8-10 May 2014 Berlin

European Conference on Rare Diseases & Orphan Products

EURORDIS
Rare Diseases Europe

National Conferences
EUCERD / Joint Action / EUROPLAN – Key Priorities and Way Forward

The development of national plans for rare diseases in Europe is the result of a two-way communication between the European level and the national level

EURORDIS, National Alliances and RD patient organisations are active at all levels

PATIENT ORG

EU LEVEL

- EU Policy Texts, Regulations, Directives, Recommendations
- Assessment of transposition in national systems
- EU Member States develop RD Plan
- Implementation at national, regional, local levels

PATIENT ORG

NATIONAL LEVEL

- Inputs from local, regional institutions to National government
- Participation of EU Member States to EU Parliament & Committees
- EU MSs provide inputs to shape EU Rare Disease Policy

25 CONFERENCES WITH 1 COMMON OBJECTIVE & 1 COMMON STRATEGY

OBJECTIVE *:*

"to support the necessary steps towards outlining high quality National Plans or Strategies on RD with concrete objectives in each field"

STRATEGY:

Integrated EU/National - Promote EU reference documents on RD policy (incl. EUCERD measures) & assess their transferability at national level

Comprehensive – Across main themes of the Council Recommendation

Long-term - Sustain grassroots movement in favour of national plans or strategies for RDs

