23-25 May 2012, Brussels

6th European Conference on Rare Diseases & Orphan Products

A better future for Patients: Shaping together the Agenda 2020

Executive Summary

A conference organised by

EURORDIS
Rare Diseases Europe

Co-organised by

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EURORDIS Membership Meeting 2013

31 May – 1 June 2013

Rixos Libertas Hotel, Dubrovnik, Croatia

The European Conference on Rare Diseases and Orphan Products

The European Conference on Rare Diseases & Orphan Products is the unique platform/forum across all rare diseases, across all European countries, bringing together all stakeholders - academics, health care professionals, industry, policy makers, patients' representatives.

It is a biennial event, providing the state-of-the-art of the rare disease environment, monitoring and benchmarking initiatives.

It covers research, development of new treatments, health care, social care, information, public health and support at European, national and regional levels.

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

Speaker presentations and poster abstracts can be found in the Programme section of the official Conference website at:

http://www.rare-diseases.eu/

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Rare Diseases Europe

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Acknowledgements and credits

Credits, support and legal information

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

Conference Organiser

The 6th European Conference on Rare Diseases & Orphan Products (ECRD 2012) is a conference organised by:

EURORDIS

Co-organised by

DIA Europe

With the support of

• European Commission, DG Health and Consumers' Protection, Executive Agency for Health and Consumers

• AFM Téléthon

In partnership with

• EU Committee of Experts on Rare Diseases

• European Medicines Agency

• ORPHANET

• NORD (National Organization for Rare Disorders USA)

• European Society of Human Genetics

• EuropaBio-EBE

The responsibility of the content and programme of the 6th European Conference on Rare Diseases lies with the speakers and Programme Committee. The Executive Agency is not responsible for any use that may be made of the information contained therein.

Continuing Education

This conference has been 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. All participants are eligible for these credits.

Introduction

Programme Committee Co-Chairs

Prof. Kate Bushby
Honorary Consultant Geneticist, Newcastle upon Tyne Hospitals NHS Foundation Trust, Joint co-ordinator, TREAT-NMD network, Vice President EUCERD, UK

Ms Avril Daly
Chair Rare Diseases Ireland (GRDO), CEO Fighting Blindness, Board Member and Officer EURORDIS, Ireland

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Chair EUCERD, France

Yann Le Cam
Chief Executive Officer, EURORDIS,
Vice Chair EUCERD, France

Ms Catarina Edfjaell
VP, Head of EU and International Regulatory Affairs, Shire HGT, Member of DIA Advisory Council Europe, Switzerland

Ms Lesley Greene
Patient Representative COMP, CLIMB, UK

Mr Torben Gronnebaek†
Rare Disorders Denmark, EUCERD Member, EURORDIS Board Member, Past Co-Chair ECRD 2010, Denmark

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Research Professor at National Institute of Health and Welfare, Helsinki, Vice Chair EUCERD, Finland

Prof. Dr. Milan Macek
Chair ESHG, Member EUCERD, Department of Biology and Medical Genetics, Department of Molecular Genetics and National Cystic Fibrosis Center, Czech Republic

Dr Mirando Mrsic
Minister of Labour and Pension System, Croatian Society of Patients with Rare Diseases, Former EURORDIS Board Member, Croatia

Dr Gabor Pogany
President Rare Diseases Hungary, Member EUCERD, Hungary

Prof. Josep Torrent i Farnell
General Director Fundacio Doctor Robert, COMP, Past Co-Chair ECRD 2010 Krakow & ECRD 2007 Lisbon, Member of the Committee of Orphan Medicinal Products, Spain

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Head of Orphan Medicines at the European Medicines Agency (EMA), EU

Mr Kevin Loth
Vice Chair Task Force Rare Diseases & Orphan Drugs of EuropaBio-EBE, EUCERD Member, Senior Director, External Relations, Europe, Celgene, UK

Prof. Dr Gert Matthijs
Head of the Laboratory for Molecular Diagnostics at the Centre for Human Genetics in Leuven, EUROGENTEST, EUCERD Member, Belgium

Ms Rosa Sanchez de Vega
Spanish Association Aniridia, Past President FEDER, EURORDIS Board Member, Spain

Mr Cees Smit
European Haemophilia Consortium, The Netherlands

Dr Domenica Taruscio
Director National Centre for Rare Diseases, European Platform for Rare Disease Registries (EPIRARE), Italy

Dr Rainald von Gizycki
Retina Europe, ECRD 2010 Programme Committee Member, EURORDIS Volunteer, Germany

orphanet Inserm

Orphanet is the reference portal for information on rare diseases and orphan drugs offering a large range of freely accessible services for all audiences. Its aim is to help improve the diagnosis, care and treatment of patients with rare diseases, and to provide information on developments in research and new therapies:
http://www.orpha.net

orphaNews Europe
Newsletter of the European Union Committee of Experts on Rare Diseases

Orphanews Europe is the freely available, twice-monthly electronic newsletter of the European Union Committee of Experts on Rare Diseases (EUCERD) that reports the latest scientific and political developments in the field of rare diseases and orphan drugs:
www.orphanews.org

EUCERD
European Union Committee of Experts on Rare Diseases

The European Union Committee of Experts on Rare Diseases was formally established on November 2009. Its mission is to aid the European Commission with the preparation and implementation of Community activities in the field of rare diseases.

www.eucerd.eu

EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

The European Medicines Agency (EMA) is a decentralised body of the European Union. Its main responsibility is the protection and promotion of public and animal health, through the evaluation and supervision of medicines for human and veterinary use.

http://www.ema.europa.eu

Key features of the conference

649 participants – 45 countries – 6 languages

Highlights of the successful 6th European Conference on Rare Diseases & Orphan Products (ECRD 2012) include:

•Highest participation since the first ECRD 2001 with 649 participants on-site,
•Participant representation from a record 45 countries including 26 from EU/EEA,
•17 of the 45 countries represented were Eastern European States,
•A comprehensive conference programme: 131 speakers/chairs, 36 sessions, 155 posters,
•A convivial event with ample time allocated to networking.

Participation

A total of 700 participants pre-registered for the conference. 649 participants actually attended the event (as compared to 545 in 2010, 420 in 2007 and 320 in 2005), including 131 speakers and session chairs. 40 full patient fellowships were awarded.

The participants at ECRD 2012 represented 45 countries (as compared to 41 in 2010, 35 in 2007 and 24 in 2005), including 25 from the EU and 26 from the EEA. Croatia as an acceding country to the EU was also represented. The total number of participants attending from EU countries was 521. This corresponds to 80% of the overall attendees. Participants attending from EEA countries corresponded to 81% of the overall number of attendees (526 persons) and 2 representatives attended the Conference from Croatia.

Representatives from 17 Eastern European states (Armenia, Bosnia and Herzegovina, Slovakia, Bulgaria, Russia, Czech Republic, Estonia, Hungary, Poland, Romania, Belarus, Serbia) were present at the Conference. In total, participants from Eastern Europe represented 13.3% of the attendance.

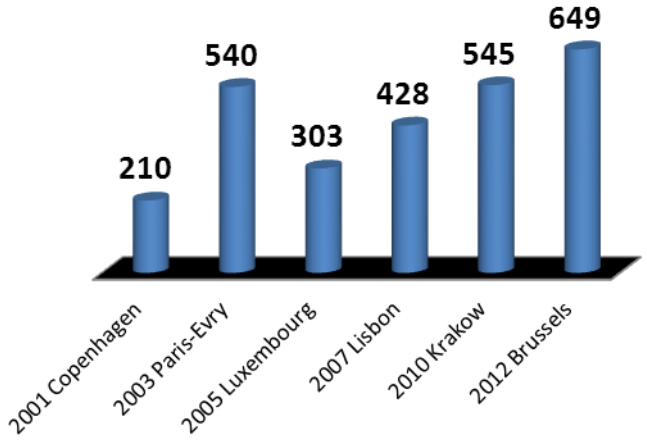
Non-European participation from USA, Canada, Japan, Russia and Argentina represented 8% of the total attendance.

By country of origin, Belgium had the largest delegation, representing 12.5% of participants (81 people). Mobilisation of Belgian patient organisations, interpretation of selected sessions in French, as well as special registration fees, were key factors for this high Belgian presence.

The other countries with an important presence at the Conference included France (78 participants), UK (55 participants), The Netherlands (51 participants), Germany (48 participants), Switzerland (43 participants), Italy (41 participants), USA (27 participants) and Spain (25 participants). This can be explained in part by the interpretation of the Opening & Plenary sessions from English into 5 languages and other selected sessions interpreted into French. The close proximity and ease of access to Belgium from these neighboring countries – with the exception of the USA – was also a contributing factor to this high presence.

Interpretation of the Opening and Plenary Sessions in 6 languages (English, French, German, Dutch, Spanish and Russian) was undoubtedly an important factor for registration: Overall, delegates originating from countries where these languages dominate represented 70% of all delegates.

The evolution of the total number of participants to the European Conference on rare Diseases since 2001 is as follow:



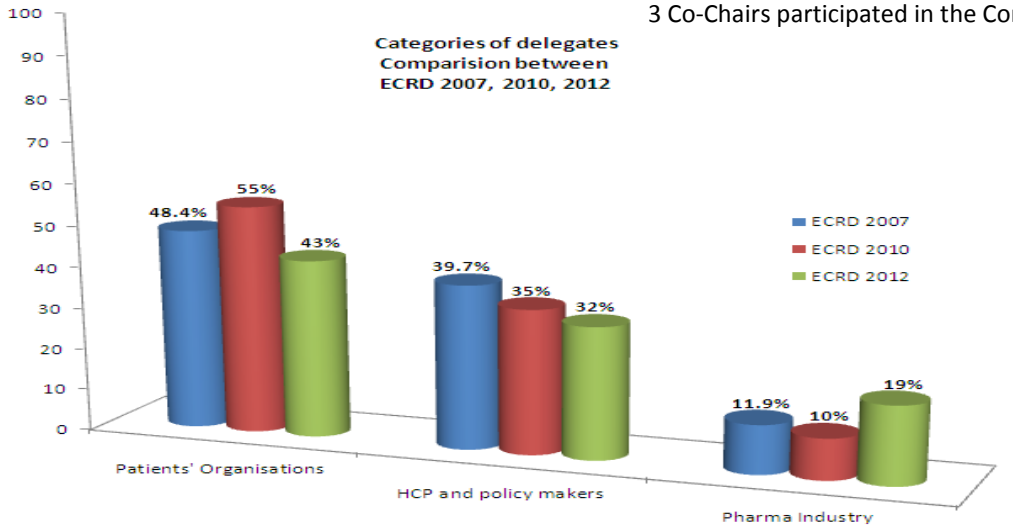
Who were the participants?

In terms of participants' categories, the composition of ECRD 2012 Brussels differed slightly compared to previous events. This evolution was projected and can be considered a direct result of the enhanced conference format and enlarged partnership. More representatives from the pharmaceutical and health industry were present (19% versus 10% in 2010 and 11% in 2007). There were proportionally fewer patient representatives than in previous years (43% versus 55% in 2010 and 48.4% in 2007). The proportion of health care professionals was more or less stable (32%).

The 131 Conference speakers and session chairs represented a

All of the official Conference partners were represented among the speakers and session chairs, including the EUCERD, EMA/COMP, EBE and EuropaBio, Orphanet, the Drug Information Association (DIA) and the European Society of Human Genetics (ESHG). In particular, 11 representatives on the Programme Committee / Advisors to the Programme Committee were members of the EUCERD.

In addition, 3 overview presentations were given during the Plenary Session based on information contained in the Annual Report of the « State of the Art of Rare Diseases in Europe » of the EUCERD prepared by the Joint Action. 25 Speakers and/or Session Chairs from EUCERD including the 3 Co-Chairs participated in the Conference.



Country	Number of delegates	%	Country	Number of delegates	%
Argentina	4	0.6%	Macedonia	1	0.1%
Armenia	1	0.1%	Malta	1	0.1%
Austria	7	1.1%	Netherlands	51	8%
Australia	1	0.1%	Norway	5	0.8%
Belarus	3	0.5%	Poland	11	1.7%
Belgium	81	12.5%	Portugal	3	0.5%
Bosnia	1	0.1%	Romania	17	2.6%
Bulgaria	4	0.6%	Russia	14	2.2%
Canada	4	0.6%	Serbia	1	0.1%
Croatia	2	0.3%	Slovakia	7	1.1%
Cyprus	2	0.3%	Slovenia	1	0.1%
Czech R.	5	0.8%	Spain	25	3.9%
Denmark	16	2.5%	Sweden	19	2.9%
Estonia	2	0.3%	Switzerland	43	6.6%
France	78	12.2%	Thailand	1	0.1%
Finland	17	2.6%	Turkey	1	0.1%
Georgia	4	0.6%	UK	55	8.5%
Germany	48	7.4%	Ukraine	2	0.3%
Greece	4	0.6%	USA	27	4.2%
Hungary	11	1.7%	Venezuela	1	0.1%
Ireland	10	1.6%	Europe	4	0.6%
Israel	1	0.1%	Not Specified	5	0.8%
Italy	41	6.3%			
Japan	2	0.3%			
Luxembourg	5	0.8%			
			Total	649	100.0%

Programme at a glance

ECRD 2012

Wednesday 23 May 2012 (Afternoon)			
14:00-17:30	**Tutorial 1** Orphan Drug Development	**Tutorial 2** Orphan Drugs in the EU: From Designation to Marketing Authorisation	**Tutorial 3** HTA 101 for Rare Diseases
Thursday 24 May 2012 (Morning)			
09:00-09:45	**Opening Session**		
10:15-12:00	**Plenary Session**		

	Theme 1 National Plans for Rare Diseases	**Theme 2** Centres of Expertise and European Reference Networks	**Theme 3** Information and Public Health
Thursday 24 May 2012 (Afternoon)			
Session 1 **14:00-15:30**	The latest information about National Plans for Rare Diseases have been integrated into sessions throughout the programme	**Session 0201** Introduction of Centres of Expertise	**Session 0301** Making Rare Diseases Visible in Society
Session 2 **16:30-18:00**	The latest information about National Plans for Rare Diseases have been integrated into sessions throughout the programme	**Session 0202** Introduction of European Reference Networks (ERNs)	**Session 0302** Making Rare Diseases Visible for Research & Public Health
Friday 25 May 2012 (Morning)			
Session 3 **09:00-10:30**	The latest information about National Plans for Rare Diseases have been integrated into sessions throughout the programme	**Session 0203** Cross-Border Health Care: Samples Mobility	**Session 0303** Improving Care through Clinical Guidelines
Session 4 **11:00-12:30**	The latest information about National Plans for Rare Diseases have been integrated into sessions throughout the programme	**Session 0204** Health Care Pathways Focusing on Transition from Childhood to Adulthood	**Session 0304** New Approaches for Training and Awareness
Friday 25 May 2012 (Afternoon)			
Session 5 **13:30-15:00**	The latest information about National Plans for Rare Diseases have been integrated into sessions throughout the programme	**Session 0205** Cross-Border Health Care: Patient Mobility	**Session 0305** Fostering Early Diagnosis & Prevention
Session 6 **15:00-16:30**	The latest information about National Plans for Rare Diseases have been integrated into sessions throughout the programme	**Session 0206** Oral Poster Session	**Session 0306** Primary and Secondary Prevention

