
Poster Session 0301 - Saphir 1  
Shaping rare disease research policy |
| 15:30 - 16:30 | Coffee Break, Posters and Exhibition                              |
| 16:30 - 18:00 | Session 0102 - Jade  
Centres of expertise - Part 2 (Designation and Evaluation)  
Session 0202 - Amethyst  
A collaborative Model to progress Knowledge and Research  
Session 0302 - Saphir 1  
Addressing the gaps in Research at international level to identify opportunities |
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<td><strong>Smaragd</strong></td>
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<td><strong>Tutorial 4</strong></td>
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<tr>
<td>How to get the best out of orphanet data</td>
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<td><strong>Keynote Addresses:</strong></td>
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<tr>
<td>Hermann Gröhe, German Federal Minister of Health, Germany</td>
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<td>Christoph Nachtigall, President, German National Alliance for Chronic Rare Diseases (ACHSE), Germany</td>
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<tr>
<td>Irene Norstedt, Head of Unit, Personalised Medicine, Directorate for Health Research at the DG Research &amp; Innovation, European Commission</td>
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<tr>
<td>Lesley Greene, Vice-Chair COMP, Vice-President, CLIMB, Co-Chair Programme Committee ECRD 2014, UK</td>
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The Opening & Plenary Session will be simultaneously interpreted from English into 5 languages: German, French, Spanish, Polish and Russian and bring together all stakeholders: patients’ representatives, health professionals, political leaders, officials from the EU, academia and industry.

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<th><strong>Coffee Break</strong></th>
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Round table discussion: The challenges ahead in the fields of organisation of healthcare, research and access to orphan medicinal products

Participants: Ségolène Aymé, Serge Braun, Kate Bushby, Pauline Evers, Wills Hughes-Wilson, Tsveta Schyns, Bruno Sepodes, Thomas Wagner

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<th><strong>Poster Session</strong></th>
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**Theme 04**
State of the Art and Innovative Practices in Orphan Products

**Session 0401 - Opal**
Current landscape of policy development on orphan products and rare disease therapies

**Session 0501 - Jade**
Early dialogue and horizon scanning of product development to address unmet medical needs

**Session 0601 - Bernstein**
Identifying specific social challenges of rare diseases

**Coffee Break, Posters and Exhibition**

**Session 0402 - Opal**
Facts on current patient access challenges to orphan products

**Session 0502 - Saphir 2 & 3**
How to shape a better framework for orphan drug development: EMA/FDA Collaboration

**Session 0602 - Bernstein**
Different approaches to the social challenges of rare diseases: Social Policy
**Saturday, 10 May 2014**

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<td>09:00 - 10:30</td>
<td><strong>Session 0103 - Jade</strong>&lt;br&gt;European Reference Networks (ERNS)**</td>
<td><strong>Session 0203 - Saphir 1</strong>&lt;br&gt;Making the invisible visible: The coding of rare diseases in health information systems</td>
<td><strong>Session 0303 - Opal</strong>&lt;br&gt;Incentives to create a favourable Eco-System</td>
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<td>10:30 - 11:30</td>
<td><strong>Coffee Break, Posters and Exhibition</strong></td>
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<td>11:30 - 13:00</td>
<td><strong>Session 0104 - Saphir 2 &amp; 3</strong>&lt;br&gt;Adressing the challenges of healthcare pathways</td>
<td><strong>Session 0204 - Saphir 1</strong>&lt;br&gt;Delivering help and support in virtual world: what will work best?</td>
<td><strong>Session 0304 - Opal</strong>&lt;br&gt;Breakthroughs in science</td>
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<td>13:00 - 14:00</td>
<td><strong>Lunch, Posters and Exhibition</strong></td>
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<td>14:00 - 15:30</td>
<td><strong>Session 0105 - Jade</strong>&lt;br&gt;Advances in diagnostic possibilities for undiagnosed patients</td>
<td><strong>Session 0205 - Bernstein</strong>&lt;br&gt;Knowledge at the point of care: Getting the facts just in time or just in case</td>
<td><strong>Session 0305 - Opal</strong>&lt;br&gt;Pre-competitive tools and resources / public-private partnership in the area of rare diseases</td>
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<td>15:45 - 17:15</td>
<td><strong>Session 0106 - Jade</strong>&lt;br&gt;Improving the quality of and access to diagnostic services</td>
<td><strong>Session 0206 - Bernstein</strong>&lt;br&gt;Hype, help or harm? the impact of media promotion of rare diseases</td>
<td><strong>Session 0306 - Opal</strong>&lt;br&gt;Whose data is it?: stimulating research and removing barriers</td>
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<td>17:15 - 17:45</td>
<td><strong>Farewell Coffee Break</strong></td>
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### Saturday, 10 May 2014