Multi-stakeholders - Involve all stakeholders, broadest possible outreach

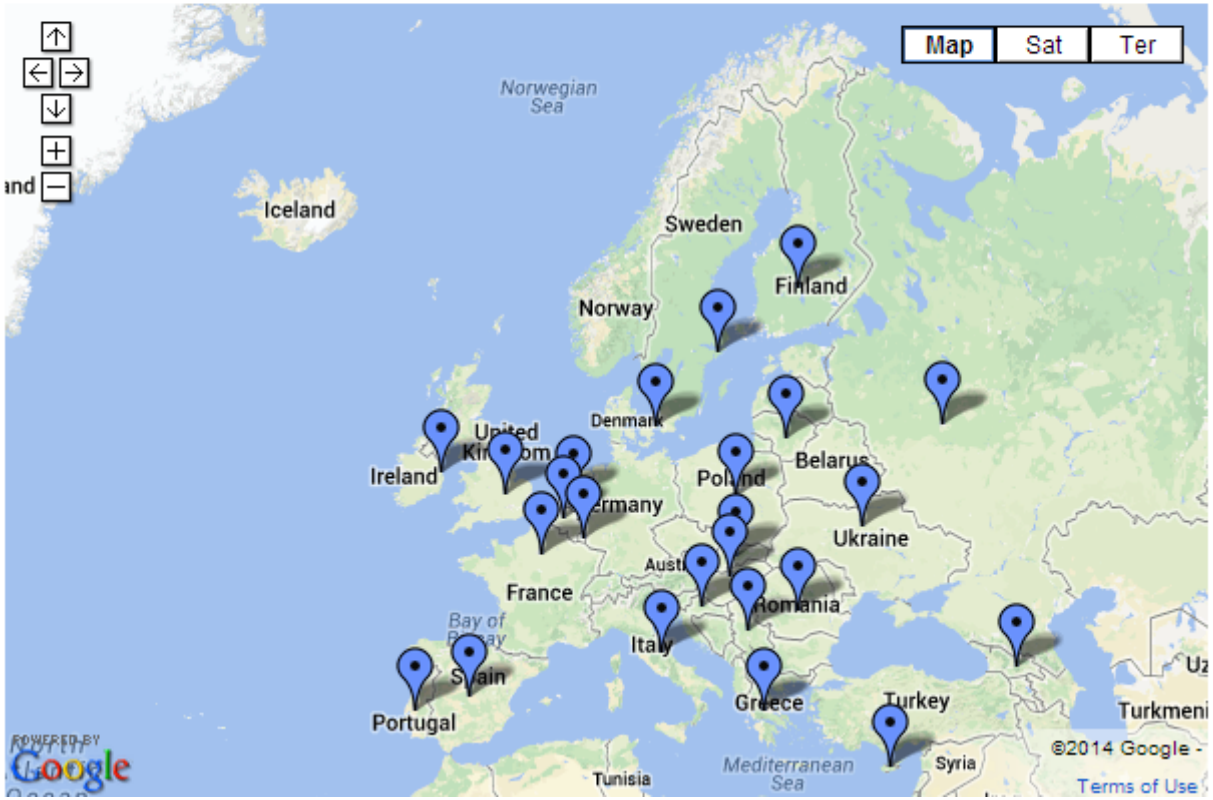
25 CONFERENCES OVER 2012 - 2015

EUROPLAN National Conferences 2012-2015

ShareThis

On this map you can find when and where the EUROPLAN National Conferences are being held and details about the organisers, in most cases National Alliances (NA) of Rare Disease patient organisations.

- Final report of each EUROPLAN National Conference



25 National Conferences
to promote RD National Plans in EU and beyond

Belgium
Croatia
Cyprus
Denmark
Finland
France
Greece
Hungary
Ireland
Italy
Lithuania
Luxembourg
Netherlands
Poland
Portugal
Slovakia
Romania
Spain
Sweden
UK

+
Georgia
Serbia
Switzerland
Russia
Ukraine

EURORDIS Rare Diseases Europe

CONFERENCES 2012 - 2015

Date	Country	EURORDIS Advisor	Conference Organiser	Debrief Session
2012				
26 Nov	SWEDEN	Maria Gardsäter	Sällsynta Diagnoser	Possibly in 2014
1 Dec	GREECE	Simona Bellagambi	PESPA	Possibly in 2014
2013				
27 -28 Feb	SLOVAKIA	Dorica Dan	Slovak Alliance of Rare Diseases	YES
28 Feb	RUSSIA	Oleg Kvlividze	Russian patients union + National Ass. of Organis. Patients with RDs "Genetica" + "Union of patients and POs with RDs"	NO
17 Apr	GEORGIA	Oleg Kvlividze	GeRad	NO
27 Mar	UKRAINE	Oleg Kvlividze	CSMA; Kharkiv Charitable Foundation Children with spinal muscular atrophy	NO
24-25 May	ROMANIA	Dorica Dan	RONARD	YES
21 Sep 2013	FINLAND	Maria Gardsäter	Finnish Rare Diseases Alliance	YES
27-28 Sep	POLAND	Vlasta Zmazek	ORPHAN	YES
25-26 Oct	HUNGARY	Dorica Dan	HUFERDIS	YES
13-14 Nov	LITHUANIA	Yann Le Cam/ Valentina Bottarelli	Ministry of Health / Vilnius University	YES
14-15 Nov	CYPRUS	Lily Cannon	CARD	YES
14-15 Nov	NETHERLANDS	Melissa Hillier/ Farhana Ali	VSOP	Possibly in 2014
19-20 Nov	LUXEMBOURG	Lily Cannon	ALAN	YES
5-7 Dec	SERBIA	Vlasta Zmazek	NORBS	YES

CONFERENCES 2012 - 2015

Date	Country	EURORDIS Advisor	Conference Organiser	Debrief Session
2014				
13 Jan	FRANCE	Christel Nourissier	Alliance Maladies Rares	NO
27-28 Jan	ITALY	Simona Bellagambi	UNIAMO	YES
27-28 Feb	CROATIA	Vlasta Zmazek	Croatian Alliance for Rare Diseases	YES
28 Feb	BELGIUM	Lene Jensen	RaDiOrg	YES
24 Jun	UK	M. Hillier / Farhana Ali	Rare Disease UK	Planned
Autumn 2014	SPAIN	Simona Bellagambi	FEDER	Planned
October	DENMARK	Lene Jensen	RDD	Planned
Nov 2014 - Feb 2015	IRELAND	Avril Daly	GRDO	Planned
2015				
Early 2015	PORTUGAL	Lene Jensen	APADR	Planned
Possibly 2014-15	SWITZERLAND	V. Bottarelli	ProRaris	?