AT A GLANCE

Tutorial 4 Registries			
Opening Session			
Plenary Session			

Theme 4 Research from Bench to Bedside	**Theme 5** Orphan Products & Rare Disease Therapies: Access	**Theme 6** Orphan Products & Rare Disease Therapies: Regulatory	**Theme 7** Patients' Empowerment
Session 0401 How Rare Disease Research can contribute to Innovation	**Keynote session 0501/0601** The Big Picture		**Session 0701** Empowering Patients and Their Families
Session 0402 The Big Picture of Rare Disease Research Policy	**Keynote Session 0502/0602** The Value and Specificity of the Rare Disease Business Model		**Session 0702** Empowerment at a Political Level
Session 0403 EU Infrastructure & Projects in the Field of Rare Diseases & Patient Registries	**Session 0503** EU Policy Developments in the Field of Access to Orphan Drugs	**Session 0603** Deployment of the Orphan Drug Regulation: Predictable Flexibility	**Session 0703** Organisational Level: Patient's Generated Knowledge in Practice
Session 0404 Breakthroughs in Research in the Field of Rare Diseases: New Genetic Diagnostics	**Session 0504** Novel Reimbursement Schemes as a Potential Way Forward	**Session 0604** Compassionate Use Programmes	**Session 0704** Projects that Empower and Inspire: Examples of the added value of working, learning and acting together
Session 0405 Breakthroughs in Research in the Field of Rare Diseases: Therapeutics	**Session 0505** Ways to look at HTA for Orphan Drugs and Rare Diseases	**Session 0605** Treatment of Rare Cancers	**Session 0705** Personal Level: The forum of "Patient Groups Innovation"
Session 0406 Oral Poster Session	**Session 0506** The Involvement of Patients in the Product Life-Cycle	**Session 0606** Oral Poster Session	**Session 0706** Oral Poster Session

Conference Programme

Opening Session

Thursday, 24 May 2012 | 09:00 – 09:45 |

Session Chair:
Avril Daly, Chair Rare Diseases Ireland (GRDO), CEO Fighting Blindness,
Board Member and Officer EURORDIS, Ireland

Welcome Remarks by conference co-organisers
Avril Daly, Chair Rare Diseases Ireland (GRDO), CEO Fighting Blindness,
Board Member and Officer EURORDIS, Ireland

Dr Brigitte Franke-Bray, Director, Global Training Officer DIA Europe, Switzerland

Keynote Addresses:
European Commission Action on Rare Diseases. John Dalli, Commissioner for Health and Consumer Policy, EU

Dr Jytte Lyngvig, Director of the Danish Health & Medicines Authority, Denmark
Representative from Danish EU Presidency.

Terkel Andersen, President EURORDIS, Denmark

Plenary Session

Thursday, 24 May 2012 | 10:15 – 12:00 |

Session Chair:
Avril Daly, Chair Rare Diseases Ireland (GRDO), CEO Fighting Blindness, Board Member and Officer EURORDIS, Ireland

Overview Focusing on EU Policy at large and based on the Annual Report of the "State of the Art of Rare Disease Activities in Europe" of the EUCERD prepared by the Joint Action.
Dr Ségolène Aymé, Chair EUCERD, France

State of the Art around the World. An Overview of the International Landscape Including Emerging Countries.
Durhane Wong Rieger, President, Canadian Organisation for Rare Disorders (CORD), Canada

Dynamic of National Plans in EU Member States based on the "State of the Art of Rare Disease Activities in Europe" of the EUCERD prepared by the Joint Action.
Dr Edmund Jessop, Medical Advisor, National Health Service, UK

Cutting edge national strategy case studies:
Germany**:** Dr Andreas Reimann, Mukoviszidose EV National Action League on Rare Diseases, Germany

France**:** Alain Garcia, Social Affairs Inspectorate General (IGAS), 2nd French National Plan for Rare Diseases, EUCERD Member, France

Belgium: Ri De Ridder, Director General at RIZIV-INAMI, Belgium

Broad Overview of the main themes addressed during the conference and highlights,
Avril Daly, Chair Rare Diseases Ireland (GRDO), CEO Fighting Blindness, Board Member and Officer EURORDIS, Ireland

Theme 1 | National Plans for Rare Diseases

Prof. Josep Torrent i Farnell, General Director Fundacio Doctor Robert, COMP, Past Co-Chair ECRD 2010 Krakow & ECRD 2007 Lisbon, Member of the Committee of Orphan Medicinal Products, Spain

Dr Domenica Taruscio, Director National Centre for Rare Diseases, European Platform for Rare Disease Registries (EPIRARE), Italy

The latest information about National Plans for Rare Diseases have been integrated into sessions throughout the programme

Theme 2 | Centres of Expertise and European Reference Networks

Prof. Helena Kääriäinen, Research Professor at National Institute of Health and Welfare, Helsinki, Vice Chair, EUCERD, Finland

Dr Ségolène Aymé, Chair EUCERD, France

The "Council Recommendation on action in the field of rare diseases" issued on 9 June 2009 recommended to the Member States to elaborate and adopt a national plan or strategy, preferably by the end of year 2013, including the identification and support to appropriate centres of expertise as nodes to establish, when necessary, European Reference Networks. Theme 2 introduces the concepts for National Centres of Expertise and European Reference Networks and the EUCERD criteria for establishing them. The possibilities and problems related to patient and sample mobility as part of cross-border healthcare are presented and discussed. Finally, the solutions focusing on transition from childhood to adulthood in the field of rare diseases are tackled.

Session 0201|Thursday, 24 May 2012, 14:00-15:30 Room A

Interpretation FR + RU

INTRODUCTION OF CENTRES OF EXPERTISE

Session Chair:

Alastair Kent, Director, Genetic Alliance UK, Chair, Rare Diseases UK, President at EGAN, UK

Overview of Current Situation: Where do we stand in Europe (scoping paper) and process to designate centres (how does the process work)

Dr Ségolène Aymé, Chair EUCERD, France

Recommendations

Dr Edmund Jessop, Medical Advisor, National Health Service, UK

Panel/Roundtable Discussion

- Dr Véronique Heon-Klin, Federal Ministry of Health, Germany
- Prof. Helena Kääriäinen, Research Professor at National Institute of Health and Welfare, Helsinki, Vice Chair, EUCERD, Finland
- Dr Odile Kremp, Director ORPHANET-INSERM US14, France

Session 0202|Thursday, 24 May 2012, 16:30-18:00 Room A

Interpretation FR + RU

INTRODUCTION OF EUROPEAN REFERENCE NETWORKS (ERNS):

Session Chair:

Kay Parkinson, Alstrom Syndrome UK (ASUK)

European Reference Network under the Framework of the Directive on the Application of Patient's Rights in Cross-border Healthcare: State of the Art

Enrique Terol, Healthcare Systems Unit, DG SANCO, European Commission, Belgium

Overview – Where do we stand – Progress to date – EUCERD recommendations

Prof Kate Bushby, Honorary Consultant Geneticist, Newcastle upon Tyne Hospitals NHS Foundation Trust, Joint co-ordinator, TREAT-NMD network, Vice President, EUCERD, UK

Criteria for Prioritisation and Quality Criteria

Prof. Olaf Hiort, Professor of Paediatrics, Paediatric Endocrinologist, Molecular Genetics of Endocrinological Disorders, University of Lübeck, Germany

Session 0203 | Friday, 25 May 2012, 09:00-10:30 Room A

Interpretation FR + RU

CROSS-BORDER HEALTHCARE: SAMPLES MOBILITY

Session Chair:

Prof. Helena Kääriäinen, Research Professor at National Institute of Health and Welfare, Helsinki, Vice Chair, EUCERD, Finland

Where Do we Stand with the Directive on In Vitro Diagnostics and their Consequences for Rare Diseases
Prof. David E. Barton, National Centre for Medical Genetics, Ireland

Cross-Border Genetic Testing: Issues and solutions
Prof. Jean-Jacques Cassiman, Center Human Genetics, Forensic Medicine, Leuven, Belgium

External Quality Assessment of Genetic Testing at EU Level: A necessity for quality of care
Prof. Dr Els Dequeker, EuroGenTest, Quality Manager Division Diagnostic UZ Leuven Biomedical Quality Assurance Research Unit, Belgium

Session 0204 | Friday, 25 May 2012, 11:00-12:30 Room A

Interpretation FR + RU

HEALTH CARE PATHWAYS FOCUSING ON TRANSITION FROM CHILDHOOD TO ADULTHOOD

Session Chair:
Dorica Dan, President Romanian Prader Willi Association, Romanian National Alliance for Rare Diseases, Board Member and Officer EURORDIS, Romania

Transition from Childhood to Adulthood
Prof. Hanns Lochmüller, Chair of Experimental Myology, Institute of Genetic Medicine, Newcastle University, UK

European Paediatric Academy
Dr Liesbeth Siderius, Representative of Shwachman diamond Syndrome Support Holland, Paediatrician, Youth Health Care, Coordinator Rare disease working group European Academy of Paediatrics, The Netherlands

Session 0205 | Friday, 25 May 2012, 13:30-15:00 Room A

Interpretation FR + RU

CROSS-BORDER HEALTH CARE: PATIENT MOBILITY

Session Chair:
Flaminia Macchia, Director of European Public Affairs, EURORDIS, Belgium

Introduction and Implementation of the EU Directive on Cross-Border Health Care
Nathalie Chaze, Head of Unit, Healthcare Systems, DG SANCO, European Commission, Belgium

The Experience of Denmark
Marianne Jespersen, Member of the Rare Diseases Task Force, Member of the Working Group on Standards of Care, National Board of Health , Denmark

Expectations from Patients
Flaminia Macchia, Director of European Public Affairs, EURORDIS, Belgium

Session 0206 | Friday, 25 May 2012, 15:00-16:30 Room A

Interpretation FR + RU

ORAL POSTER SESSION

Session Chair:
Christel Nourissier, Prader Willi France, General Secretary, EURORDIS

The Hunter Outcome Survey (HOS), a Value-Adding Disease Registry (82)
Maria Paabol Larsen, Shire, Denmark

Scope of Centres of Expertise for Rare Diseases in European Countries Where they Exist (96)
Charlotte Rodwell, Scientific Seceteriat of the EUCERD, France

Drafting a National Plan for Rare Diseases in Germany by Concerted Action: (117)
The National Action League for People with Rare Diseases
Dr Véronique Héon-Klin, Federal Ministry of Health, Germany

Theme 3 | Information and Public Health

Avril Daly, Chair Rare Diseases Ireland (GRDO), CEO Fighting Blindness, Board Member and Officer EURORDIS, Ireland
Dr Ségolène Aymé, Chair EUCERD, France

Making rare diseases visible in society is a challenge which has now good reasons to be met. The public at large can be involved with appropriate initiative which already proved their efficacy. The interest of researchers is enhanced by the availability of organised data freely accessible on the web.
The healthcare practitioners now have sources of information on good practice guidelines to guide them, and information technologies allow innovative teaching approaches to train online. This will foster early diagnosis and secondary prevention.

Session 0301 | Thursday, 24 May 2012, 14:00-15:30
Room E + F

MAKING RARE DISEASES VISIBLE IN SOCIETY

Session Chair:

Avril Daly, Chair Rare Diseases Ireland (GRDO), CEO Fighting Blindness, Board Member and Officer EURORDIS, Ireland

Rare Disease Day (Highlighting Community Level and National Level Initiatives)

Avril Daly, Chair Rare Diseases Ireland (GRDO), CEO Fighting Blindness, Board Member and Officer EURORDIS, Ireland

The French Téléthon (Overview of the past 25 years, how it is organised, the way information is disseminated and the legal framework behind it)

Monique Karkatcharian, Communications & HR Director, AFM Généthon, France

Resources Available Online: The web 2.0 experience

Denis Costello, Web Communications Senior Manager and Rare Connect Leader, EURORDIS, Spain

Panel Discussion: Focusing on Powerful Tools for Broad Dissemination

Session 0302 | Thursday, 24 May 2012, 16:30-18:00
Room E + F

MAKING RARE DISEASES VISIBLE FOR RESEARCH & PUBLIC HEALTH

Session Chair:

Dr Ségolène Aymé, Chair EUCERD, France

Classification and Coding of Rare Diseases: Overview of where we stand, rationale, why it matters and what it can change

Dr Peter Robinson, Institute of Medical and Human Genetics, Charité Berlin, Germany

Speeding up Research Using the Semantic Web

Marco Roos, BioSemantics group, Human Genetics department, Leiden University Medical Centre, Belgium

Solving Bottlenecks in Data Sharing

Richard Kidd, Manager Informatics, Fellow of the Royal Society of Chemistry, UK

Panel Discussion: Availability of Data and the Challenge of Making Data Freely Accessible

Session 0303 | Friday, 25 May 2012, 09:00-10:30 Room H

IMPROVING CARE THROUGH CLINICAL GUIDELINES

Session Chair:

Dr Odile Kremp, Director ORPHANET-INSERM US14, France

Professional Clinical Guidelines: Methodology and impact

Dr Odile Kremp, Director ORPHANET-INSERM US14, France

How Reference Networks Develop, Implement and Monitor Guidelines (Methodology)

Dr Janbernd Kirschner, Consultant Paediatric Neurologist, Uniklinik Freiburg (TREAT-NMD and CARE-NMD), Germany

The Involvement of Patients in Developing Clinical Guidelines

Kay Parkinson, CEO Alstrom Syndrome, UK

Session 0304 | Friday, 25 May 2012, 11:00-12:30 Room H

NEW APPROACHES FOR TRAINING AND AWARENESS

Session Chair:

Dr Rainald von Gizycki, Retina Europe, ECRD 2010 Krakow Programme Committee Member, EURORDIS Volunteer, Germany

WeBSurg.com, a web-based surgical university dedicated to MIS surgery

Thomas Parent, Chief Technical Officer, WebSurg, France

Training of Medical Students on the Issues of Rare Diseases

Dr Paula Byrne, Senior Lecturer, Conway Institute, UCD School of Medicines, UCD College of Health Sciences, University College Dublin, Ireland

The Italian Model

Simona Bellagambi, UNIAMO Italy

Panel Discussion

Inge van Niewerburgh, Digital projects manager Ghent University Library, project partner EC project OpenAIRE, Belgium

Session 0305 | Friday, 25 May 2012, 13:30-15:00 Room H

FOSTERING EARLY DIAGNOSIS AND PREVENTION

Session Chair:

Anne-Sophie Lapointe, President, Vaincre les maladies lysomales, France

Part 1: Fostering Early Diagnosis

State of the Art of Screening in Europe and Recommendations

Dr Peter Burgard, Centre for Pediatric and Adolescent Medicine Dept. of General Pediatrics, University of Heidelberg, Germany

HTA on Neonatal Screening for Rare Metabolic Disorders Faced Misconceptions and Blurred Objectivity

Prof. Ilona Autti-Rämö, Chief of Health Research, Research Professor, The Social Insurance Institution Research Department, Finland

Panel Discussion: Patient Perspective and Expectations

Anne-Sophie Lapointe, President, Vaincre les maladies lysomales, France

Session 0306 | Friday, 25 May 2012, 15:00-16:30 Room H

PRIMARY AND SECONDARY PREVENTION

Session Chair:

Avril Daly, Chair Rare Diseases Ireland (GRDO), CEO Fighting Blindness, Board Member and Officer EURORDIS, Ireland

Genetic Counselling for Family Planning

Prof. Helena Kääriäinen, Research Professor at National Institute of Health and Welfare, Helsinki, Vice Chair, EUCERD, Finland

The Spina Bifida Example

Pierre Mertens, President, International Federation for Spina Bifida and Hydrocephalus (IF), Belgium

The Porphyria Example

Samantha Parker, Director of External Affairs and Rare Disease Partnerships, Orphan Europe, France

Theme 4 | Research from Bench to Bedside

Prof Kate Bushby, Honorary Consultant Geneticist, Newcastle upon Tyne Hospitals NHS Foundation Trust, Joint co-ordinator, TREAT-NMD network, Vice President, EUCERD, UK

Prof. Dr Milan Macek, Chair ESHG, Member EUCERD, Department of Biology and Medical Genetics, Department of Molecular Genetics and National Cystic Fibrosis Center, Czech Republic

Prof. Dr Gert Matthijs, Head of the Laboratory for Molecular Diagnostics at the Center for Human Genetics in Leuven, EUROGENTEST, EUCERD Member,
Belgium

There are specific issues which are relevant to rare diseases when considering research that is likely to lead to changes in diagnostics or therapy development (so called translational research). This theme will address the political climate for funding translational research in RD, as well as examples of the resources which have been successfully developed to overcome some of the particular barriers for these disorders. Finally we will review some areas of outstanding scientific progress where translation to the clinic can be foreseen and where trials of personalised medicines have become a reality.