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<th>Theme 06</th>
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<td>State of the Art and Innovative Practices in Orphan Products</td>
<td>Emerging Concepts and Future Policies for Rare Disease Therapies</td>
<td>Beyond Medical Care</td>
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**Session 0403 - Saphir 2 & 3**
EMA - HTA interfacing on rare disease therapies

**Session 0503 - Amethyst**
Progressive patient access schemes and patient involvement in benefit-risk assessment

**Session 0603 - Bernstein**
Concrete solutions to social challenges: essential tools for the integration of rare diseases into social services

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**Coffee Break, Posters and Exhibition**

**Session 0404 - Jade**
Shortages in authorised medicines for rare diseases

**Session 0504 - Amethyst**
Mechanism of coordinated access (MOCA) and transparent value framework, managed entry agreements

**Session 0604 - Bernstein**
Can people living with a rare disease be independent? Inspiring personal stories

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**Lunch, Posters and Exhibition**

**Session 0405 - Amethyst**
Understanding of orphan therapies off-label uses and their new challenges

**Session 0505 - Saphir 1**
Emerging ideas for sustainable access to orphan medicinal products

**Session 0605 - Saphir 2 & 3**
Can people living with a rare disease be independent? Inspiring solutions by providers

**Session 0406 - Saphir 1**
Empowering patient advocates in drug development

**Session 0506 - Amethyst**
Rare disease treatments beyond medicinal products

**Session 0606 - Saphir 2 & 3**
How centres of expertise should/could interface with social services

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**Farewell Coffee Break**

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### DON’T MISS

**EURORDIS Membership Meeting 2016**
8th European Conference on Rare Diseases & Orphan Products
Dates available soon!
Edinburgh, Scotland
POSTERS

Visit the poster presentations:
Friday, 9 May 2014, 9.45 - 10.15, 13.00 - 14.00 and 15.30 - 16.30 Saturday, 10 May 2014, 10.30 - 11.30 and 13.00 - 14.00

Theme 1 - Improving Healthcare Services

P 1  Haemoglobinopathies in Europe: Health & Migration Policy Perspectives
Patricia Aguilar Martinez, Michael Angastiniotis, Androulla Eleftheriou, Beatrice Gulbis, Maria del Mar Mañú Pereira, Roumyana Petrova-Benedict, Joan-Lluís Vives Corrons

P 2  se-atlas: Cartographic Representation of Experts on Rare Diseases
Holger Storf, Tobias Hartz, Wulf Pfeiffer, Kathrin Rommel, Mareike Derks, Elisabeth Nyoungui, Jörg Schmidtke, Holm Graessner, Mirjam Knoell, Thomas Wagner, Frank Uckert

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

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

P 5  The impact of rare diseases on the health care system: linking the Veneto Region Rare Diseases Registry with health current statistics
Laura Visonà Dalla Pozza, Michela Biasio, Erma Toto, Paola Facchin

P 7  The first year of activity of Students’ Scientific Circle on Rare Diseases in Pomeranian Medical University in Szczecin, Poland
Michał Skoczylas, Jacek 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 Miedzybrodzka on behalf of the EHDN Standards of Care Working Group

P 10 A Survey on the Centres of Expertise for Rare Diseases in Italy
R. Mingarelli, S. Ciampa, R. Ruotolo, M. Di Giacinto, S. Festa, E. Cocchiara, B. Dallapiccola