APPROACH (I)

Since its inception in 2008, EUROPLAN National Conferences promote and accompany the development and adoption of National Plans or Strategies for Rare Diseases in the EU Member States based on a common policy and legal framework:

- → Communication of the Commission « Rare diseases, Europe's challenge » (2008)
- → Council Recommendation on rare diseases (2009)
- → Directive on the application of patients' rights in cross-border healthcare (2011)
  - establishing European Reference Networks

APPROACH (II)

This vision and robust policy base is turned into reality through technical guidance & support in priority areas:

→ EUCERD Recommendations:

- on **Quality Criteria of Centres of Expertise**, 2011
- on the information flow on **Clinical Added Value of Orphan Medicinal Products** (**CAVOMP**), 2012
- on **European Reference Networks**, 2013
- on **Rare Disease Patient Registries and Data Collection**, 2013
- on **Core indicators for National Plans**, 2013

→ EUCERD Joint Action (Work Package 6):

- Guiding Principles for advising **Specialised Social Services** on the integration of People Living With Rare Diseases

In National Conferences 2012-2015 integrates the guidance documents adopted by the EUCERD and promote their dissemination

Content Guidelines

- 6 Content Guidelines, 6 **main themes,** one for each National Conference's Workshop
- Provide the conference organisers / Steering Committees /Workshops' Chairs with the necessary background information and questions to prompt the discussion during the Workshops
- Each CG contains:
  - All **relevant EU documents & materials** providing the necessary background on the theme
  - a list of questions to be addressed during the Workshop
- Reflected in the Final Reports

➔ **CONTENT GUIDELINES:**
http://www.eurordis.org/content/documents-organise-national-conference

6 MAIN THEMES

1. Methodology and Governance of a NP
2. Definition & inventorying (Information & Training)
3. Research on RD
4. Care: Centres of Expertise & EU Reference Networks
5. Orphan Drugs
6. Social Services for RD

Same themes as in:

- 2009 EU Council Recommendation
  EUROPLAN I Recommendations
- EUCERD Core Indicators
- National Conferences Content Guidelines and EUCERD Recommendations
- EUROPLAN II NC Workshops

EMERGING OUTCOMES OF EUROPLAN NATIONAL CONFERENCES

From the thematic discussions at National Conferences (*a still ongoing process!)*

Key areas of discussions and hurdles emerge as well as recurring needs and problems

Possibly areas for more coordinated action, exchanges and policy guidance

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Theme 1 - Methodology, Governance and Monitoring of the National Plan

Sub-Themes:

1.1 Mapping policies and resources
1.2 Development of a National Plan /Strategy
1.3 Structure of a National Plan /Strategy
1.4 Governance of a National Plan
1.5 Dissemination and communication on the National Plan
1.6 Monitoring and evaluation of the National Plan
1.7 Sustainability of the National Plan

Theme 2 - Definition, codification and inventorying of RD

Sub-Themes:

2.1 Definition of RD
2.2 Codification of RD and traceability in national health system
2.3 Registries and databases
2.4 Information on available care for RDs in general, for different audiences
2.5 Help Lines
2.6 Training healthcare professionals to recognise and code RD
2.7. Training healthcare professionals

Theme 3 - Research on RD

Sub-Themes:

3.1 Mapping of existing research resources, infrastructures and programmes for RDs
3.2 Dedicated RD research programmes and governance of RD research funds
3.3 Sustainability of research programmes on RD
3.4 Needs and priorities for research in the field of RDs
3.5 Fostering interest and participation of national laboratories and researchers, patients and patient organisations in RD research projects
3.6 RD research infrastructures and registries

patient involvement

codification of RDs

registries and DBs

comprehensive info systems on RDs

RD research

EURORDIS Rare Diseases Europe

Theme 4 – Care for RDs - Centres of Expertise and European Reference Networks for Rare Diseases