Session 0401 | Thursday, 24 May 2012, 14:00-15:30 Room G

HOW RARE DISEASE RESEARCH CAN CONTRIBUTE TO INNOVATION

Session Chair:

Dr. Ilro Eerola, EC Project Officer, European Commission, EU

Potential Link between Gaucher Pathways and those of Parkinson's Disease

Dr Hanna Rosenbaum, Head of Hematology Day Care Unit, Hematology and Bone Marrow transplantation Department Rambam Health Care Campus Haifa, Israel

FDA Perspective

Dr Anne Pariser, Associate Director for Rare Diseases Office of New Drugs CDER, FDA, USA

How AFM has Succeeded to Encourage and Promote Rare Disease Research and Innovation

Dr Serge Braun, Scientific Director, French Association against Myopathies (AFM), France

Session 0402 | Thursday, 24 May 2012, 16:30-18:00
Room G

THE BIG PICTURE OF RARE DISEASE RESEARCH POLICY

Session Chair:

Prof. Dr Milan Macek, Chair ESHG, Member EUCERD, Department of Biology and Medical Genetics, Department of Molecular Genetics and National Cystic Fibrosis Center, Czech Republic

FP7 and FP8 Outlook and IRDiRC

Dr. Ilro Eerola, EC Project Officer, European Commission, EU

Canadian Policy on Rare Disease Research

Dr Paul Lasko, Department of Biology, McGill University, Canada

French Rare Disease Foundation - How to address rare disease research issues at a national level

Prof. Nicolas Lévy, Univ Prof, Head of a research team on laminopathies and neuromuscular disorders at INSERM U491, Head of the Laboratory of Molecular Genetics of the Department of Medical Genetics at the Children's Hospital La Timone

Panel Discussion: Discussion of RD Funding Mechanism

Session Speakers and Sophie Koutouzov, Secretary General, eRare-Coordnator, GIS Instut des Maladies Rares, France

Session 0403 | Friday, 25 May 2012, 09:00-10:30 Room B

Interpretation FR + RU

EU INFRASTRUCTURE & PROJECTS IN THE FIELD OF RARE DISEASES & PATIENT REGISTRIES

Session Chair:

Antoni Montserrat Moliner, Policy Officer for Rare and Neurodevelopmental Diseases Health and Consumers General-Directorate (SANCO) Directorate C 'Public Health', Unit C-2 European Commission, Belgium

Disease Specific Registries vs Product Registries

Prof. Hanns Lochmüller, Chair of Experimental Myology, Institute of Genetic Medicine, Newcastle University, UK

State of Play of Registries in Europe

Dr Domenica Taruscio, Director National Centre for Rare Diseases, European Platform for Rare Disease Registries (EPIRARE), Italy

Panel Discussion:

Prof. Hanns Lochmüller, Chair of Experimental Myology, Institute of Genetic Medicine, Newcastle University, UK

Dr Anil Mehta, Reader in Molecular Medicine Division of Maternal & Child Health Sciences, University of Dundee, UK

Session 0404 | Friday, 25 May 2012, 11:00-12:30 Room B

Interpretation FR + RU

BREAKTHROUGHS IN RESEARCH IN THE FIELD OF RARE DISEASES: NEW GENETIC DIAGNOSTICS

Session Chair:

Prof. Dr Gert Matthijs, Head of the Laboratory for Molecular Diagnostics at the Center for Human Genetics in Leuven, EUROGENTEST, EUCERD Member, Belgium

Example of Genetic Diagnostics in a Real Life Setting

Prof. David E. Barton, National Centre for Medical Genetics, Ireland

Technological Advances in Next Generation Sequencing

Dr Joris Veltman, Associate Professor Genomic Disorders, Department of Human Genetics, Radboud University, The Netherlands

Next Generation Sequencing: A Policy Perspective

Prof. Dr Koen Devriendt, Center for Human Genetics, KU Leuven, Belgium

Session 0405 | Friday, 25 May 2012, 13:30-15:00 Room B

BREAKTHROUGHS IN RESEARCH IN THE FIELD OF RARE DISEASE THERAPEUTICS

Session Chair:

Prof Kate Bushby, Honorary Consultant Geneticist, Newcastle upon Tyne Hospitals NHS Foundation Trust, Joint co-ordinator, TREAT-NMD network, Vice President, EUCERD, UK

Finding New Medicines to Fight CF: Multiple steps of a success story"

Prof. Margarida Amaral, Faculty of Sciences, University of Lisbon, Portugal

Exon Skipping for Treating Rare Diseases

Annemieke Aartsma-Rus, DMD Genetic Therapy Group Department of Human Genetics Leiden University Medical Center, CF, The Netherlands

Panel Discussion: Success Factors in RD Translation Research

Session 0406 | Friday, 25 May 2012, 15:00-16:30 Room B

ORAL POSTER SESSION

Session Chair:

Prof. Hanns Lochmüller, Chair of Experimental Myology, Institute of Genetic Medicine, Newcastle University, UK

From Front Room to Research Laboratory - How Alstrom Syndrome UK madethat journey (25)

Dr Mike Hales, Alström Syndrome International, UK

Italian Research on Genetic Diseases: Worthy ideas deserve care (364)

Dr. Francesca Sofia, Research Program Manager Comitato telethon Fondazione ONLUS, Italy

Theme 5 | Orphan Products and Rare Disease Therapies: Access

Wills Hughes-Wilson, Chair of Task Force RDs & ODs of EuropaBio-EBE, Member EUCERD, Vice President Global Public Policy & Government Relations Swedish Orphan Biovitrum (sobi), Member of DIA Advisory Council Europe, Belgium

Yann Le Cam, Chief Executive Officer, EURORDIS, Vice Chair EUCERD, France

Authorised safe and effective Orphan Medicinal Products to treat rare diseases are – where available – a key element in our approach to diagnosing, treating or even preventing rare diseases. But as more such treatments come to market, and against a backdrop of / in times of economic uncertainty, costs of treating often find themselves under the spotlight. Where do we go from here in ensuring that the interest in orphan drugs continues in a way that is sustainable for all stakeholders? Initiatives at a country-level, European level and even global level are pointing out the potential pathways for the orphan drug and rare disease community. Are there lessons we can learn by cooperating across borders or even across regions? And are there lessons the healthcare systems can learn for approaching other, innovative treatment solutions in an increasingly targeted healthcare environment?

Session 0501/0601 | Thursday, 24 May 2012, 14:00-15:30 Room B

Interpretation FR + RU

KEYNOTE - THE BIG PICTURE (TOGETHER WITH THEME 6)

Session Chair:

Josep Torrent i Farnell, General Director, Fundacio Doctor Robert, COMP, Past Co-Chair ECRD 2010 Krakow & ECRD 2007 Lisbon, Member of the Committee of Orphan Medicinal Products, Spain

Implementation of the OD Regulation and Consistency with the Policy on Rare Diseases

Paola Testori Coggi, Director General of the Directorate-General for Health and Consumers of the European Commission, Belgium

Historic Achievements and New Paradigms for Development and Access to Orphan Drugs

Yann Le Cam, Chief Executive Officer, EURORDIS, Vice Chair EUCERD, France

Looking to the Future: What's next?

Prof. Guido Rasi, Executive Director, European Medicines Agency, EU

Panel Discussion: Understanding the Global Trends for Orphan Drugs. How to align the different agencies/bodies worldwide

Panellists:

Dr Gayatri Rao, Acting Director, FDA Office of Orphan Products Development, USA

Peter Saltonstall, President & Chief Executive Officer, National Organization for Rare Disorders, USA

Session 0502/0602 | Thursday, 24 May 2012, 16:30-18:00 Room B

Interpretation FR + RU

KEYNOTE - THE VALUE AND SPECIFICITY OF THE RARE DISEASE BUSINESS MODEL (TOGETHER WITH THEME 6)

Session Chair:

Thomas Heynisch, European Commission, EU

Rare Disease Unit Setting up within big Pharma Company

Marc Dunoyer, Global Head, GlaxoSmithKline Rare Diseases, UK

Orphan Drugs & Rare Diseases as the new Paradigm?

Theresa Heggie, Senior Vice President, Global Commercial Operations, Shire Human Genetic Therapies, Switzerland

Point of View from a Dedicated Orphan Company
Philippe Van Holle, President, Celgene Europe, Switzerland

Session 0503 | Friday, 25 May 2012, 09:00-10:30
Room E+F

EU POLICY DEVELOPMENTS IN THE FIELD OF ACCESS TO ORPHAN DRUGS

Session Chair:
Yann Le Cam, Chief Executive Officer, EURORDIS, Vice Chair EUCERD, France

The Coordinated EU Approach to Access: CAVOD EC proposals
Wills Hughes-Wilson, member of the drafting group on EUCERD, Belgium

Mechanism of Coordinated Access (MOCA)
Dr Ri De Ridder, Director General at National Institute for Health and Disability Insurance (RIZIV-INAMI), Belgium

Session 0504 | Friday, 25 May 2012, 11:00-12:30
Room E+F

NOVEL REIMBURSEMENT SCHEMES AS A POTENTIAL WAY FORWARD

Session Chair:
Jakub Adamski, Chief Expert Drug Policy and Pharmacy Department Ministry of Health, Poland

Belgium Proposed System in Development under the Proposed National Plan
Françoise Stryckman, Pharma.be, Belgium
Dr Ri De Ridder, Director General at National Institute for Health and Disability Insurance (RIZIV-INAMI), Belgium

Risk-sharing Schemes: The Italian experience in the field of rare cancers
Luca De Nigro, Clinical Trials Unit, Drugs Monitoring Registers, Italian Medicines Agency, AIFA, Italy

Coverage with Evidence Development: The example of The Netherlands
Dr Ad Schuurman, CVZ & Chair of MEDEV, The Netherlands

Session 0505 | Friday, 25 May 2012, 13:30-15:00
Room E+F

WAYS TO LOOK AT HTA FOR ORPHAN DRUGS AND RARE DISEASES

Session Chair:
Wills Hughes-Wilson, Vice President, Global Public Policy & Government Relations, Sobi – Swedish Orphan Biovitrum, Belgium

IQWIG's Approach in Germany
Dr Stefan Lange, Deputy Director, Institute for Quality and Efficiency in Healthcare (IQWiG), Germany

A New Methodology for HTA Orphan Drugs – The Experience of AGNSS NHS
Josie Godfrey, Head of Policy and Coordination, AGNSS, NHS, UK

CVZ Experience
Dr Ad Schuurman, CVZ & Chair of MEDEV, the Netherlands

How to Consider Orphan Drugs in the Mix – Including multi-criteria decision making analysis
Prof. Steven Simoens, Chair of Pharmacoeconomics, Catholic University of Leuven, Belgium

Session 0506 | Friday, 25 May 2012, 15:00-16:30
Room E+F

THE INVOLVEMENT OF PATIENTS IN THE PRODUCT LIFE-CYCLE

Session Chair:
Dr Michele Lipucci Di Paola, Vice-President AVLT (Associazione Veneta Lotta alla Talassemia), EURORDIS Volunteer, Italy, Patients' Representative Committee Advanced Therapies/EMA

Involving Patient Advocacy Organisations in Research
Sharon Gibsztein, Norwegian Cystic Fibrosis Association, Norway

Collaboration Between Patient Advocacy Organisations and Sponsors of Clinical Trials for Rare Diseases
Susanna Leto di Priolo, Novartis Oncology Region Europe, Head of Patient Advocacy and Professional Relations, Italy

Involvement of Patient Representatives in Marketing Authorisation and Post-Marketing Authorisation Surveillance Processes
François Houÿez, Director of Health Policy, EURORDIS, France

Panel Discussion: Involvement in Reimbursement Issues
Dr Pauline Evers, Member of the COMP, Federation of Cancer Patients Organisations, The Netherlands

Theme 6 | Orphan Products and Rare Disease Therapies: Regulatory

Catarina Edfjaell, VP, Head of EU and International Regulatory Affairs, Shire HGT, Member of DIA Advisory Council Europe, Switzerland
Dr Jordi Llinares, Head of Orphan Medicines at the European Medicines Agency (EMA), EU
Prof. Kerstin Westermark, Senior Expert, Medical Products Agency, Adjunct Professor of Medicine at Uppsala University, Chairperson, COMP, EUCERD Member, Sweden

This regulatory theme will analyse the delivery of the OD Regulation, the success of its implementation and consistency with the policy on rare diseases, while also looking to the future for orphan drugs and how predictable regulatory flexibility could enhance orphan drug development. Regulators, patient organisations, academia and industry will share experiences in specific areas including compassionate use, paediatric patients and rare cancers. Opportunities and challenges resulting from the evolving regulatory landscape and global trends for orphan drugs will be discussed in panels including all stake holders.