P 11 The Projects 12 months 12 Therapeutic Groups and everyone adds
B. Dallapiccola, R. Mingarelli, S. Ciampa, R. Ruotolo, M. Di Giacinto, S. Festa, E. Cocchiara, B. Dallapiccola

P 12 Only the strong survive
Birthe Byskov Holm, Lene Jensen

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

P 14 Health care transition in rare diseases patients: results from a population-based Registry
Monica Mazzucato, Cinzia Minichiello, Martina Bua, Paola Facchin

P 16 Treatment of patients with rare diseases: individual treatments or protocols?
M. Volta, M. Vizioli, E. Calzolari, E. Rozzi, E. Di Rusci

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

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

P 19 Comparative assessment of family’s experience of patients with Dravet Syndrome on the use of rectal diazepam and buccal midazolam
Nathalie Coqué, Nicole Chemaly, Rima N.About

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”
Africca Villanueva, Richard Woolley, Virginia Corrochano, Beatriz Gómez, Jordi Molas, José David Barberà, Francesca Palau

P 22 A Clinical advisory board for a Rare disease (Prader-Willi Syndrome)
Susanne Blichfeldt; Stense Farholt

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

P 24 Improving healthcare in adult patients with rare diseases in Poland
Joanna Sulicka, Joanna Pera, Magdalena Strach, Jaroslaw Królczyk, Izabela Kierzkowska, Agnieszka Słöwik, Tomasz Grodzicki

P 26 Newborn Screening For Inherited Metabolic Diseases: the Network Of Emilia-Romagna Region
M. Volta, E. Rozzi, A. Cassio, G. Biasucci, F. Righetti, M.O. Bal, F. Baronio, I. Bettocchi, E. Di Rusci

P 27 I amcing the cost of hereditary angioedema in England
M. Helbert, T. Holbrook, A. MacCulloch, A. Mannan

P 28 Quality analysis of health care network through patients’ and families’ judgement
Silvia Manea, Sara Barbieri, Miriam De Lorenzi, Paola Facchin

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

P 30 Gene Panel Diagnostics for Disorders with abnormal Bone mass
Izabella Kierzkowska, Agnieszka Słöwik, Tomasz Grodzicki

P 31 Establishment of narcolepsy-centres in Germany
Ulff Kallweit, Geert Mayer, Norbert Dahmen, Volkmar Westdickenberg, Christine Pitzen, Claudia Schitto

P 32 An appropriate way of organizing services for rare disorders
Lena Lande Wekre, Lena Haugen, Kjersti Vardeberg

P 33 Genodermatoses Network – Towards a Rare And Severe Skin Diseases European Reference Network sharing experience and knowledge with Mediterranean and Middle-Eastern experts and centres
Christine Bodemer, Johann Bauer
P 35 | Understanding the healthcare experiences and needs of people living with Huntington’s Disease (HD): an exploratory study
D Rae, S McCann, Z Miedzybrodzka on behalf of the EHDN Standards of Care Working Group

P 36 | Rare professionals for rare diseases
Piergiorgio Miottello, Giulia Mariani

P 37 | Multidisciplinary clinic for patients with Gorlin syndrome in Norway
Charlotte von der Lippe, Lajla Schulz

P 38 | Boys with sex chromosome aneuploidies (SCA) compared to a clinical sample
K. Fjermestad, S. Stokke

P 39 | Improving health care and social services for patients with Neuro-muscular diseases in the Southeast healthcare region in Sweden
Rebecca Pestoff, Charlotte Lilja, Olof Danielsson, Cecilia Gunnarsson

Theme 2 - Knowledge Generation & 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 Bladen, Hanns Lochmuller

P 42 | Strategies for citizen control: patient engagement in rare disease biobank and genomics research in Europe.
Pauline McCormack, Anna Kole

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

P 45 | OSSE – Open Source Registry Software Solution
Martin Lablans, Marita Muscholl, Tobias Hartz, Holger Storf, Thomas Wagner, Frank Ückert

P 46 | Characterization and classification of Rare Disease Registries by using exploratory data analyses