Sub-Themes:

- 4.1 Designation and evaluation of CE
- 4.2 Scope and functioning of CEs
- 4.3 Multidisciplinarity, healthcare pathways & continuity of care
- 4.4 Access to information
- 4.5 Research in CEs – How to integrate research on RDs and provision of care
- 4.6 Good practice guidelines
- 4.7 Diagnostic and genetic testing
- 4.8 Screening policies
- 4.9 European and international collaboration – Cross-border healthcare and ERNs (European Reference Networks)
- 4.10 Sustainability of CEs

Theme 5 – Orphan Medicinal Products

Sub-Themes:

- 5.1 Support to Orphan Drug (OD) development
- 5.2. Access to treatments
- 5.3. Compassionate use programmes
- 5.4. Off label use of medicinal products
- 5.5. Pharmacovigilance

Theme 6 –Social Services for Rare Diseases

Sub-Themes

- 6.1. Social resources for people with disabilities
- 6.2. Specialised social services for rare diseases
- 6.3. Policies to integrate people living with rare diseases into daily life
- 6.4. International–supranational dimension

Additional Workshops (*optional*)

Report of the Closing Session - Conclusions

CEs & healthcare pathways

good practice guidelines

genetic testing & NGS

ERNs

access to treatments

social policy

EURORDIS Rare Diseases Europe

Areas with enough guidance
but might need additional support
through exchange of best practices
across EU Member states

Implementation, monitoring of NP

- **Patient representatives are not involved in the actual implementation and monitoring of the NP/NS in a number of countries**

what exists

- **EU Council Recommendation**
- **EUROPLAN Recommendations for the development of RD NPs (Guidance document)**

what additional support?

→ Exchange of **best practices across countries** is an urgent need, with focus on specific solutions adopted in specific countries

Centres of Expertise & Healthcare Pathways

- **In many EU countries (even where a NP is adopted) identification and designation of CEs are still ongoing**
- **A point of central relevance – how to structure relations with expertise & care provision in the country and network with expertise outside the country (ERNs)**
- **How to define and individual patient healthcare pathways**

what exists

- **EUCERD Recommendations establish CE Quality Criteria**
- Outcomes of **EUCERD Joint Action Work Package 7** on "*Improving access to higher quality healthcare in RD*" – how CE are structured and integrated in HC systems

what additional support?

→ Exchange of **best practices** (e.g. filières in France) among countries of similar size, healthcare system, historical process…

European Reference Networks

- **Participation to ERNs spurs the ongoing process of CE designation**
- **A lot of unclear points, calls for greater clarity and guidance – how to join ERNs in practice, links to Cross-border Healthcare (CBHC) Dir.**

what exists

- **CBHC Directive**

+ EC Implementing and Delegated Acts

- **EUCERD Recommendations on RD ERNs**

what additional support?

→ Identification of ERNs by therapeutic areas

→ Exchange of **best practices**

→ Capacity building /information activities to support ERN establishment and MS in CE designation role

Registries, Databases

- **Setting up national registries is a priority action in many countries where it does not exist**
- **Also emerging priorities: better linking of existing ones; sustainability; interoperability**

what exists

- **EUCERD Recommendations on RD patient registries and data collection**
- EPIRARE project /EUCERD Joint Action WP8 - **Minimum Dataset**
- Upcoming **JRC Platform**

what additional support?

→ Advisory role to JRC and Exchange of **best practices**

→ Including in broader context of **information flow**, **eHealth for RD** –how to integrate for ex. Electronic Health Records while preserving specific RD needs?

Research on RD

- **In most countries no RD specific research programmes**
- **Some countries already members of E-RARE and/or IRDiRC**

what exists

- **Consortia-type initiatives E-RARE, IRDiRC**

what additional support?

→ Exchange of **best practices** (e.g. experiences of specific RD calls or programmes) ?