Session 0601/0501 | Thursday, 24 May 2012, 14:00-15:30 Room B

Interpretation FR + RU

KEYNOTE - THE BIG PICTURE (TOGETHER WITH THEME 5)
Session Chair:
Josep Torrent i Farnell, General Director, Fundacio Doctor Robert, COMP, Past Co-Chair ECRD 2010 Krakow & ECRD 2007 Lisbon, Member of the Committee of Orphan Medicinal Products, Spain

Implementation of the OD Regulation and Consistency with the Policy on Rare Diseases
Paola Testori Coggi, Director General of the Directorate-General for Health and Consumers of the European Commission, Belgium

Historic Achievements and New Paradigms for Development and Access to Orphan Drugs
Yann Le Cam, Chief Executive Officer, EURORDIS, Vice Chair EUCERD, France

Looking to the Future: What's next?
Prof. Guido Rasi, Executive Director, European Medicines Agency, EU

Panel Discussion: Understanding the Global Trends for Orphan Drugs. How to align the different agencies/bodies worldwide
Panellists:
Dr Gayatri Rao, Acting Director, FDA Office of Orphan Products Development, USA
Peter Saltonstall, President & Chief Executive Officer, National Organization for Rare Disorders, USA

Session 0602/0502 | Thursday, 24 May 2012, 16:30-18:00 Room B

Interpretation FR + RU

KEYNOTE - THE VALUE AND SPECIFICITY OF THE RARE DISEASE BUSINESS MODEL (TOGETHER WITH THEME 5)
Session Chair:
Thomas Heynisch, European Commission, EU

Rare Disease Unit Setting up within big Pharma Company
Marc Dunoyer, Global Head, GlaxoSmithKline Rare Diseases, UK

Orphan Drugs & Rare Diseases as the new Paradigm?
Theresa Heggie, Senior Vice President, Global Commercial Operations, Shire Human Genetic Therapies, Switzerland

Point of View from a Dedicated Orphan Company
Philippe Van Holle, President, Celgene Europe, Switzerland

Session 0603 | Friday, 25 May 2012, 09:00-10:30 Room G

DEPLOYMENT OF THE ORPHAN DRUG REGULATION: PREDICTABLE FLEXIBILITY
Session Chair:
Prof. Kerstin Westermark, Senior Expert, Medical Products Agency, Adjunct Professor of Medicine at Uppsala University, Chairperson, COMP, EUCERD Member, Sweden

Successful Use of Existing Opportunities for Orphan Drug Approval
Catarina Edfjaell, VP, Head of EU and International Regulatory Affairs, Shire HGT, Member of DIA Advisory Council Europe, Switzerland

Specific Challenges for Orphan Drugs with Paediatric Development
Dr Tsveta Schyns-Liharska, Patient Representative, Member of the PDCO, EMA, Belgium

Panel Discussion:
Dr Jordi Llinares, Head of Orphan Medicines at the European Medicines Agency (EMA), EU

Session 0604 | Friday, 25 May 2012, 11:00-12:30 Room G

COMPASSIONATE USE PROGRAMMES

Session Chairs:
Etelka Czondi, Programmes Coordinator, Sense International, Romania
Arielle North, Ancre Consultant, formerly European Medicines Agency, Belgium

Presentation of Main Outcomes from EURORDIS ERTC Workshop on 21/11/2011
Arielle North, Ancre Consultant, formerly European Medicines Agency, Belgium

Survey Results: Overview of recent compassionate use programmes for OMPs and issues raised
François Houÿez, Director of Health Policy, EURORDIS, France

Session 0605 | Friday, 25 May 2012, 13:30-15:00 Room G

TREATMENT OF RARE CANCERS

Session Chair:
Adam Heathfield, Science Policy Director, Pfizer, UK

Regulator's Perspective
Dr Francesco Pignatti, Head of Oncology, Haematology and Diagnostics, European Medicines Agency, EU

Academic Perspective
Prof. Jean-Yves Blay, President of the European Organisation for Research and Treatment of Cancer, University Claude Bernard Lyon, France

Physician's Perspective
Dr Paolo G. Casali, Medical Oncologist, Head of the Adult Sarcoma Medical Oncology Unit, Istituto Nazionale dei Tumori, Italy

Session 0606 | Friday, 25 May 2012, 15:00-16:30 Room G

ORAL POSTER SESSION

Session Chair:
Dr Jordi Llinares, Head of Orphan Medicines at the European Medicines Agency (EMA), EU

From Rationing to Rationality: An N-Of-One Trial Service for Off-Label Medicines for Rare (Neuromuscular) Diseases (43)
Stephanie Weinreich, EMGO Institute, VUMC, The Netherlands

Consensus "Pathways of Care" as a Mean to an End (55)
Hanka Meutgeert, Volwassenen, Kinderen en Stofwisselingsziekten (VKS), The Netherlands

Listening to Children and Parents Voices: Using Patient Reported Outcomes to Empower Patients with Orphan Diseases and their Parents (64)
Linda Abetz, Mapi Values, UK

Review of Marketing Authorisation Applications of Orphan Medicinal Products (122)
Dr Jordi Llinares, Head of Orphan Medicines at the European Medicines Agency (EMA), EU

Theme 7 | Patients' Empowerment

Lesley Greene, Patient Representative COMP, CLIMB, UK

Torben Grønnebaek†, Past Co-Chair ECRD 2010 Krakow / Member EUCERD (EU Presidency 1st semester 2012) / Rare Disorders Denmark / Board Member EURORDIS, Denmark

Review of how /why patients and families have been empowered and how the Empowerment of Patients and their families has driven positive developments and change in policy and practice for rare diseases in relation to research, therapy and social care issues. The theme will also examine how patient empowerment can continue to be harnessed for the benefit of all stakeholders at political, operational and personal levels in the Rare Disease community.

Session 0701 | Thursday, 24 May 2012, 14:00-15:30
Room C + D

EMPOWERING PATIENTES AND THEIR FAMILIES

Session Chair:
Lesley Greene, Patient Representative COMP, CLIMB, UK

Academia Perspective
Isabelle Aujoulat, Université Catholique de Louvain, Institut de Recherche Santé & Société (IRSS), Belgium

Society Perspective
Caroline Huyard, Author Rare. Sur la cause politique des maladies peu fréquentes, Paris, EHESS, Coll. "En temps et lieu", 2012, France

Patients Perspective
Lise Murphy, Swedish Marfan Association, EURORDIS Volunteer, Sweden

Panellists:
Anna Arellanesova, Czech Cystic Fibrosis Association, Czech Association for Rare Diseases, Czech Republic
Kevin Loth, Vice Chair of Task Force Rds & ODs of EuropaBio-EBE, EUCERD Member, Senior Director, External Relations, Europe, Celgene, UK
Jana Petrenko, Chair Czech Coalition for Health, Czech Republic

Session 0702 | Thursday, 24 May 2012, 16:30-18:00
Room C + D

EMPOWERMENT AT A POLITICAL LEVEL

Session Chair:
Rosa Sanchez De Vega, Spanish Association Aniridia, Past President FEDER, EURORDIS Board Member, Spain

Development in the Political Role of Patients and Patients Organisations Both at EU and National Level
Terkel Andersen, President EURORDIS, Denmark

Building Organizational Capacity in Romania through Failure and Successes
Dorica Dan, President Romanian Prader Willi Association, Romanian National Alliance for Rare Diseases, Romanian Association for Rare Cancers, Board Member and Officer EURORDIS, BoD, IPWSO, Romania

The Involvement of Patients in Developing UK National Plan (RDUK)
Stephen Nutt, Executive Officer Rare Diseases, UK

Panel Discussion: Steering Committee
Dr Pauline Evers, Member of the COMP, Federation of Cancer Patients Organisations, The Netherlands

Session 0703 | Friday, 25 May 2012, 09:00-10:30
Room C+D

ORGANISATIONAL LEVEL: PATIENTS GENERATED KNOWLEDGE IN PRACTICE

Session Chair:
Dorica Dan, Romanian Prader Willi Association, Romania

Impact of Social Media and Patient Knowledge
Denis Costello, Web Communications Senior Manager and Rare Connect Leader, EURORDIS, Spain

Denmark's Documentation Strategy
Lene Jensen, CEO Rare Disorders Denmark (RDD)

Climb & The Bemis Project
Pam Davies, Family Services Manager, Climb, UK

Session 0704 | Friday, 25 May 2012, 11:00-12:30
Room C+D

PROJECTS THAT EMPOWER AND INSPIRE: EXAMPLES OF THE ADDED VALUE OF WORKING, LEARNING AND ACTING TOGETHER

Session Chair:
Dr Gabor Pogany, President Rare Diseases Hungary, Member EUCERD, Hungary

Special International Empowerment Camps
Zsuzsanna Poganyné Bojtor, President of the Hungarian Williams Syndrome, Hungary

Oral Poster Presentation: Hole in the Wall Volunteering Programme to Support Life-Changing Experiences to Children with Serious Medical Conditions (326)
Terence Dignan, Orphan Europe Recordati Group, Italy

Oral Poster Presentation: Rare Family Days "A Family Empowerment Programme" (32)
Birthe Byskov Holm, President, Rare Disorders Denmark (RDD), Member of the COMP, Denmark

Oral Poster Presentation: Developing the Institutional Capacity of Rare Disease Patient Associations: The Promise of Social Entrepreneurship Projects in Business Schools (153)
David Forse, EMLYON Business School, France

Session 0705 | Friday, 25 May 2012, 13:30-15:00
Room C+D

Interpretation FR + RU

PERSONAL LEVEL: THE FORUM OF "PATIENT GROUPS INNOVATION" THE WAY IN WHICH THE PATIENT EXPERIENCE HAS INSPIRED, INFORMED AND DEVELOPED ORIGINAL APPROACHES TO MEDICAL KNOWLEDGE AND DRUG DEVELOPMENT

Session Chair:
Lesley Greene, Patient Representative COMP, CLIMB, UK

Debate with focus on patients: Engaging the Support for Drug Development
Dr Nick Sireau, AKU Society and Nitisinone, UK

Is More Involvement Needed in the Clinical Trial Design & Endpoints?
Elizabeth Vroom, Duchenne Patient Project, the Netherlands

Patient Perspective of Clinical Trial Involvement: Are they listening to my needs?
Ulrike Pypops, Association Muco Vereniging, Belgium

Session 0706 | Friday, 25 May, 16:30-18:00 Room C + D

Interpretation FR + RU

ORAL Poster SESSION

Session Chair:
Josep Torrent i Farnell, General Director, Fundacio Doctor Robert, COMP, Past Co-Chair ECRD 2010 Krakow & ECRD 2007 Lisbon, Member of the Committee of Orphan Medicinal Products, Spain

What Price Do we Pay for Repurposing Medicines for Rare Diseases? (81)
Eline Picavet, Catholic University of Leuven, Belgium

EuroGentest: Harmonization, validation and standardization in genetic testing (103)
Valerie de Groote, Center for Human Genetics, Belgium

Estimating the Budget Impact of Orphan Medicines in Europe: 2010 – 2020 (125)
Carina Schey, Global Market Access Solutions LLP, UK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Summary of Opening Session

Avril Daly, Chair Rare Diseases Ireland, CEO Fighting Blindness, Board Member and Officer EURORDIS


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Rare diseases represent a federating field of action and ECRD has become a truly global event, with people from all over Europe, USA, Canada, Latin America, Asia and Africa. Since 2001, the rare disease movement has gone "from strength to strength": rare diseases have become a public health priority in Europe while EURORDIS celebrated its 15th anniversary. National Plans on Rare Diseases are under development in all EU Member States and, despite the crisis, there have been substantial breakthroughs in research.

"The innovation spirit of the rare disease community must be protected as it is a key element to address unmet medical needs."

Brigitte Franke-Bray, Director, Global Training Officer, DIA Europe, Switzerland

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ECRD is a global event which embodies what DIA does: partnering with experts, providing a platform not only to exchange information about drug development but also generate ideas to make things better. Through the EUPATI project and together with the Innovative Medicines Initiative's Public Private Partnership, EFPIA, patient organisations and others, DIA Europe is developing a comprehensive training material on drug development for patient experts, patient advocates and the public at large.

"There is a need to share information and work together to tackle unmet medical needs."

John Dalli, Commissioner for Health and Consumer Policy, EU

The work of the European Commission is to help bring together scarce knowledge and fragmented resources to maximise synergies and results. In 2008, a Commission Communication on Rare Diseases put forward a co-operation system between the Commission, Member States and various stakeholders. This paved the way for a Council Recommendation in 2009 and a number of joint actions under the EU Health Programme, as well as to the creation in 2010 of the EUCERD. In the Council Recommendation, Member States have committed to adopting National Plans by the end of 2013. The Commission has developed technical assistance to help Member States create these Plans with the EUROPLAN project and actions on rare diseases also feature prominently in the Commission proposal for the new Health Programme.


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Another important aspect or our work concerns access to information for both patients and health professionals, which is indispensable for good quality care. The EU-funded Orphanet database has become the world reference for knowledge on rare diseases.

Patients also need access to highly specialised centres and to the right medicines. The Commission action addresses this by creating a system for establishing European Reference Networks under the Cross Border Health Care Directive, as well as by financing additional networks and rare disease registries. We are also committed to creating specific incentives for pharmaceutical companies to develop and market new medicines for rare diseases.

Since 2008, the Commission has allocated over 300 million euros to research on rare diseases. Rare disease research also serve as models to better understand common conditions. The EC spearheaded the International Rare Disease Research Consortium (IRDiRC) which aims at delivering 200 new therapies and diagnostic tools for most rare diseases by 2020.

"Our common objective is to improve the lives of over 30 million Europeans who suffer from a rare disease."


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Jytte Lyngvig, Director at the Danish Health and Medicines Authority, Denmark, on behalf of the Danish EU Presidency

To face the challenges of rare diseases, the EU Public Health Programme will place great focus on European Reference Networks of Centres of Expertise. This is especially relevant in the field of rare diseases, where there are many diseases with low prevalence, where knowledge is scarce, research difficult, diagnosis delayed. This networking is essential, especially for smaller countries and the Cross Border Health Care Directive will help in this respect. Although actions on rare diseases feature prominently in the Commission proposal for the new Health Programme and there have also been great achievements in the field of Orphan Drug designation, there is the need to follow the placing of these drugs on the market.

"The EU is inspiring other parts of the world. There are the right policies, there is support and there are the opinions developed by the EUCERD: We are on the right track."

Terkel Andersen, President EURORDIS, Denmark

The rare disease field is attracting more and more dedicated people but it is useful to remember that 15 years ago rare disease patients were ignored and isolated. Today, rare diseases are recognised as a Public Health priority with high unmet medical needs but also with high Community added value. We are building a unique, integrated, comprehensive and long term strategy to address patients' needs in Europe. The current priorities are the EU Directive on Cross Border Health Care, the *Health for Growth Programme* in Public Health, and *Horizon 2020* Framework Programme in Research. These programmes must translate into concrete results for patients. In order to achieve this we need a politically supportive environment, with adequate funding at both national and EU levels. In order to turn this vision and robust policy base into reality we need the technical guidance of the EUCERD and the support of structured networks. Next steps include: adoption of the National Plans by all EU MSs by the end of 2013 and the development of the EUCERD's Joint Action on Rare Diseases, in which EURORDIS is involved and which will add the social dimension to the current strategy.


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"Rare diseases are a good model of EU collaboration to inspire optimal use of resources and innovative health solutions."

Summary of Plenary Session


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Ségolène Aymé, Chair EUCERD, France

We have made great achievements in the last 12 years.

We have now 1883 genes with a test. This is a major breakthrough which goes in the right direction for patients. We have now more tests available in Europe than in the US.

The directory of medical laboratories providing testing is useful but there must also be a cross-border flow of diagnostic tests. We need to think global. This is a new essential challenge for Europe. We also need more registries, which are crucial for clinical research, to spur Research & Development in Europe.

We also need to promote research at international level: The EU Framework Programmes for Research have allowed for the creation of large networks and, cooperation between funding agencies has increased within E-Rare. This trend has also been consolidated with the launch of International Rare Disease Research Consortium. We must encourage more countries to join these initiatives.

Today, it is unquestionable that there is the need to create Centres of Expertise. In two years hopefully most countries will have Centres of Expertise designated in their National Plan.

Last but not least we have created better information through Orphanet which currently has 20.000 visitors a day and is rapidly expanding outside of Europe.