P 47 | National RD registry in Spain: Pilot study of the Spanish Rare Diseases Registries Research Network (SpainRDR)

P 48 | Implementation of and first experiences with the Central Registry for Rare Diseases in Belgium
Elifriede Swinnen, Annelies Mallezie, Montse Urbana Paz, Viviane Van Casteren

P 49 | UK FSHD Patient Registry

P 50 | Sanfilippo Syndrome Registry Project and Natural History Studies: An Example of Patients, Parents and Researchers Collaborating for a Cure

P 51 | The RE(ACT) Initiative and the use of online communities to enhance research on rare diseases
Olivier Menzel

P 52 | e-Enerca: New e-Health Services for the European Reference Network on Rare Anaemias
Maria del Mar Mañú Pereira, Laura Olaya Costa, Juan Luis Vives Corrons

P 53 | The TREAT-NMD Duchenne muscular dystrophy registries: conception, design and utilisation by industry and academia
Catherine L Bladen, Hanns Lochmuller

P 54 | Rare Diseases are Common
Ela Malarsowska

P 55 | Needs for Information on rare diseases: the most used Orphanet services and products as an indicator

P 56 | A review of the data collection features of international and national platforms for the registration of rare disease patients
Domenica Taruscio, Rosa Giuseppa Frazzica and CNMR Researchers

P 57 | Spanish National Rare Disease Biobank (BioNER)

P 59 | Empowerment of rare diseases patients organisations by a searchable, crosslinking website
Marianne G. Nijnuis, Mariette H.E. Driessens, Ildikó Vajda, Silvia van Breukelen, Cor Oosterwijk

P 60 | OrphanAnesthesia – Anesthesia recommendations for patients suffering from rare diseases
Tino Münster, Nina Schnabel

P 61 | Telethon Infoline: supporting patients through updated and useful information on genetic diseases
Alessia Daturi, Vera Bianchi, Barbara Gentilin

P 62 | The TREAT-NMD Care and Trial Site Registry: A Powerful Tool for Clinical research on Neurodegenerative and Neuromuscular Diseases
Kirsten König, Kathrin Gramsch, Adrian Tassoni, Sunil Rodger, Cathy Turner, Brigitte Paap, Thomas Klockgether, Volker Straub, Kate Bushby, Hanns Lochmuller, Janbernd Kirschner

P 63 | Unmet needs in the Rare Disease Community
Dianne Petrie, Catherine Spinks, Bronwyn Burgess

P 64 | TOSCA – An International Disease Registry for Tuberous Sclerosis Complex
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Eline Picavet, Thomas Morel, David Cassiman, Steven Simoens

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G Santoni, F Capozzoli, F Paoli, P Torreri, C Borzacchello, C Frank

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P 158 | The role of research in understanding the psychosocial needs of families affected by serious illness and innovative programme responses
Caroline Stott, Suzanne Guerin

P 159 | Romanian Prader Willi Association - a model for approaching rare diseases in Romania
Dorica Dan, Maria Puiu

P 160 | Wing Test Project - independent living camps for Williams Syndrome youth
Beáta Boncz, Krisztina Pogány, Katalin Heiszer, Ágnes Várheleyiné Monostori, Zsuzsanna Pogányné Bojtor

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P 163 I School- starters with a rare disorder - a model for supplying information to school personnel in Norway
Susan Sadal, Synne Heilvang

P 164 I Giving a Voice to Parents: Experience of Caring for Children, Adolescents and Young Adults with Mucopolysaccharidoses (MPS)
Suja Somananadan

P 165 I The healing power of being together
Velia Maria Lapadula, Giulia Mariani

P 167 I DebRA Slovakia and its experience with Specialised Social Services in Epidermolysis bullosa
B. Ramiljaková, T. Foltánová, K. Martinásková, J. Pěč

P 170 I Chromosomal Disorders. Developing a design for a multidisciplinary study of Norwegian patients with Kleeftstra syndrome and Wolf-Hirschhorn syndrome
Lise B. Hoxmark, Heidi E. Nag, David K. Bergsaker, Bente S. Hunn, Susanne Schmidt