→ Sharing roadmap and funding priorities amongst IRDiRC members

Areas where there is need for new guidance

To be provided by EUCERD (CERD)

Codification of RD

- **Recurring call for solid RD codification as a basis for a healthcare system that can effectively respond to RD patient needs**
- **Overall use of ICD-10 and widespread wish to integrate and expand use of OrphaCode**

what exists

- **EUCERD Joint Action WP5 and ongoing work in ICD-11 with specific RD nomenclature (but not before 2017 !)**

what additional support?

→ Technical CERD Recommendations on use of using OrphaCode alongside existing systems of codification at national level?

→ Exchange of best practices on implementation?

Comprehensive information system for RDs

- **Emphasis on the difficulties in accessing high-quality information and general call for improved information on RDs**
- **Create and make better use of quality assured helplines for both professionals and people/families living RDS**

what exists

- Key role of **ORPHANET** and national websites
- **Dir. 2011/24/EU on Cross Board HealthCare** creating **National Contact Points**
- **Help lines - European Network of RD Help Lines**
- **Patient groups helplines & website + new CoE website**

what additional support?

→ Technical CERD Recommendation – common principles

(Included in broader context of **information flow for RD** hence linked to registries, codification, pharmacovigilance, cross border healthcare and ERNs)

Good practice guidelines

- **Topic addressed in a limited number of National Conferences**
- **Subsidiarity - share work at appropriate level within the country (regional/national level) e.g. in Belgium, or between the country and the EU level, e.g. Sweden**

what exists

- **RARE-BestPractices project (ongoing)**

what additional support?

→ Technical CERD Recommendations on Good Practice Guidelines ? Principles, methods, gathering of experience at EU level, sharing of work between MSs

Genetic testing, Next Generation Sequencing

- **Availability and cost of genetic testing, accessibility for all patients and cost borne by HC system**
- **Cross-border genetic testing often required esp. in small/medium countries**

what exists

- **EUCERD Joint Action WP8 – ongoing research to explore crossborder genetic testing for RD**
- **EuroGenTest**

what additional support?

→ Technical Recommendations /guidance on how to implement and use Next Generation Sequencing technology for RD

Access to treatment

- **Access to OMPs having received Centralised Market Authorisation depends on national mechanism for pricing and reimbursement**
- **Gathering of expertise at EU level for coordination of clinical value assessment, linkage of price to value + volume + post-MA research**

what exists

- **EUCERD Recommendations on Clinical Added Value of OMPs (CAVOMP)**
- **Draft « Long-term provision on EU cooperation on HTA » by HTA network**

what additional support?

→ Greater coordination in HTA pricing and reimbursement – backing of HTA ongoing Strategy

→ Build on HTA network, MOCA Working Group & MEDEV, Managed Entry Agreement working group

→ Greater post-marketing collaboration

Social Policy

- **Still a general lack of coordination between HC and social systems for People Living With Rare Diseases (PLWRD)**
- **A number of pending issues - how to ensure coordinated care for PLWRD including social care, visibility of RD in the disability schemes**

what exists

- **EUCERD Joint Action WP6 notably Guiding Principles advising Specialised Social Services on the integration of PLWRD**

what additional support?

→ Exchange of **best practices** (interaction of social services with HC, case studies of patient-need based services rather than organisation-based services) ?

→ Technical Recommendations on Social Policy measures pertaining to PLWRD ?

→ Support to Orphanet for indexing the functional consequences of rare diseases with the Orphanet Disability Thesaurus?

WHERE ARE WE ?

1) The EU framework is established and in place

- **18 out of 28 EU MS have a NP/NS adopted….**
- **…but :**
  - not all areas covered and/or
  - most actions without funding allocations and/or
  - many policy measures difficult to implement and/or
  - some disease areas left uncovered

So we need to think to the next phase of National Plans to reach an integrated, comprehensive and long term strategy !

WHAT'S NEXT ?