"The role of patients can be more than decisive: nothing happens related to orphan drug development if there is not a patient organisation behind it"

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

The impetus received from Europe, and the impact of EURORDIS, have been essential to advance the rare disease movement in Canada. Back in 1996, the position of the Canadian Government was to say: "There is no need for a specific Orphan Drug law in Canada - as the US has one". A great deal of progress has been achieved since, culminating with the recent adoption in Canada of the Undiagnosed Disease Research and Collaboration Act. Through an overview of the situation in Asia (China, Singapore, Japan, Korea, Taiwan, Province of China), as well as in Australia and Argentina, it is clear that there has been a direct impact of the work from Europe. Assistance to patients and caregivers from support by public and private Social Security schemes should be encouraged.

"The main issue is of course real access to treatment when marketed. This is a tremendous challenge to all of us. We must work hard with funders to support access."

Edmund Jessop, Medical Advisor, National Health Service, UK

The Council Recommendations have had an enormous effect throughout the EU. Even though they are all at different stages of development, every Member State is now committed to produce a Plan for people with rare disease by focusing on the most advanced and to learn from their experience.

Every country is now debating its rare disease strategy, focusing on important questions such whether there should be one or more registries; the purpose of screening and type of screening that should be performed or how to train doctors on more than 5000 different diseases. The main question though is whether plans will be implemented.

"The French National Plan has changed in two ways after its evaluation: it has been simplified and the focus is on European Reference Networks rather than on Centres of Expertise. This is a message to the EU as a whole."

Dr Andreas Reimann, Mukoviszidose EV National Action League on Rare Diseases, Germany

Since 2009, there is a roadmap for the adoption of a National Plan in Germany by 2013. The multi-stakeholders negotiations followed a bottom up approach aimed at reaching consensus. A National Action League (NAMSE) has been set up by the National Alliance of Rare Disease patient organisations (ACHSE) and the Federal Ministry for Health and is working on a National Plan that will rest on four main pillars: information, diagnosis, care, and research.

Alan Garcia, Social Affairs Inspectorate General, National Plan for Rare Diseases, EUCERD Member, France

France has laid down the foundations for European Reference Networks, by recognising the specificities of rare diseases and by defining new criteria and simpler evaluation for re-designation of more than 600 Centres of Expertise.

In 2011 France adopted a new National Plan for Rare Diseases with the aim to cover every rare disease and every patient. To achieve this goal, more European and international cooperation is needed. Important objectives are: to share resources, collect clinical data, produce good practice guidelines, improve training, connecting with local regional platforms in order to coordinate healthcare and social care delivery.

Ri de Ridder, Director General at National Institute for Health and Disability Insurance, Belgium

Following the Council Recommendation, measures were taken to create a Belgian Plan. Today, there is a strong push to adopt it by 2013 despite important budget constraints. A multi-stakeholders group, gathering patient organisations, clinical centres, experts, academia, industry, agencies and private health insurers, has been working since 2008. Currently the Plan's budget is estimated at 17 million over 5 years. The first measure will be to set up a National registry compatible with Orphanet and EPIRARE strategy. Belgium is greatly benefiting from what is going on in Europe and from the EUCERD's guidance.

Across the themes

Theme 1: National Plans for Rare Diseases

The latest information about National Plans for Rare Diseases has been integrated into sessions throughout the programme.

Theme 2: Centres of Expertise and European Reference Networks

If rare diseases are rare, expertise on them is also scarce and fragmented within Member States and across the EU. The goal of Theme 2 was to describe and discuss the state of art as well as the future needs related to Centres of Expertise and European Reference Networks (ERNs) from the viewpoints of different actors including politicians, health care professionals, patient organisations and individual patients. Theme 2 also introduced the present work and future plans of the EUCERD Committee.


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In the first Session, Ségolène Ayme, Chair EUCERD, France, presented the state of art of Centres of Expertise in different EU countries. At present, a national plan for rare diseases has been adopted in 5 countries but fully implemented only in France which has already finalised a second version of its national plan. In the other countries, centres dedicated to a rare disease or group of diseases exist but are not yet designated officially, except in the UK.

Edmond Jessop, Medical Advisor, National Health Service, UK, presented "the EUCERD recommendations on the criteria for establishing national Centres of Expertise". A panel discussion presented three examples of problems and solutions identified at national level: France as an example of a good process to designate and fund centres of expertise, Finland taking the first steps and trying to fit the demands with its small population in a large country, and Germany in between these two extremes. As stated by Véronique Héon-Klin, Federal Ministry of Health, Germany, the process of developing and discussing national plans is itself already important and promotes the issues related to rare diseases.

European Reference Networks (ERNs) was the topic of the second session. The criteria for selecting the ERNs are still under discussion in the EUCERD Committee and the final conclusions are not expected before late autumn this year. Enrique Térol, Healthcare Systems Unit, DG SANCO, European Commission, Belgium explained that one of the aims of the Directive on Cross-Border Health Care is to provide assurance about safety and quality of cross-border health care and, in this, ERNs will have an important role as officially recognised for their expertise and quality. Kate Busby, Honorary Consultant Geneticist, Newcastle upon Tyne Hospitals NHS Foundation Trust, Joint coordinator, TREAT-NMD network, Vice President, EUCERD, UK, reminded us that a main aim of the EUCERD Joint Action is to find an optimal model for ERNs. Antoni Montserrat, Policy Officer for Rare and Neurodevelopmental Diseases, DG SANCO, European Commission, Belgium proposed that some of the past rare disease networking projects financed by DG SANCO could be the first pilot ERNs.

The EU Directive aiming at facilitating access to safe and high-quality cross-border health care and promoting cooperation on healthcare between Member States was adopted early 2011. The new directive was written to agree upon and clarify the rights of patients who seek healthcare in another Member State supplementing the rights that patients already had at EU level through the legislation on the coordination of social security schemes (regulation 883/04). Many still consider that there are a lot of questions that need further clarification before the Directive can really be considered as implemented. Theme 2 devoted two sessions to these issues.

Session 3 presented the questions related to sample mobility. In diagnostic (genetic) testing of rare diseases it happens hundreds of times every day in Europe that a sample has to be sent for diagnostic work up to a laboratory in another country. The three speakers in this session, David Barton, National Centre for Medical Genetics, Ireland, Jean-Jacques Cassiman, Center Human Genetics, Forensic Medicine, Leuven, Belgium and Els Dequeker, EuroGenTest, Quality Manager Division Diagnostic UZ Leuven Biomedical Quality Assurance Research Unit, Belgium, presented their wide experience on the issues and solutions related to sample mobility. There is, on one hand, detailed regulation (Directive on In vitro Diagnostics) which at present is under much needed revision, which will have a significant effect on how genetic tests will be regulated in the future. On the other hand, there are important practical issues related to the quality of health care services in the country sending the samples as

well as to the quality of the diagnostic laboratory in the country where the samples are received. The EU funded Eurogentest project and its continuation Eurogentest2 have concentrated on solving these quality issues by discussing them in several workshops, creating guidelines, establishing together with Orphanet a database presenting the accreditation status of the diagnostic laboratories and organising training courses.

Session 5 discussed cross-border health care from the point of view of patient mobility. Smaller Member States cannot provide high quality health care for each rare problem. The idea of the ERNs, according to Nathalie Chaze, Head of Unit, Healthcare Systems, DG SANCO, European

Commission, Belgium is partly to offer a reliable centre for patients seeking the best experience but especially to offer knowledge and guidelines so that specialists will better be able to treat their patients in the country where they live. Marianne Jespersen, Member of the Rare Diseases Task Force, Member of the Working Group on Standards of Care, National Board of Health, Denmark explained that referring patients abroad has long been practiced even before the Cross-Border Directive in Denmark, which has created a flexible system for sending patients for specific, very rare treatments to neighbouring countries, especially to Germany. She stressed that for rare disease patients and especially related to cross-border health care, a coordinating health care person is imperative.

Rare diseases exist in all fields of medicine and they can present at any time of life. A considerable part of them, however, present in childhood. Thus paediatricians have traditionally had most experience in diagnosing and treating those diseases. When the child reaches adulthood, he or she often loses the experienced medical team that has been following him/her for many years and may feel deserted. Moreover, improving treatments have changed the prognosis of some of the diseases and enabled adult life in some conditions that used to be lethal in childhood. For these reasons the important topic of health care pathways focusing on transition from childhood to adulthood was chosen for Session 4. Hanns Lochmüller, Chair of Experimental Myology, Institute of Genetic Medicine, Newcastle University, UK explored the issue with Duchenne Muscular Dystrophy as an example: healthcare and families have to change their attitude to raising a child to adult life which was not the case before. Liesbeth Siderius, Representative of Shwachman diamond Syndrome Support Holland, European Academy of Paediatrics, The Netherlands working as a paediatrician in primary health care, stressed the important role of local doctors in transition from childhood to adulthood. A presentation on family planning in rare diseases by Helena Kääriäinen, Research Professor, National Institute of Health and Welfare, Helsinki, Finland introduced the sensitive topic of the wishes of the families to avoid further affected children.


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Overall, Theme 2 focused on the burning questions that touch everybody involved in the treatment of rare diseases. When the issues suggested in the Council Recommendation on European Action in the field of Rare Diseases will be part of national health care in the Member States in coming years, the quality of health care for patients with rare diseases can be expected to improve while the costs may remain the same or even decrease due to more effective ways of utilising the existing expertise.

Theme3: Information and Public Health

Making rare diseases visible in society is a challenge that we now have the means to achieve. The general public can be involved with appropriate initiatives that have already proven their effectiveness. Researcher's interest is enhanced by the availability of organised data freely accessible on the web. Healthcare professionals have sources of information on good practice guidelines, and information technologies allow for innovative training approaches. This will foster early diagnosis and early prevention.

Making rare diseases visible in society: powerful tools for broad dissemination


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Ms Avril Daly, Co-Chair ECRD 2012 Brussels, Vice-Chair EURORDIS' Board of Directors, Chair Rare Diseases Ireland (GRDO), CEO Fighting Blindness, Ireland, opened the session by presenting the achievements of the 2012 Rare Disease Day awareness campaign. The focus was on what worked and how the campaign has been successful over the past 5 years at making rare diseases more visible to the general public, as well as complementing national advocacy actions targeting public authorities and policy-makers. Avril highlighted Community Level and National Level Initiatives, using the Irish Rare Disease Alliance (GRDO) video and postcard campaign, as an example.

Ms Monique Karkatcharian, Communications & HR Director, AFM Généthon France presented the strategy of the French Téléthon to collect donations while heightening the public's awareness of rare diseases. This strategy that rests on three pillars: 30-hour live TV broadcast marathon, development of a volunteer network and a donation appeal using the Internet and social media. The French telethon is an example of brand building associated with key values: solidarity, transparency and innovation.

Mr Denis Costello, Web Communications Senior Manager and Rare Connect Leader at EURORDIS demonstrated information resources available online – the so-called Web 2.0 revolution. He explained how to evaluate all these forms of on-line information and networking, what to look out for and how patients and families can shape this new and growing environment. More than 50% of Internet users in the EU are seeking health information and almost all patients in Europe have greater access to information about their symptoms, as well as healthcare options. At the same time some rare disease patient groups have started transforming their disease experience into novel research data. Patient organisations are in a pivotal position to promote the conversation with healthcare professionals, build a community and play a quality assurance role.

He illustrated his arguments with three case studies: classical social media (i.e. Facebook); online patient communities tailored to rare diseases (i.e. RareConnect) and online patient reported outcomes and registries (i.e. Duchenne Connect or Patients Like Me). There is a growing importance attached to patient reported data and therefore growing opportunities for patient groups. However, patients should be aware of the challenges posed by data ownership and privacy issues and the emergency of for-profit companies that can spread misinformation and take advantage of vulnerable people.

Availability of data and the challenge of making data freely accessible

The session chaired by Dr Ségolène Aymé, Chair EUCERD, France, gave an overview of the classification and coding of rare diseases: where we stand, rationale, why it matters and what it can change. Dr Peter Robinson from the Institute of Medical and Human Genetics, Charité Berlin, Germany, focused on the importance and complexity of including rare diseases in the International Classification of Diseases (ICD) managed by the World Health Organisation (WHO). He came to the alarming conclusion that with only 500 rare diseases listed in the current classification system (ICD-10) and only 3% of all rare diseases having a specific code, most rare diseases have been and still are invisible to policy-makers.

He went on to explain the work being undertaken by Dr Ségolène Aymé and her team with the WHO to add a comprehensive nosology of most currently known rare diseases in the next version of the international classification of diseases (ICD-11). The classification is particularly important for rare disease to be represented in hospital IT systems, to have more accurate statistics on the prevalence of these diseases, to improve the ability to find data on databases (such as Orphanet), to perform health electronic record mining for rare disease data and to better allocate health care resources in general.

Improving Care through Clinical Guidelines

Dr Odile Kremp, Director ORPHANET-INSERM US14, France, explained the challenges related to establishing guidelines for rare diseases and their importance. She described the methodology used to produce Orphanet emergency guidelines and Professional Clinical guidelines, using the French experience with national protocols for diagnostics and care (PNDS). In France, PNDS constitute a best practice reference document for use by health professionals, the patient and their families. There have been only 45 PNDS validated and published since 2005. The second French National Plan for Rare Diseases aims at simplifying the methodology in order to accelerate the production of diagnosis and treatment guidelines.

Dr Janbernd Kirschner, Consultant Paediatric Neurologist, Uniklinik Freiburg, Germany described how reference

networks develop, implement and monitor guidelines taking the reference network for Duchenne Muscular Dystrophy network (TREAT NMD , CARE NMD) as an example. Care guidelines for Duchenne are the result of a consensus process between patient organisations, translational research networks and health agencies. An international coalition of 84 experts was consulted and a literature review was conducted to produce care recommendations that were published in a scientific journal and then turned into a Family Guide.

Ms Kay Parkinson, CEO, Alström Syndrome, UK explained the involvement of patients in developing clinical guidelines. Since she founded Alström Syndrome UK in 1998 they have developed a website, patient information and a medical handbook. They have organised and developed multi-disciplinary clinics in partnership with Torbay Hospital and Birmingham's Children Hospital and have instigated and obtained NHS funding to run them. The work performed by Alström Syndrome UK has been hailed as a model to improve clinical guidelines for other multi-organ rare diseases.

New approaches for training and awareness

The session started with an example of highly-specialised training of medical doctors using the Internet. Mr Thomas Parent, Chief Technical Officer, WeBSurg, France described WeBSurg.com, a web-based surgical university dedicated to MIS surgery.

Dr Paula Byrne, Senior Lecturer at the School of Medicine of the University College Dublin, Ireland, described the training available to medical students on rare diseases at her institution. The training consists of a novel elective module that runs on a 4-month period. The module is designed to enhance awareness of rare genetic disorders amongst future medical professionals. The learning outcomes are: to understand what is meant by a "rare disease", to reflect on the obstacles and difficulties, to appreciate the importance of research, to recognise the importance of identifying novel treatments. In addition the students taking this course are informed of the government policies in this field, they are taught to identify the information resources available to them and to recognise that "it is not rare to have a rare disease". Amongst the guest lecturers invited to contribute to the course, are representatives of patient organisations, clinicians, scientists and the pharmaceutical industry. In order to assess the impact of the training, students are asked to produce a pamphlet for medical healthcare professionals on one particular rare disease and keep a learning journal to record their increasing awareness of these disorders.