P171 I Change of Emotional Perspective in Prader-Willi Syndrome (PWS)
Norbert Hoedebeck-Stuntebeck

Theme 7 - Rare Disease Patient Groups Innovations

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S. Masefield, N. Cassidy, P. Powell

P 174 I Promote Therapeutic Patient Education for rare diseases in France
Marie-Pierre Bichet, Paul Gimenes

P 175 I Creating a European Chromosome 18 Clinical Research Network
Alistair McLaughlan, Bonnie McKerracher, Apostolis Ritsos, Jannine D. Cody

P 177 I A Funding Program for the autonomy, the autonomy and the social integration of the people affected by Alternating Hemiplegia of Childhood (AHC) in Italy, and for the empowerment of their families
Rosaria Vavassori, Nicolò Bona, Denise Previtali

P 178 I Integrating personalized medicine into patient organizations’ tasks: a workshop with moral deliberation
Stephanie S. Weinreich, Elisabeth Martens, Bert Molewijk

Theme 8 - National Plans

P 179 I Evolution of national and European policies in the field of rare diseases and their impact over the past four years
Charlotte Rodwell, Ségoîène Aymé

P 180 I Selecting Core Indicators for National Plans According to the Delphi Technique
Marta De Santis, Amalia Egle Gentile, Rita Maria Ferrelli, Manuel Posada De La Paz, Luciano Vittozzi, Rosa Giuseppa Frazzica, Agata Polizzi, Domenica Taruscio

P 181 I Rare disease patient organisations’ opinions about the implementation of the national plan in Finland: a survey
The Finnish Network for Rare Diseases

P 183 I German National Plan of Action for People with Rare Diseases
Miriam Schlangen, Katharina Heuing

P 184 I Strengths and Weaknesses in Developing National Plans for Rare Diseases: Results of a Europlan Survey
Rita Maria Ferrelli, Marta De Santis, Giuseppa Rosa Frazzica, Amalia Egle Gentile, Domenica Taruscio

P 185 I The role of Patient Organisations in the creation of the Hungarian National Plan for Rare Diseases
Gábor Pogány, Helga Süli-Vargha, Judit Váradiné Csapó, Katalin Brunner

P 186 I National Conferences in the frame of the EUROPLAN project in post-Soviet countries
Oleg Klvividize

Theme 9 - Other/open topic

P 187 I VEMSE-CF – A Prospective Controlled Care-Research Study Investigating the Effects of a Comprehensive Psycho-Social Intervention

P 188 I Genetic Analysis of Mucopolysaccharidoses in Russia

P 189 I Are there Differences between People with Prader-Willi Syndrome (PWS) and Healthy Controls in Heart Rate Variability Parameters in Response to Eating-Related Pictures?
Hubert Soyer, Svetlana Labun, Arne Wittling, Elisabeth Schweiger

P 190 I The Porphyrias in Southern Italy: Clinical, Therapeutic and Social Services Aspects
Claudio C. Guida, Maria Savino, Maria Garrubba, Annabella Di Giorgio, Maria Placentino, Adelaide Potenza, Lazzaro di Mauro, Filippo Aucella

P 191 I Intravenous Neridronate in Patients with Osteogenesis Imperfecta
Ombretta Viapiana, Davide Gatti, Maurizio Rossini, Elena Fraccassi, Sonila Troplini, Ilaria Piazza, Giovanni Orsolini, Luca Idolazzi, Silvano Adami

P 193 I Preclinical studies: research discoveries to find a cure
Alba Di Pardo, Vittorio Maglione, Mariagrazia Favellino, Enrico Amico and Ferdinando Squitieri

P 194 I APMAR (Italian non-profit association of People with Rheumatic Diseases): supporting rheumatic diseases’ patients' volunteer

P 195 I Physical Activity and Walking Capacity in Persons with Down Syndrome, Williams syndrome and Prader-Willi syndrome
Marianne Nordstrøm, Bjørge Herman Hansen, Benedicte Paus, Svein Olav Kolset