2) There is still a need to work together on those essential areas where concrete actions should be expected in all MS

- EU needs to c**ontinue providing support in the implementation of cost-effective measures** to reinforce NP/NS in specific **essential areas** for the **next phase of NP/NS**
- There is a need to continue conveying EU guidance - role of **Commission Expert Group on RD**
- Focused on the implementation at national level and the **cross-feeding** of experience and **good practices** developed across Europe

BACKGROUND SLIDES

EUROPLAN – 25 CONFERENCES:

1 **COMMON FORMAT** THAT CAN BE ADAPTED TO THE NATIONAL SITUATION

- → Conferences are **outcome-oriented**
- → Conferences are organised around **Workshops**, with opening and closing plenary session(s)
- → Workshops set up according to specific **themes**
- → Workshops deliver concrete proposals for the plenary
- → Specific guidance questions and presentations are provided (CONTENT GUIDELINES) based on
- → EUCERD Core Indicators are discussed at NC
- → Conferences deliver a Final Report using a common template

➔ **COMMON FORMAT document:**
http://www.eurordis.org/sites/default/files/europlan-ntional-conferences-2012-2015-common-format.pdf

COMMON FORMAT - PLENARY

Plenary Session (opening session)	• Key EU /EUROPLAN documents • Country's status quo
Parallel Workshops	6 Main THEMES (Content Guidelines)
Plenary Session (closing session)	• Report from Workshops • Wrap-up and debate

EUROPLAN – CONTENT GUIDELINES

Background documents

Sub-theme

Full list of questions to select for discussion

RESOURCES	TOPICS for DISCUSSION
4.1. Designation and evaluation of CE	
Council Recommendation 11. Identify appropriate centres of expertise throughout their national territory by the end of 2013, and consider supporting their creation. **EUROPLAN recommendations** R 4.1 Well defined mechanisms of designation of centres of expertise are established and their quality is assured, efficiency and long term sustainability. *See also page 43 of EUROPLAN Recommendations for case studies of Identification and Designation of Centres of Expertise in DK, FR, IT, SP, UK; and page 44 for case studies of evaluation of CEs in DK, FR, SP, UK.* R 4.4 A national directory of Centres of expertise is compiled and made publicly available. **EUCERD Recommendations for Quality Criteria for CEs** **"Mission and scope of centres of expertise (CEs) for rare diseases (RD) in Member States (MS)"** 16. A national directory of formally designated CEs is compiled and made publicly available, including on the Orphanet portal. **"Criteria for designation of CEs for RD in MS" (17-32)** 17. Capacity to produce and adhere to good practice guidelines for diagnosis and care. 18. Quality management in place to assure quality of care, including National and European legal provisions, and participation in internal and external quality schemes	**Mapping of CEs** • Starting from the recognition that expertise on RDs exist in all countries, (see Final Report of EUROPLAN I conferences, Area 4) what is the level of knowledge of the existing expertise in the country? Is there a mapping of structures providing expertise on rare diseases? • Have their different roles and competences been acknowledged? **Designation criteria** • Are designation criteria being defined? If not, is there a procedure in place to define and approve such designation criteria? • Are the designation criteria such to adapt to the characteristics of the disease or group of diseases covered by each CE? • What sort of quality management is ensured within CEs throughout the national territory? • Please compare the designation criteria adopted in your country with the EUCERD Recommendations on Quality Criteria for CEs (*see left column*). What recommended criteria are missing? Which ones could be incorporated? **Designation process of CEs** • At what stage of development is the process of designation of CEs in your country?

Centres of Expertise & healthcare pathways

→ Consider a step wise & realistic approach :

- Identification of experts, supported to coordinate multidisciplinary skills with some budget allocation
- Candidate centres encouraged to define their current actions, their goals and their strategy to attain designation criteria
- Patient organisations actively involved at all levels: identification, collaboration in their activities, internal and external evaluation
- Collaboration with other centres and experts at national, European and international level being essential
- Mechanisms to measure performance and progress should be laid out
- KEEP in MIND: The combined scope of centres should cover all patients needs at national level in the long term