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Ms Simona Bellagambi, Rare Diseases Italy (UNIAMO), presented two initiatives to improve information and awareness about rare diseases in Italy. The first is a project aimed at raising the diagnostic suspicion and developing procedures to handover the daily care from paediatric age to adult age. "Conoscere per assistere" (Knowledge for Helping) started in 2009 and is funded by the pharmaceutical industry trade union in Italy. It is the result of collaboration between the rare disease patient umbrella organisation (UNIAMO) and several federations gathering together various medical specialities. The main goal of the project is to train trainers who will be able to transfer these messages at the regional and provincial level through the organisation of local educational courses as part of health professional's continuing education. To date, trainees are recruited by the medical societies collaborating with the project. Between November 2010 and January 2012, 10 courses were held in 10 regions involving 514 participants. Another six courses are planned to be held in other regions in 2012-13.

Another initiative in Italy has been promoted by UNIAMO to make information on rare diseases more accessible. The website www.malatirari.it is a simple and comprehensible database to meet the daily needs of patients and their families and in a second phase will be expanded to include the information needs of doctors. Health professionals are meant to be users and authors/ guarantors of the scientific validity of the scientific information.

Fostering early diagnosis and prevention

Dr Peter Burgard, Centre for Paediatric and Adolescent Medicine, Department of General Paediatrics, University of Heidelberg, Germany described the State of the Art of screening in Europe and presented the recommendations of the European Network of Experts on Newborn Screening. He went over some basic concepts already been used to decide on screening programmes. In particular the Wilson Jungner's Principles and Practice of screening for disease

and the concept of "Number Needed to Screen" to evaluate a screening programme. He went on to explain the work undertaken by the European Commission Public Health Programme to evaluate all the new-born screening practices for rare disorders in Member States. This initiative has already produced a report on current practices and regulations by country and an Expert Opinion document with 70 recommendations to decide on the implementation of new-born screening programme.

Dr Ilona Autti-Rämö, Chief of Health Research, Research Professor, The Social Insurance Institution Research Department, Helsinki, Finland presented the Finnish case. Since 2002 the Finnish government has been running a Health Technology Assessment on neonatal screening for rare metabolic disorders faced misconceptions and blurred objectivity. The assessment is supposed to consider the specific needs for Finland in terms of resources, prevalence of disorders and organisation. As part of this initiative, a screening model was constructed using the best available evidence from literature and clinical experts. Special emphasis was given to ethical and equity issues.

The results of the pilot are that 5-10 children with rare metabolic diseases could be identified yearly. 1 to 3 deaths and 1 to 3 severe handicaps could be avoided per year. The estimated cost is 2.5 million euros/year, that is to say 45 euros per new-born. In conclusion, three scenarios were proposed to the National Screening Committee to decide on the follow-up: 1) No change to the current situation (screening for hypothyreosis from naval cord blood); 2) screening for CAH only, 3) screening for all five disorders.

Primary and Secondary Prevention

Prof. Helena Kaariainen, Vice Chair EUCERD, Research Professor at National Institute of Health and Welfare, Helsinki, Finland presented an overview of genetic counselling for family planning and the considerations surrounding this issue. She attempted to answer the following questions: Who should benefit from this service and when (before or during the pregnancy)? She drew examples of several rare genetic conditions. The conclusion is that genetic counselling should be available for families with an increased risk of a rare disease in the offspring. It should be carried out by appropriately trained professionals, and should provide information and support. The decisions should be based on the information and the values, wishes and cultural norms of the couple.

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Mr Pierre Mertens, President, International Federation for Spina Bifida and Hydrocephalus, Belgium presented Spina Bifida as an example of a rare disease that can greatly benefit from information and awareness of primary prevention. IF promotes folic acid as an effective way of preventing the disease before birth. They advocate for mandatory fortification of staple foods with folic acid and has started a European campaign to raise awareness about primary prevention with folic acid. They have presented a report on Prevention to Neural Tube Defects in Europe at a Public Hearing in 2010 and have recently come out with a second report on this topic.

Ms Samantha Parker, Director of External Affairs and Rare Disease Partnerships, Orphan Europe, France presented the European Porphyria Network (EPNET). EPNET is a pilot reference network which was set up to mitigate the disparity in the content and accessibility of information for this rare genetic disease. To date EPNET has managed to connect 31 centres in 21 countries and has as an objective to have a registered specialist centre in every EU country and to improve diagnostic and analytical quality of existing centres in order to reduce delays in diagnosis. EPNET has already produced a consensus-agreed information leaflet for all types of porphyria in all European languages for patients and health professionals.

Theme 4: Research from Bench to Bedside

The opening session of Theme 4: Research from Bench to Bedside illustrated how the complexity and the challenges that need to be tackled in order to understand the physio-pathological mechanisms underlying rare diseases represent a driving force for new scientific paradigms, evolution of regulatory frameworks and technological innovation.

Hannah Rosenbaum, Head of Hematology Day Care Unit, Rambam Health Care Campus Haifa, Israel provided a striking example of how the molecular mechanisms at the origin of a rare metabolic disease, Gaucher disease (GD), advanced understanding of the commonest neurodegenerative movement disorder, Parkinson disease (PD). One of the first clues that there might be a link between PD and GD arose from a small number of case reports describing parkinsonian features in GD patients.

This was followed by the finding that PD occurs at an increased frequency in the relatives of GD patients who are carriers of *GBA1* mutations. These findings have led to extensive subsequent studies that have opened new exciting research venues. New and improved mouse models of GD and PD will undoubtedly aid this process, leading to the development of new therapies that successfully cross the blood-brain barrier and ameliorate the central nervous system disease in both conditions.

Anne Pariser, Associate Director for Rare Diseases, Office of New Drugs CDER, FDA, USA provided the perspective of the US Food and Drug Administration (FDA) on rare diseases, orphan drugs and innovation. Her presentation included a review of rare disease and orphan drugs history showing how rare diseases have often led the way for medical advances. An early example comes from the study of familial hypercholesterolemia (FH) described in 1938 by Carl Müller and that led in 1987 to the market approval of lovastatin, the first lipid-lowering therapy (HMG-CoA reductase inhibitor) in US. Presently, 60 million Americans are receiving lipid-lowering therapies, the all-time highest grossing prescription drugs in US. The early wave of orphan drugs (ODs) brought the development of therapies for rare hematopoietic neoplastic diseases. Some of these ODs have changed the standards of medical practice e.g. Imatinib for Chronic Myelogenous Leukemia or Rituximab for Chronic Lymphocytic Leukemia and Non-Hodgkins Lymphoma. According to the Center for Drug Evaluation and Research (CDER, part of the FDA), the recent history of ODs is blossoming with new approvals (46 in the last 2 years), 50% of each is represented by biologics. Forecast for the future of ODs developments is indeed very promising and it is more and more apparent that ODs have changed the model of drug development from a linear concept to a "foundation-building"-parallel concept, where the traditional phases of drug development run parallel or superimposed instead of following one another. In summary, orphan drug development and approvals have produced a major impact on the treatment of both rare and common diseases; the advent of targeted therapy in orphan diseases is associated with the application of regulatory flexibility and scientific judgment; as new targeted therapeutics continue to evolve, approvals for orphan drugs will likely continue to grow in importance.

Serge Braun, Scientific Director, AFM, France traced the very successful 25 years long history of how the Association Francaise contre les myopathies (AFM) has succeeded in encouraging and promoting research on rare disease and innovation. The AFM is composed of patients and their families who are affected by neuromuscular diseases. There are about 200 neuromuscular diseases (NMD), most of them have a genetic origin and are rare. In order to fight these diseases, AFM chose to initiate innovative actions and took a strategy of raising general interest on all rare diseases and persons with disabilities. In order to find a cure, AFM supports the development of innovative therapies for neuromuscular diseases and for rare diseases altogether. AFM has developed an integrative approach to accelerate and overcome the challenges that its mission poses by financing a) basic research projects and fellowships, b) strategic programmes, including 35 on-going clinical trials in both NMD and other genetic disorders, c) research infrastructures such as Genethon, Institut de Myologie, I-Stem, GIS Maladies Rares, Biobanks and patient registries.

Clinical trials supported by the AFM naturally mainly concern NMD but also other rare diseases affecting the immune system, blood, eyes, skin, nervous system. The therapeutic pathways explored include gene therapy and cell therapy or approaches combining both. Recently, great emphasis has been given to the renewed interest in gene therapy approaches with the creation of the Genethon Bioprod, the largest worldwide GMP production plant for gene-therapy products to support clinical trials (currently opening). Genethon Bioprod will offer unprecedented production capacity for clinical-grade gene therapy products. The mission of Genethon Bioprod will be to manufacture treatment candidates for clinical trials being conducted in France or at international level, while at the same time ensuring the strictest respect for pharmaceutical regulations.


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During the second session, Iiro Eerola, EC Project Officer, European Commission, Belgium presented the current status of the International Rare Diseases Research Consortium: IRDiRC. This "transatlantic" research consortium, launched in April 2011, has had a very successful start with 25 executive members comprising

major funding bodies and research organisations. The overall aim is to reach 3000 diagnostics and at least 200 new therapies by 2020. The rapid introduction of next generation sequencing into diagnostics is providing ground breaking results in the area of rare diseases research and diagnostics. Dr Eerola highlighted the impact of the FP7 Health Programme and in particular the model role of rare diseases research for future calls of the Horizon 2020 Programme in the area of common diseases.

Paul Lasko, Department of Biology, McGill University, Canada demonstrated the integrated Canadian CIHR 33 institute virtual network of researchers and funding partner activities in the field of rare diseases. Their scope is impressive, comprehensive and involves nation-wide integration of complementary resources. As such, important lessons could be drawn for European Commission-funded research, with the Canada Genomics Enterprise providing a model example of successful collaboration in the area of sequencing of selected rare diseases. Moreover, Dr Lasko elaborated on dedicated rare diseases projects - Atlantic medical genetic and genomics initiative (AMGGI); The Canadian Pediatric Cancer Genome Consortium: Translating next-generation sequencing technologies into improved therapies for high-risk childhood cancer; Orphan Diseases: Identifying genes and novel therapeutics to enhance treatment (IGNITE), Finding of Rare Disease Genes in Canada (FORGE Canada). In particular, the last consortium is currently making seminal discoveries in the identification of genes associated with rare diseases and sets the world-wide stage in this domain. The need for international databases, avoidance of overlap of research activities, need for integrated bio banking and sharing of resources framed the main message coming out of Dr Lasko´s presentation.

To conclude this session, Nicolas Lévy, University Professor, Head of research team on laminopathies and neuromuscular disorders INSERM, Head of Laboratory of Molecular Genetics of the Department of Medical Genetics at the Children's Hospital La Timone, France presented an outline of the French Rare Disease Foundation that aims to gather expertise and funding for the implementation of coordinated research activities in France. This foundation has ambitious aims and multiple corporate partners, aimed in particular at the development of new therapies.

In summary, this session highlighted the need for transnational collaboration in the field of rare diseases research, as exemplified by successes and challenges of the E-rare programme presented in the final overview by Sophie Koutouzov, Secretary General, e-Rare Coordinator, GIS Institut des Maladies Rares, France.

The third session focused on the increasing and urgent need for both the expanded use of rare disease patient registries and for a more systematic and standardized approach to their creation, maintenance, and accessibility on an international and global basis.

Hanns Lochmuller, Chair Experimental Myology, Institute of Genetic Medicine, Newcastle University, UK presented the success story of the TREAT-NMD network. TREAT-NMD is a "network of excellence" funded by the European Union (but with global collaborations) with the aims to help promising new treatments for neuromuscular diseases by facilitating the transition from basic research to treatments for patients. It is an infrastructure project that creates the "tools" for trial-readiness in the neuromuscular field, helping researchers and expert centres to collaborate better and improve patient care worldwide.

Since 2011, it has transitioned into the TREAT-NMD Alliance. Among many other activities, TREAT-NMD had created a global database of patients with the genetic and clinical data necessary for trial recruitment. This initiative has brought many benefits to registered patients such as feedback on standards of care and new research developments, feeling a sense of "belonging" to a broader community, not being left behind as clinical trials develop, and last but not least a link to the research community. The benefits extend to industry as well with easy and transparent access to the patient community, clear concept of target market, feasibility and planning of clinical trials, and recruitment of patients into clinical trials.

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The TREAT-NMD global registry activities are regulated by a strong and transparent governance structure that includes a strong participation of patients. This global registry model can be used as a compelling example to underlie the weakness of the many registries focused on a treatment for a rare disease instead of the disease itself, as is the case for Fabry disease. Indeed, multiple product registries for the same disease fragment

the care pathway for patients, the data are not shared on critical issues (only few stakeholders are involved), knowledge on the appropriate use of the drugs is delayed, and the same patient population is being 'targeted' each time. The development of multi-purpose disease specific registries would ensure a multi-stakeholder involvement, a shared cost model, it would allow comparison of appropriate use of all novel therapeutics, would emphasise patients not just drugs and would provide a way of monitoring the natural history of the disease. Indeed, regulators themselves are advocating for the development of disease focused registries vs. multiple product specific registries.

Domenica Taruscio, Director, National Centre for Rare Diseases, European Platform for Rare Disease Registries (EPIRARE), Italy presented EPIRARE, a European project funded by DG SANCO (April 2011-October 2013). Its main objective is to build consensus and synergies to address regulatory, ethical and technical issues associated with the registration of rare disease patients, to elaborate possible policy scenarios and lay the foundations for a common European rare disease (RD) registry platform. The key problems affecting the utility of registries for future research are: a) lack of harmonisation, with high variability among RD databases, geographical coverage and type of data collected; b) the majority of databases are national or regional, only a minority are European or international; c) lack of data sharing: about half of the registries share some data with other databases but only a minority share data with biobanks or centres of expertise; d) lack of sustainability: a significant proportion of RD registries expire due to lack of commitment from data providers, lack of funding or study termination, and so discontinuation often leads to loss of data and loss of investment; e) lack of utility for research: owes to absence of quality control, standardised data elements, and genetic data.

Epirare has studied the landscape of RD registries in detail. The preliminary data analysis confirms, as expected, that the reality of the RD registries currently operating on the European territory is quite complex and has revealed the weaknesses and contradictions that need to be tackled with concerted efforts from all stakeholders. Full data analysis, soon to be completed, will constitute the base for laying the foundations of a common European rare disease registries platform.

In the fourth session, perspectives on the use of the newest genomic technologies for the diagnosis and research on rare diseases were presented by three practitioners in the field. Over the last few years, fantastic breakthroughs have occurred in the technology and tools to sequence DNA. It is possible today to sequence an individual's genome in days to weeks. These technologies rely on what is called 'massive parallel sequencing' and they are referred to as 'next generation sequencing' (NGS).

David Barton is the Director of the National Centre for Medical Genetics in Dublin, Ireland. As a laboratory geneticist, he is responsible for providing DNA testing for genetic diseases and introducing novel technologies into the diagnostic laboratory as they arise. Of course, David Barton's laboratory is also preparing for NGS, but during the session he had task to (re)introduce the audience to laboratory testing in human genetics as it is performed today. For instance, a clinical diagnosis of cystic fibrosis (CF) can relatively easily be confirmed by mutation analysis using (commercial) kits that test for a limited number of mutations. This is due to the frequency distributions of mutations in the CF gene that cause the disease: in most patients, it suffices to test for the more common CF mutations. The same tests are useful for carrier testing in families and couples. Duchenne Muscular Dystrophy (DMD) is often caused by a deletion in the DMD gene. Hence, a simple technology that specifically detects deletions is the prime choice for the confirmation of the diagnosis in patients, and for carrier testing in their mothers (DMD is an X-linked disease). Thus, the molecular diagnosis of these diseases and of a plethora of other rare, genetic diseases, is relatively straightforward, the tests are widely available today and in the immediate future are unlikely to become obsolete.