P 198 I EUCERD Joint Action - Working for Rare Diseases: Goals and Achievements
Victoria Hedley, Stephen Lynn, Teresinha Evangelista, Antonio Atalaia, Kate Bushby

P 199 I STEPS-2, a 2-Year, Multicenter, Open-Label Clinical Trial: Safety and Efficacy of Long-Term Teduglutide 0.05-mg/kg/day Treatment of Adults With Intestinal Failure Associated With Short Bowel Syndrome (SBS–iF)
Simon M. Gabe, Palle B. Jeppesen, Ulrich-frank Pape, Nader N. Youssef

P 200 I The unique difficulties of rare disease patients to access health and social care in Europe and the role of early diagnosis: the case of Prader-Willi Syndrome (PWS)
S.K Manollas, K. Politis, H. Frysira
Tutorial 1
Thursday, 8 May 2014 14:30 - 17:00
Room Diamant

**HTA 101 FOR RARE DISEASES**

Josie Godfrey, Associate Director, Highly Specialised Technologies, National Institute for Health and Care Excellence (NICE), UK

Elena Nicod, Research Officer, LSE Health and Social Policy

Vinciane Pirard, Director Public Affairs, Genzyme, Belgium

Health Technology Assessment is a multidisciplinary process that summarises information about the medical, social, economic and ethical issues related to the use of a health technology in a systematic, transparent, unbiased and robust manner. Its purpose is to guide policymakers to make decisions on the adoption of new and existing technologies. This tutorial will introduce the participant to the principles of health technology assessment, describe the methods of economic evaluation, illustrate the unique circumstances surrounding rare disease evidence, provide an overview of the different approaches for assessing rare disease drugs across countries and discuss other factors which may contribute to funding decisions.

**Learning Objectives**

At the conclusion of this tutorial, participants will have a better understanding of:

- HTA in order to contribute and engage more effectively in future assessments of medicines for rare diseases
- Have an understanding of the principles and process of health technology assessment. The focus being on the various approaches and application to rare diseases
- Gain knowledge and skills in how various stakeholders, particularly patient organisations can contribute and engage in the process of HTA

**Target Audience**

Those with limited / no technical understanding or experience of health technology assessment in relation to rare diseases. Participants from patient organisations, government agencies, other non-profit organisations and industry are encouraged to attend.

Tutorial 2
Thursday, 8 May 2014 14:30 - 17:00
Room Quartz

**RD-CONNECT: AN INTEGRATED PLATFORM CONNECTING DATABASES, REGISTRIES, BIOBANKS AND CLINICAL BIOINFORMATICS FOR RARE DISEASE RESEARCH**

Lucia Monaco, Chief Scientific Officer, Fondazione Telethon, Italy

Peter Robinson, Professor of Medical Genomics, Institute of Medical and Human Genetics, Charité, Universitätmedizin Berlin, Germany

Despite many examples of excellent practice, rare disease research is still mainly fragmented by data type and by disease. Individual efforts have little interoperability and almost no systematic connection of detailed clinical information with genetic information, biomaterial availability or research/trial datasets. By developing robust mechanisms and standards for linking and exploiting these data, RD-Connect will develop a critical mass for harmonisation and provide a strong impetus for a global “trial-ready” infrastructure ready to support the IRDIRC goals for diagnostics and therapies for rare disease. During this tutorial, participants will gain a greater appreciation of the aims and objectives of RD-Connect and how different user groups e.g. patients/patient representatives, researchers and industry can best utilise the platform. Specifically, we will discuss topics such as the integration of bioinformatics tools, registries and biobanks into the platform, and also the legal, ethical and regulatory issues that need to be considered.

**Learning Objectives**

- Gain a greater understanding of the aims and objectives of the RD-Connect platform
- Learn how the RD-Connect platform can be utilised by different groups of individuals e.g.
  - How to set up and manage a registry or a biobank for a disease
  - How to utilise RD-Connect to study rare disease biospecimen
  - How to utilise RD-Connect to plan a rare disease trial
- How to analyse exomes for rare disease diagnostics and novel gene discovery

**Target Audience**

This tutorial will be of interest to all those working in the rare disease field who would like to learn more about the RD-Connect platform, including clinicians, researchers, patient advocacy groups and industry.