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EuroGentest was created several years ago, to help laboratories in validating their assays and in working towards accreditation. New technologies need to be subject to the same rigorous evaluation before they can replace existing techniques. But the future looks bright for diagnostics in previously poorly characterised diseases, as shown by Joris Veltman, a 'next generation' scientist at the Department of Human Genetics at the Radboud University Nijmegen Medical Centre, The Netherlands. Using the latest NGS platforms, his group has been able to identify mutations in novel genes in a number of syndromes, for

which the genetic defect had remained elusive up to date. Using specific, molecular 'capture' tools, the DNA corresponding to the approx. 25.000 genes in a human's genome is isolated. The sequence of that DNA is then determined using fast sequencing machines that 'read' millions of DNA fragments in hours or days. These fragments are then compiled again to result is an 'exome' i.e. information on all the exons of the genes in an individual's genome. Needless to say it requires novel and powerful bioinformatics tools to 'browse' through the raw data and built a correct data set. The technology has been optimized to identify de novo defects (mutations that occur first in the patient, and are thus not present in the parents). The group in Nijmegen has shown that such mutations play a prominent part in rare and common forms of neurodevelopmental diseases, including intellectual disability, autism and schizophrenia. After a thorough validation of the technology, the group is now offering exome sequencing for diagnostics.

Koen Devriendt is a clinical geneticist at the Center for Human Genetics, University of Leuven, Belgium. Using the example of the Long QT (LQT) syndromes, he showed how NGS and exome sequencing will improve diagnostics for heterogeneous diseases. The search for the causal mutation is a patient with a clinically detected LQT can be very lengthy using the current (old) technology, which is largely based on Sanger sequencing of the many individual genes implicated in this condition. It is not only cumbersome, eventually the cost becomes prohibitive and for many of the 'rare' genes, the tests are simply not diagnostically available. This changes when all the genes of a patient can be sequenced in parallel. The information on the entire set of genes, known to be associated with LQT, can now be interrogated using bioinformatics tools. Still, it is not trivial to pinpoint the culprit mutation among thousands of genetic variants. Indeed, our genomes (and exomes) are very variable and it is often quite a challenge to distinguish common variants or polymorphisms for pathogenic mutations. The tools will require constant optimization and fine-tuning. Also, the sequence can be stored and re-analysed later as new evidence for other genes with a role in LQT is published in literature. Thus, NGS is very promising for testing heterogeneous diseases and for rare syndromes. However, given the fact that all genes are sequenced at once, one is faced with an overload of genetic information. Indeed, unsought findings may also be revealed. They are called 'incidental findings'. It will be important for parents and patients to discuss about this possibility with the clinical geneticist beforehand, and to develop the appropriate informed consent. The situation will become increasingly complex when companies will start to offer total genome analysis directly to consumers. At first glance, they may be offers for lifestyle advice only, but intrinsically, they are medical tests. Therefore, the medical professionals, the patients and the community as a whole has to think about how those novel technologies will best be introduced in clinical practice.

Session five presented success stories in translational research in rare disease therapy development and concentrated on two diseases, cystic fibrosis and Duchenne muscular dystrophy where there have been significant advances towards therapy development in the past few years. The support that these efforts have had from the patient organisations, and the infrastructures set in place to facilitate translational research including trial networks and patient registries provide interesting models for the push to therapy development for other rare diseases. Such resources can address the challenges of translational research in rare diseases such as the identification of sufficient patients to perform adequate trials, the identification, standardization and harmonization of clinically relevant outcome measures and access to appropriate tissues for preclinical studies.

Margarida Amaral, Faculty of Sciences, University of Lisbon, Portugal provided an overview of the efforts to define therapies for cystic fibrosis based on an enhanced understanding of the pathogenesis of the disease. Cystic fibrosis affects 1 in 2500 to 1 in 6000 new-borns and is a multisystem disorder where good basic therapy has very much improved quality of life and life expectancy over the last twenty years. New advances in the understanding of the cellular defect in cystic fibrosis have led to multiple new approaches to therapy going beyond symptom control to targeting the underlying disease mechanism, some of which are now becoming available as treatments. The principles of defining clear molecular targets resulting from specific mutations can apply across other rare genetic diseases. Nearly 2000 mutations in the CFTR gene have been identified as causing cystic fibrosis. They can be classified into different groups depending on the way they affect protein structure or function, subsequently provoking the cascade of cellular events that leads to the symptoms of cystic fibrosis. The cystic fibrosis drug pipeline now includes multiple compounds targeting the specific defects induced by particular mutations.

Just as in the field of cystic fibrosis, highly "personalised medicines" look like the way forward. A similar process is happening in Duchenne muscular dystrophy (DMD). Annemieke Aartsma-Rus, DMD Genetic Therapy Group, Department of Human Genetics Leiden University Medical Center, The Netherlands outlined the progress achieved in a particular kind of therapy for DMD (which affects around 1 in 3500 male live births). Like with cystic fibrosis, better baseline medical care has improved life expectancy in this condition, and a range of therapeutic options are under development based on the knowledge of the underlying gene and protein defect.

Antisense oligonucleotides are being trialed in DMD in an approach known as "exon skipping" which aims to change the effect of harmful mutations (particularly deletions), which do not allow the dystrophin protein to be made, to less harmful mutations which allow for partial production of protein. This mimics the situation seen in patients where people with Becker muscular dystrophy have mutations in the gene which allow some production of protein and have a much milder disease than DMD. The process by which the exon skipping approach has moved from basic studies in cells and animal studies and into human trials has to date given promising signals that this may modify disease progression. The therapy would be dependent on the mutation present in individual patients, so a large number of different molecules would need to be developed to benefit the majority of patients.

These examples of drug development for two rare diseases showed how concepts for therapy, based on an understanding of the molecular basis and how mutations cause disease, can progress from laboratory experiments to drug company sponsored placebo controlled trials, and hopefully in the future to more registrations of approved drugs. They also illustrate the manner in which rare diseases can lead the field in personalised medicine approaches, particularly when targeting specific disease causing mutations. These are important paradigms for the rare disease field.

Theme 5: Orphan Products & Rare Disease Therapies: Access

Authorised safe and effective Orphan Medicinal Products to treat rare diseases are – where available – a key element in our approach to diagnosing, treating or even preventing rare diseases. But as more such treatments come to market, and against a backdrop of and in times of economic uncertainty, costs of treating often find themselves under the spotlight. Where do we go from here in ensuring that the interest in orphan drugs continues in a way that is sustainable for all stakeholders? Initiatives at a country-level, European level and even global level are pointing out the potential pathways for the orphan drug and rare disease community. Are there lessons we can learn by cooperating across borders or even across regions? And are there lessons the healthcare systems can learn for approaching other, innovative treatment solutions in an increasingly targeted healthcare environment?

The Theme opened with a review of the current situation, experience to date and future perspectives, taken from the key stakeholders involved in building, refining and maintaining the orphan drug policy in Europe. Paola Testori Coggi, Director General of the Directorate-General for Health and Consumers of the European Commission, echoed Commissioner Dalli's comments in the Opening Session about rare diseases being an area where European cooperation is a policy area of unique high European community added value, vital to patients, by bringing together scarce knowledge and resources.

Mrs Testori Coggi highlighted that, although prices are a key concern, this is not an area where the European Commission can regulate. This is and would remain a competence addressed at Member State level, where the drugs are paid for by the governments that are responsible for their individual healthcare budgets and systems. So, the only option is to increase collaboration between Member States with European Commission support.

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During the conversations, it was also acknowledged that, if we are to stand any chance of reaching the target of 200 new orphan drugs by 2020 as set by the International Rare Diseases Research Consortium (IRDiRC), we need to look at how we can use the systems better. Simply doing the same as today is not going to provide us these results. One

example of using the system better to help Member States make informed decisions could be to include information about the specific orphan drug within the recommendation for Marketing Authorisation. Adaptive authorisation procedures – also called Progressive Licencing - should be explored and we should make better use of the tools that we already have available, such as conditional Marketing Authorisation – something that exists but is very seldom used, although it would appear to be ideally suited to orphan drugs. The key is to use the existing legislative tools well and potential of the scientific agencies better while being creative in finding elements that can further support the development and availability of treatments for rare disease patients.

The European Medicines Agency (EMA), by the voice of its Executive Director, Guido Rasi, underlined its commitment to making orphan drugs available: "patients' access is the ultimate goal"; and highlighted one concrete example of making better use of the system – data that EMA has in hand could be useful to the other parts of the access chain, such as HTA Agencies. Even if certain data is not useful in terms of the Marketing Authorisation process, it could be helpful for someone else. The Agency highlighted its continued contribution to the development of the Clinical Added Value of Orphan Medicinal Products Information Flow (CAVOD) process in development and stressed that they would "continue to contribute in any way that they can" to building better understanding of orphan drugs. One key element of this is to continue the process of early and on-going dialogue between all stakeholders all along the development and availability processes.

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The 1000 orphan drug designations in Europe are a good indication of the number of attempts to develop orphan drugs, but we are still lagging behind the US in the number of designations per year. And, further, the fate of those designated drugs remains an open question – it is now essential to understand the barriers to turning those designations into actual, available orphan drugs. Collaboration along the development chain, as well as collaboration internationally are going to continue to be a key element – something that was echoed by the speakers from the Food & Drug Administration (FDA) and the US National Organisation for Rare Disorders (NORD), both also participants in the International Rare Disease Research Consortium (IRDiRC) along with the European authorities.

A key recurring element throughout the conversations was the need to understand the value of individual orphan drugs and their contribution to society and the healthcare systems. Society is paying a high price for the burden of diseases and, in some cases, the treatments of patients. Better data collection all along the lifecycle is crucial to understanding the disease and the interventions. Again, cooperation is a key element. As was highlighted in the Danish Presidency intervention in the opening session: why use a database of 5 million patients when you can cooperate to have a database of 500 million? This is particularly important when dealing with low numbers of patients for an individual rare disease – countries can use the legal tools at their disposal to work across borders. The new "Cross-Border Healthcare Directive" supporting European Reference Networks for Rare Diseases and the ability to gather data via working together opens up new avenues to continue to build on European cooperation – rare diseases were a key focus in the legislation, with good reason.

It was stressed that – in the end – patients are asking for action now, not just at some undefined moment in the future. And governments within Europe are responding – for example, exploring opportunities to work together to increase access to orphan drugs in a more uniform way by working together on potential collaborative negotiation approaches. The 15 countries involved in the "Mechanism of Coordinated Access to Orphan Drugs" (MOCA) as part of the EU initiative on Responsibility in the Field of Pharmaceuticals led by the EU Presidency and the European Commission are looking at ways to work together to advance this in a concrete way.

The pharmaceutical industry, too, has a strong role to play. It was noted that, although orphan drug prices are under the spotlight, rare disease patients make up some 6-8% of a country's population, but spending on their therapies remains at just 2-3% of drug spending, while a possible peak of expenses in coming years is expected not to go beyond 4 to 5 %. So are the concerns justified? Or are these concerns simply in the spotlight because they are new? The dynamics of the business model mean that there is an inherent risk for industry and their investors in going into rare disease therapy research and development. Pledges by the authorities to examine new ways of promoting rare disease therapies were welcomed – for

example, repurposing drugs could be a source of many new therapies and hopefully at a lower risk. Companies accept their responsibility in being able to present the value of a product and this could even be reflected in, for example, modulated pricing based on evidence generated and actual value revised years after approval. If authorities are being asked to be innovative in their thinking, industry should also be doing the same. Partnership remains a key focus – across borders, across agencies and institutions, but also within the drug development process. Bringing big and small companies together and looking for more partnership to address more technologies is a key element of continued and future success. This will enable us not only to discover new treatments for diseases without any therapy, but also better ways to treat the diseases that are still only partially addressed – for example, Central Nervous System involvement for certain diseases where the treatments are today only peripherally active.

Building collaborative approaches to authorisation procedures, data collection and novel pricing, reimbursement and access schemes will require dialogue and trust. This is not something that always exists between authorities and the pharmaceutical industry. Behaviours by individual companies influence the way the entire sector is seen. The leadership of companies needs to come together and to talk openly and transparently about how they see their responsibility towards the rare disease communities and the governments that are paying for the treatments. Because this trust will be a vital element of the potential success – or not – of any of the new systems being proposed.

For example, the "CAVOD" process, currently in development to improve informed decisions on access to orphan drugs via optimising processes to share information between countries, based on EU collaboration, will see early dialogue between all stakeholders. If these result in commitments or agreements, a certain trust level needs to exist in order to ensure that people are willing to engage in them. HTA bodies and payers are increasingly talking between each other and together with regulators. Bringing in the industry – or the industry volunteering to step in – is the next logical step that needs to be undertaken. Unfortunately, experiences to date are not always as encouraging as they might be. Coverage with evidence development schemes piloted in the Netherlands as a way to address access issues for orphan drugs have been disappointing in their ability to deliver results. This could be due to several factors, but the suspicion that companies have not always lived up to their promises does lurk in the background as one element contributing to this failure. Rarity of the disease and the complexity of data collection are, of course, confounding factors. But if this is to form the model for the future, trust between all stakeholders will need to be built, earned and protected.

The European legislative system is robust and is delivering – although the continued search for more flexibility and ways within the framework to develop and approve more drugs is strongly welcomed and supported. The key to addressing countries concerns around value and pricing of orphan drugs is something that will have to be developed between the Member States – and, again, a shared approach to a common challenge could be one way forward. New ways of thinking about orphan drugs – for example, multi-criteria decision-making frameworks that take into account the specific characteristics of a given drug and the disease in question – are needed. And, since countries are facing similar challenges, it would make sense to share these across borders. Frameworks are in development by academic researchers as well as being applied already in real-life settings, such as the evaluation system being used by AGNSS in the UK, a direct result of the realisation by NICE back in 2004-2005 that the thresholds being used for "regular" drugs were inappropriate for treatments for very rare conditions.

The ability to develop data is, of course, hampered by the small numbers of patients. But, again, cross-border collaboration and effective systems can be developed to support evidence generation. Rarity, while raising specific challenges, should not be used as a "catch-all" reason to avoid building evidence. Although this might need to be done over longer timeframes than with more conventional drugs. And, further, the processes for doing so should not prevent patients from getting access where a positive risk-benefit is shown and where there is an area of high unmet medical need. The Centres of Expertise provide a controlled, expert-driven environment within which to provide access and the European Reference Networks being established under the new EU Directive on Cross Border Health Care, with a particular focus on rare diseases, will provide the


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infrastructure to support quality evidence-gathering at European level.

Finally, the involvement of the patients at every stage of the research, development and availability chain is a fundamental "must" in the field of rare diseases. This has been one of the cornerstones of the orphan drug system in Europe since the very beginning – patients were included in the COMP as the first committee to allow patient participation. The contributions have been so valued and respected that all new scientific committees established at European level have included patient representatives. In a field where expertise is scarce and scattered and where patients are often the true experts in their condition, it is vital to ensure that their contributions are formally included at all stages. There is no set time for starting the dialogue – it needs to start as early as possible and continue as regularly as possible. This holds true for patients, but also true for all stakeholders within the research, development and availability chain if we are to stand any realistic chance of achieving our shared goal of 200 new orphan drugs by 2020.