Tutorial 3
Thursday, 8 May 2014 14:30 - 17:00
Room Topaz 1

**SUPPORTING THE PATHWAY TO TRIALS FOR RARE DISEASES: CLINICAL TRIAL DESIGN AND OTHER CONSIDERATIONS**

Cristina Cismon, TACT Core Group Member, CEO, Cydan Development, USA

Dominic Wells, TACT Chair, Professor in Translational Medicine, Royal Veterinary College, UK

Ralf-Dieter Hilgers, IDEAl coordinator, Department of Medical Statistics, RWTH Aachen University, Germany

**Part 1**

Clinical trial design for small sample population groups

Ralf-Dieter Hilgers, IDEAl coordinator, Department of Medical Statistics, RWTH Aachen University, Germany

There is a pressing need to integrate a broad range of innovative methodologies improving clinical trials in the setting of small sample population groups (SPG). The objective of the IDEAl project is to produce methods of general applicability irrespective of indication by Integrated Design and Analysis of clinical trials in SPG through a multidisciplinary closely collaborating consortium of researchers from European universities, research institutes and industry. By combining, enhancing and developing different statistical methodologies and assessment methods, this research programme will impact the scientific discussion in promoting efficient statistical methodology for clinical trials in SPG, also in view of existing regulatory guidance in the EU. This tutorial will stimulate this discussion by relating our research to known design settings. The IDEAl project received funding from the European Union’s Seventh Framework Programme for research, technological development and demonstration under Grant Agreement No. 602552.

**Learning Objectives**

Attendees will:

- Gain knowledge in some specific design and analysis challenges of clinical trials for small population groups
- Understand the importance of appropriate design and analysis of small population group trials
Target Audience
This tutorial will be of interest to all stakeholders who would like to hear more about the design challenges in small population group trials, which will be solved by the IDEAL project. This includes clinicians, researchers, patient advocacy groups and industry.

Part 2
TACT, the TREAT-NMD Advisory Committee for Therapeutics
Dominic Wells, TACT Chair, Professor in Translational Medicine, Royal Veterinary College, UK
Cristina Csima, TACT Core Group Member, CEO, Cydan Development, USA

TACT, the TREAT-NMD Advisory Committee for Therapeutics, is an expert multidisciplinary body that provides the neuromuscular community (clinicians, researchers, patient advocacy groups and industry) with independent and objective guidance on advancing new therapies (whether novel or repurposed) for neuromuscular diseases. During this tutorial we will share our experience in advising on the drug development pathway for neuromuscular diseases and explain how this could be exported as a model for other rare diseases.

Learning Objectives
Attendees will:
- Gain a greater appreciation of how TACT reviews act as a constructively critical and educational resource for researchers and drug development sponsors evaluating research programmes with a translational goal toward entry into clinical trials in neuromuscular diseases
- Understand the value of a TACT review and how it can be utilised in a wider context including funding

Target Audience
This tutorial will be of interest to all stakeholders who would like to learn how the TACT model guides development of therapies for rare diseases. This includes clinicians, researchers, patient advocacy groups and industry.

Tutorial 4
Thursday, 8 May 2014 14:30 - 17:00
Room Smaragd

HOW TO GET THE BEST OUT OF ORPHANET DATA

Virginie Hivert, Orphanet Pharmaceutical Affairs & Expert Resources Team Manager, Orphanet-Inserm, France
Sandra Peixoto, Orphanet Resources Expert Team, Orphanet-Inserm, France

Over the years, Orphanet has developed a broad range of products which are of use for different purposes and/or different types of users. Apart from the information displayed through the website which is now quite well-known, emphasis can be put for example on: review articles that are produced or indexed to the classification of diseases; Orphanews, the newsletter which offers scientific, as well as political, literature survey; the documents of synthesis called 'Orphanet Report Series' dedicated to highlight a specific topic. Resources have also been put on providing the data in a re-usable format in order to foster the interoperability and analysis capacity. This could be useful for research or R&D purposes, as to identify gaps and opportunities in the field of Rare Diseases, but also in the public health sector, as for example with the OrphaCode which is more and more inserted in the national healthcare information systems. This tutorial is made to show you our current portfolio, how you could contribute to improve its quality and the coverage of data collected and produced by Orphanet and how we could help you to make the best use of this data for your own project.
THE OPENING SESSION

FRIDAY, 9 MAY 2014

09.00 – 09.45 I 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

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)
Tsventa 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 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.

**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 Sarnecki, 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 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 of ERNs in different Member States.
and management of European networks in the fields of healthcare and research, as well as possible future strategies to ensure sustainability of ERNs once established.

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

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

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

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

Session 0104
Saturday, 10 May, 11:30 – 13:00
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 Kaariainen, 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
THE ROLE, RISKS AND RELEVANCE OF REGISTRIES IN SHAPING THERAPY DEVELOPMENT TO 2020

Session Chair: Christine Laverty, 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
Lizzie Olszenco, Senior Scientific Advisor, NICE, UK

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
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, EUFORDIS, 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. This approach helps through multiple media. Is there still a place for the traditional telephone helpline or are the opportunities offered through email, the internet, and social networking sites of greater value and importance? In this session, we examine the issues involved and which sources and delivery 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 Iskov, Institute of Rare Diseases, Bulgaria

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 in point of care, specifically in an emergency setting, 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

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 bioclasters, 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 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-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

**Panel Discussion**

**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 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

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.

The theme will include presentations and panel discussions, enriched with the experience and expertise of the main stakeholders shaping the orphan landscape today.
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 Dooms, 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 Mavis, Director Therapeutic Development, EURORDIS, France
**Theme 5 | Emerging Concepts and Future Policies for Rare Disease Therapies**

**Jordi Llinares García**, 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 García**, 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

**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 García**, 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: Duchenne 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:**  
**Jaroslaw Waligora**, Policy Officer, Directorate-General for Health and Consumers, European Commission, EU

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

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

**Luca Pani**, Director General, Italian Medicines Agency (AIFA), Italy
Patient View on Progressive Patient Access Schemes
Pauline Evers, EGAn (European genetic alliances network / Dutch Federation of Cancer Patient Organisations), the Netherlands

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

Session 0504
Saturday, 10 May, 11:30 – 13:00
Room Amethyst

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

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

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

**Session 0601**
**Friday, 9 May, 14:00 – 15:30**
**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, Årgenska, 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

Session Chair:
Christoph Nachtigäler, 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, EURoDIS, Spain

Session 0606
Saturday, 10 May, 15:45 – 17:15
Room Saphir 2 & 3

HOW CENTRES OF EXPERTISE SHOULD/COULD INTERFACE WITH SOCIAL SERVICES

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.

THEMES

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 Grüters-Kieslich, Medical Director of the Charité Center for Women and Child Health and Human Genetics, Germany
BLUEPRINT GENETICS, Finland | Space 10

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

CENTOGENE, Germany | Space 6

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

COMRADIS, UK | Space 4

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

DNA GENOTEK, Canada | Space 1

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

INTERMUNE, Switzerland | Space 3

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. 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. For additional information about InterMune please visit: www.intermune.com.

MAPIGROUP, France | Space 7

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.

OPENAPP, Ireland | Space 5

Clinical Insight from OpenApp is a proven patient centric rare disease platform that supports clinical assessment & monitoring at patient, healthcare provider and health service levels. From a single patient encounter to a multi-national registry, Clinical Insight empowers providers of clinical programmes to monitor performance through intelligent capture of data, analytics and reporting.

ORION, UK | Space 2

ORION Clinical Services is a niche Clinical Research Organisation offering a full range of services in support of the clinical development process.

ORION has experience in many therapeutic areas but we have specialised in complex, hospital-based indications including oncology, HIV, sepsis in ITU patients, Acute Respiratory Distress Syndrome and many others.

QUINTILES, USA | Space 8

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. Visit our website at www.Quintiles.com.

REGULIANCE, USA | Space 9

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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