Theme 6: Orphan Products & Rare Disease Therapies: Regulatory

The first two sessions of this theme were run jointly with Theme 5. The session descriptions of those sessions can be found in the Theme 5 summary.

In the third session of Theme 6, three perspectives on the topic of "Deployment of the Orphan Drug regulation: predictable flexibility" were presented i) industry, ii) patient representative and iii) regulator.

Catarina Edfjaell, VP and Head of EU and International Regulatory Affairs at Shire HGT, Switzerland highlighted that tools and procedures such as accelerated review and conditional marketing authorisation (CMA) exist within the European Union (EU) legislative framework that could provide more flexibility and speed for Orphan Medicinal Product (OMP) marketing authorisations. Currently we observe that these are being used in a minority of applications. In addition, approval time lines for OMPs in the USA are around 3 months faster than in the EU.

Early dialogue between the applicant and the European Medicines Agency (EMA) is encouraged for study design for OMPs (e.g. acceptance of surrogate endpoints and supplementing with post-marketing and compassionate use data).

Success would ultimately depend on early dialogue between the applicant and the EMA with continuous support from the Committee for Orphan Medicinal Products (COMP) and the EMA orphan drug sector. Increased collaboration between the EMA's scientific bodies could also help e.g. parallel discussions with the Paediatrics Committee (PDCO) and the Scientific Advice Working Party (SAWP). Although the regulatory procedures for OMPs in the EU are working fairly well there is still room for improvement.

Tsveta Schyns, Patient Representative, Member of the PDCO, Belgium reminded the audience that 20% of the EU population is aged under 16 years old and that 50-90% of paediatric medicines have not been evaluated in the correct population. The situation is worse for OMPs in children with rare diseases. The number of orphan designations (OD) for the treatment of conditions affecting children exclusively or adults and children respectively has increased to 60% of all OD. Paediatric Investigation Plans (PIP) should be seen as an integrated part of OMP development. Major hurdles and concerns exist in orphan drug development in the paediatric population such as low numbers of patients, lack of expertise and clinical centres and lack of knowledge about the natural history of the diseases. 'Predictable flexibility' such as flexible criteria regarding endpoints, study population sizes and study design is called for. Collaboration between companies was suggested to perform multi-product, multi –company studies when applicable.

Jordi Llinares, Head of Orphan Medicines at the EMA, EU commented during the panel discussion that the Agency has several projects assessing the development bottlenecks and regulatory hurdles. Several analyses of data on marketing authorisations for orphan and non-orphan products have shown an association between compliance with scientific advice and a higher chance of a positive marketing authorisation opinion. There is a need for sponsors to use the regulatory tools supporting drug development to maximise the chances of successful development. The EU regulations are complex and the Agency is making a new effort to ensure coordination between committees through the 'EMA Scientific Coordination Board'. In summary, 'predictability',

'flexibility' and 'dialogue' between stakeholders were presented as success factors for the continuous improvement of development of medicinal products for rare diseases and paediatric conditions in the EU.


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It is clear that the regulatory process for medicinal products is long and complex. The average time for a medicinal product from research to market authorisation is approximately 10 years. During this time, efficacy has been demonstrated but patients do not have access to the treatment outside of a clinical trial. Compassionate Use Programmes are intended to facilitate access for patients either via cohort or on a named patient basis.

Compassionate Use Programmes were the topic of the next session. The European Compassionate Use Programmes (CUP) legislation is not applied the same way in all Member States in the European Union (EU). The experience of the European Medicines Agency (EMA), Member States and companies is that this lack of harmonisation makes the early access to important new medicines particularly difficult for rare diseases. In order to improve the situation it has been proposed to pursue the dialogue between Member States and companies as well as between Member States themselves by setting up a "Facilitation group" to exchange information and build on common experiences.

The session explored how patients and their organisations can initiate a CUP in their country, and what role they can play. This was illustrated in particular by Czondi, Programmes Coordinator, Sense International, Romania, who described her experience with the national competent authorities, doctors and the company. Chantal Bélorgey, Head of Department of Evaluation of Clinical Trials and Medicinal products of special status, French National Authority ANSM, highlighted the benefit to public health systems that CUP can represent. France has been able to provide 72% of the 64 authorised orphan medicinal products on a compassionate basis on average **35 months prior to the marketing authorisation (MA).**

Arielle North, Ancre Consulting, formerly EMA, is working together with EURORDIS on an initiative to improve information and transparency about CUP in Europe, and also to propose good practices in this domain as programmes vary across Europe and across pharmaceutical companies. Michele Lipucci di Paola, Member of the Committee for Advanced Therapies at the EMA, and Vice-President of the Italian Association for Thalassemia, concluded the session reminding all participants how compassionate use programmes can be useful in saving lives.

As an immediate action, it was agreed to use the opportunity of the next meeting of the Heads of Medicines Agency in Copenhagen in June 2012 where EURORDIS is invited to raise the issue and ask National Competent Authorities to work jointly for an increased transparency on their compassionate use programmes.

Theme 7: Patients' Empowerment

Why do patients need to be empowered?

Terkel Andersen, President EURORDIS affirmed during his presentation that patients are overcoming the "injustice of fate" that has meant we are affected by this challenge of a rare disease in ourselves or our family. Caroline Huyard, author, confirmed we have a desire to change the situation and overcome a sense of loss and weakness, a drive to take back control, and a need to influence improved care, attitudes and policy.


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How are patients empowered?

Empowerment can be a word that is misused just to mean "collective action". Isabelle Ajoulat, Belgian Institute of Health & Societal Research (IRSS) affirmed that empowerment starts with YOU the person. The patient acquires personal growth through action: by giving strength we gain strength; by helping others we help ourselves. The first step is to acquire self-knowledge, recognise the boundaries of our personal capacity, and discover if we are "a (caged) lioness or a pussycat." Both identities are totally acceptable but this process helps us to know our limits and work with them. In this way we win back control of our situation.

Personal knowledge leads to greater self-awareness which leads to better self-care and acceptance. We are then emotionally fitter to access information that is delivered according to our needs as opposed to what professionals think we need. Pam Davies, Climb UK and Denis Costello, EURORDIS stressed that tailored methods and media of communication are particularly important for ultra-rare conditions and specific ethnic groups. Lene Jensen, Rare Disorders Denmark and Stephen Nutt, Rare Diseases UK both affirmed that self-knowledge is gained through personal experience which feeds evidence based documentation as illustrated in Denmark and UK.


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Empowerment through education and training

Personal training provides the building blocks for empowerment, and develops the tools for coping strategies and communication skills to systems that will provide additional support.

CAMPS act as therapeutic training and can help extend the limits (real or imagined) of the individual affected by a rare disease. They encourage a "CAN DO" attitude, which is built through social interaction, cultural exchange, and holistic programmes bringing participants out of isolation. Examples of this were presented by Zsuzsanna Poganyné Boitor, Hungarian Williams Syndrome and Terence Dignan, Orphane Europe Recordati Group. By adding training to our experience and collective work processes, we can, if we chose, become patient advocates.

Operational influence. Terkel Andersen, EURORDIS and Dorica Dan, Romanian Prader Willi Association both testified that patient organisations provide tools to create a more caring society. They are often pioneers in this field. They act as part of the system not just the beneficiaries of it. The input of all stakeholders is complementary.

Political Influence. Moreover, Josep Torrent i Farnell, Fundacio Doctor Robert, Spain explained that the advocacy actions of rare disease organisations has forced political strategists to consider the approach to health policy development from "bottom up" rather than "top down". This influence is also evident in the involvement of patients in the development of National Plans at drafting and consultation levels, as expressed by Stephen Nutt, Rare Diseases UK, and the channelling of patient experience and knowledge into skills as advocates in scientific committees at the European Medicines Agency (EMA), as asserted by Lise Murphy, Swedish Marfan Association.

Patient organisations have proved an effective and respected link between the medical, social and educational professionals and the politicians, assisting in providing policy makers with evidence of the needs of the professionals who act for those with rare diseases.

Patient Organisations often have an innovative approach to problem solving in several areas as demonstrated in the development of rare disease collaborative projects. They have become skilled at learning how to link patient needs to emerging political agendas. Consequently there is a growing recognition by policy makers that they need the Rare Disease Community for policy development as much as it needs them.

What is stronger through the action of empowered patients?

Identity of the Rare Disease Concept is more widely recognised and a willingness to collaborate in projects and share best practices. Dorica Dan, Romanian Prader Willi Association and Birthe Byskov Holm, Rare Disorders Denmark share the opinion that this is having a beneficial influence on social care and interaction. There is a growing acceptance by politicians of our added value with less suspicion and more respect. We have EARNED greater legitimacy.

Empowerment through Research and Registers

Patients approach Patient Organisations for support of many kinds but of huge support is the knowledge that there is research because this brings hope for the future.

Registers help researchers and the community to understand the disease natural history, disease mechanism, building evidence, building partnerships, and medical models. Nick Sireau, AKU Society and Nitisinone UK explained that this is most effectively done by building a community to support the research base for a clinical trial that will lead to a treatment which could ultimately cost a fraction of the existing total annual care costs relating to a specific rare disease.

People Living with Rare Diseases have harnessed their personal skills to create research projects, influence clinical trial development, and clinical trial design focusing on

meaningful endpoints and patient outcomes. Elisabeth Vroom, Duchenne Patient Project, Netherlands and Ulrike Pypops, Association Muco Vereniging, Belgium stated that they have shown how patient needs must be part of the study design.

Mapping of Rare Disease Associations, laboratories where Rare Diseases are diagnosed, Rare Disease clusters, centres of expertise and experts are all exercises that increase our visibility, pool of knowledge and critical mass via both qualitative and quantitative studies.

Challenges that remain

Jana Petrenko, Czech Coalition for Health and Anna Arellanesova, Czech Cystic Fibrosis Association affirmed that volatile political backgrounds make it hard to maintain stable relationships with policy makers. Pauline Evers, Federation of Cancer Patients Organisations, Netherlands stressed that connections with relevant Government departments and ministries rather than ministers who can change with political parties is a valid approach. The rare disease movement can struggle to maintain the influence won over several years because of changing political environments and challenging economies.

Patient Organisations have limited resources both human and financial while policy makers have more demands placed on diminishing budgets as the economic crisis continues.

There is a continuing challenge to gaining a speedy diagnosis, promoting research and accessing medicinal products that are available and at an acceptable cost.

There needs to be more focus on developing social support for people with rare diseases in a sustained way, while maintaining a flexible approach according to the cultural, ethical and political environment of each country. It is essential to adopt a different focus and approach for different populations in different countries and consequently, to develop different methods of sustainability.

Tribute to Torben Gronnebaek

On behalf of EURORDIS and the members of the ECRD 2012 Programme Committee.

Torben was a person who experienced the pain of living with a rare disease and from that the passion, the drive and the hope that things could be made better. He knew we are stronger together and embraced the empowerment that working together provides to patients and the hope it brings to our future. Torben knew the umbrella brings shelter.

Torben was one of those “exceptions” as are all those people who are touched by a rare disease.

Poster Authors with Titles

Best poster awards

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A Route Map for the Patients Journey, 24-May

P 54 *Rabea Wagstaff, Vincent Brandenburg, Markus Ketteler*
Report from the International Collaborative Calciphylaxis Network
(ICCN, Calcific uremic Artiolopathy Registry) http://www.calciphylaxis. de, 24-May

P 265 *Holm Graessner, Julia Giehl, Olaf Riess*
First German Academy for Further Medical Training on Rare Diseases
(FAKSE, www.fakse.info), 24 & 25 May

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Kate Bushby, Co-Chair ECRD 2012 Programme Committee, presenting Paola Pierri with certificate for poster winner.

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Exhibitors

Centogene GmbH, Germany

Centogene is one of the leading laboratories focusing on genetic testing for rare hereditary disorders. We now offer more than 1650 routine genetic and biochemical tests.
In addition we perform analysis of biomarkers for lysosomal storage disorders. Centogene works with academic and industrial partners to develop cutting– edge diagnostic techniques and new orphan drugs. www.centogene.com

DIA Europe, Switzerland

The DIA is a global association of approximately 18,000 members who are involved in the discovery, development, regulation, surveillance or marketing of pharmaceuticals or related products. The DIA is committed to the broad dissemination of information on the development of new medicines or generics, biosimilars, medical devices and combination products with continuously improved professional practice as the goal. The DIA is an independent non-profit organisation. The voluntary efforts of DIA members and speakers allow the DIA to organise conferences, workshops and training courses and provide educational publications. www.diahome.org

European Commission, Luxembourg

The strategic objective of the European Commission in the field of rare diseases is aimed at improving the chance for patients to get appropriate and timely diagnosis, information and care through European action to support Member States in acting more efficiently than they can on their own.
To put in place their strategy the Commission will be assisted by an EU Committee of Experts on Rare Diseases (EUCERD) to advise on the implementation of the Communication and the Recommendation. The Committee will be assisted by a Scientific Secretariat, supported through the Health Programme.
Composed of 51 members representing Member States, patient's organisations, Pharmaceutical industry, rare diseases research Projects, Health Programme projects and the ECDC.

European Society of Human Genetics (ESHG) & EuroGentest (EGT), Austria

ESHG promotes research in basic & applied human & medical genetics, to ensure high standards in clinical practice, and to facilitate contacts between all persons sharing these aims.
EGT encourages harmonization of standards & practice in all areas of genetic testing in the EU & beyond. Patient leaflets with general information in a comprehensive way are created in over 27 languages: www.eurogentest.org

Serious Fun Children's Network, Hungary

Serious Fun Children's Network is a European and American family of camps that organise therapeutic camps for children with serious illness and their families free of charge. The camp is not about illness but fun, hope and overcoming personal limitations. The camp in Ireland, Italy, UK, France, Hungary offers life-changing experiences for children and volunteers. We serve about 3000 children per year from 26 European countries with the help of 2200 volunteers: www.batortabor.hu

Intermune International AG, Switzerland

InterMune is a biotechnology company focused on the research, development and commercialization of innovative therapies in pulmonology and fibrotic diseases. In pulmonology, we are focused on therapies for the treatment of idiopathic pulmonary fibrosis, a progressive and fatal lung disease. www.intermune.com

Orphanet, France

Orphanet is a free access European database dedicated to information on rare diseases and orphan drugs. Orphanet offers services adapted to the needs of patients and their families, health professionals and researchers, support groups and industry. Orphanet includes an encyclopaedia and a directory of services in Europe: www.orpha.net

PPTA, Belgium

PPTA is the primary advocate for the world's leading producers of plasma-derived and recombinant analogue medicinal products. The medicines produced by PPTA members are used to treat patients suffering from rare life-threatening/impairing disorders and serious conditions including bleeding disorders, immune system deficiencies, burns and shock. More under: www.pptaglobal.org

Visit Brussels

VISITBRUSSELS is the communications agency for tourism in the Brussels-Capital Region; its aim is to promote and strengthen the image of the capital of 500 million Europeans: www.visitbrussels.be

Notes

Berlin 2014
7th European Conference on Rare Diseases & Orphan Products
Dates & Venue to be announced soon

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