Report

EUCERD Joint Action Workshop

"EUROPLAN National Plans/Strategies Capacity Building Activities"

Federal Ministry of Health, Berlin

8 May 2014

Background

In the framework of the EUCERD Joint Action (EJA) N° 2011 22 01, EUROPLAN WP4 has organised a four hour workshop on 8 May 2014, in Berlin, back to back with the EURORDIS biannual European Conference on Rare Diseases and Orphan Products (ECRD).

The workshop meant to highlight the results of two studies and other relevant information that provide the base for the development of an integrated, participated, bottom up and country-tailored approach to capacity building in the field of National Plans / Strategies (NP/S) for Rare Diseases (RD) across Europe. The results of the National Conferences and the ensuing Debrief sessions, the results of a newly conducted study on the Indicators and the analysis of the NP/S for RDs, have contributed to provide a picture of the main expressed and derived needs of MS that can be translated into a plan of action for capacity building.

However, it was felt that, in order to elaborate a plan of action, additional information was needed. For this reason, a questionnaire was designed to be administered on-site to the participants of the workshop. The questionnaire was meant to match the priority needs with a number of actions but it was decided instead to view it as a pilot for future insights in order to analyse some trends and methodological issues which must be substantiated by further data collection.

The Workshop opened at 14:00

A. Introduction

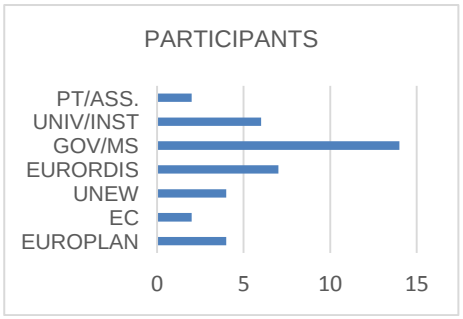
The EUROPLAN NP/S capacity building activities had two general objectives:
1. Highlight the implementation status of the NP/S of RD in Europe; and
2. Translate MS needs into effective actions.

The workshop was chaired by Dr Domenica Taruscio, EUROPLAN (WP4) Leader and by Prof. Kate Bushby, EJA Leader. From the European Commission Dr Antoni Monserrat Moliner and Mr Georgios Margetidis took part in the meeting.

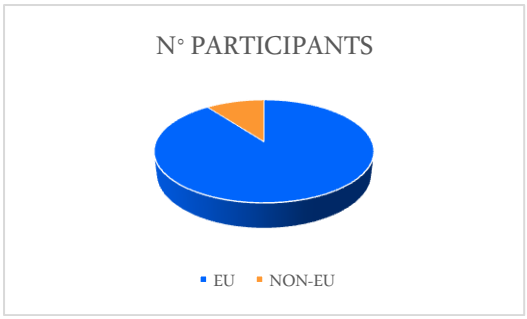
After the conventional words of welcome and a brief presentation of participants, Dr Taruscio opened the workshop with a short introduction that set the pace for the activities of the workshop. She presented the *Council Recommendation on an action in the field of RDs* of 8 June 2009 and highlighted, in particular, the seven areas of the Council Recommendation. After the presentation of the programme, the workshop commenced with the planned presentations and the other activities. Dr Taruscio's presentation is in Annex 1.

The participants

The total number of registered participants was 39, including the 2 EC Representatives, coming from 25 European and non-European Countries. Graphic 1 shows the breakdown of participants according to the institution they represent while Graphic 2 illustrates where participants come from. A list of Participants is found in Annex 2.



Graphic 1



Graphic 2

B. Presentations

After the introduction, the first part of the workshop comprised five presentations.

1. Overview of the Rare Disease National Plans/Strategies (NP/S) in Europe
A. Monserrat Moliner (European Commission)

Dr. Monserrat Moliner provided a general overview of current situation of Rare Diseases NP/S in Europe highlighting the fact that most member states have sought to comply with with the *Council Recommendation on an action in the field of RDs* of 8 June 2009.

According to the Recommendation, MS should have elaborated and adopted a plan or strategy for RDs by the end of 2013 and the Commission initially provided European guidelines for the definition of the action plans for RDs through the EUROPLAN project (2008-2011). He has underlined the importance of EUROPLAN, this being the only EU funded project explicitly mentioned in the text of the Council Recommendation.
The stress on the development of specific NP/S derived from the fact that the lack of health policies specifically targeted at RDs may have resulted in delayed diagnosis and difficult access for RD patients to appropriate treatment and care.

NP/S should contain integrated and comprehensive health policy actions for RD, to be carried out at national level. A plan should contain specific objectives, actions, resources, a time frame and an evaluation process based on specific indicators (EUROPLAN Definition). Given the nature of RDs, the international dimension regarding key topics such as, classification and codification, reference networks, orphan drugs, and research should also be included in the NP/S.

In this respect, most of the MS, by the end of 2013, had adopted a NP/S. This is a good achievement considering the fact that in 2009 only a few MS had produced a NP/S (Bulgaria, France, Portugal and Spain). Meanwhile, the remaining MS are in the process of defining/adopting a NP/S. On the other hand, there is the case of France which is well underway in implementing the second NP/S on RDs.

So far as the existing NP/S are concerned, Dr Monserrat Moliner has pointed out that these can be catalogued into four categories:
1. Some NP/S could be defined as declarative, since they are based on declarations of intentions;
2. NP/S that concentrate on issues regarding the solution of problems between competent authorities as in the case of Regions and the Central State. This is the case, for example, of the Spanish and German NP/S.
3. Some NP/S are monothematic, concentrating efforts and resources on, for example, public health, epidemiology, orphan drugs, etc.
4. The remaining NP/S are examples of integrated plans.

The issue regarding the budgetary situation of NP/S was also mentioned, whereby only few NP/S indicate the monetary investment into the planned activities.

Special mention was made regarding rare cancers, whose plans run separately. It was proposed that perhaps efforts should be made to integrate them into the RD larger picture. Montserrat's presentation is in Annex 3.

2. Analysis of specific aspects of the Rare Disease NP/S

R. G. Frazzica (EUROPLAN Coordinating Team)

The results of a review of all NP/S for RDs in Europe was presented (Annex 4). The review was based on the *Council Recommendation on an action in the field of RDs* adopted on 8 June 2009 which involves twenty Recommendations gathered into seven main areas: 1) Plans and strategies in the field of RDs; 2) Adequate definition, codification and inventorying of RDs; 3) Research on RDs; 4) Centres of expertise (CEs) and European reference networks (ERNs); 5) Gathering the expertise on RDs at European level; 6) Empowerment of patient organisations; 7) Sustainability.

The goal of the current review was to analyse the available NP/S to determine if the elements of the Council Recommendation were included and what criteria were used in the definition of the plans/strategies. When feasible, a cross-country comparison was made. The review process comprised two main phases:

- Pre-Review Phase
  During this phase, the preliminary work was carried out. All documents included in the EUROPLAN website (Section "National Plans" - List) were assembled ,ready to be studied. Then, some relevant terms in relation to planning were selected and defined and these became the assumptions used in reviewing the NP/S. Thereafter, the documents were read extensively. Once this activity was completed, a number of items were identified for collection and a number of grids were designed to be used for the data collection.

- Review Phase
  The screening of these documents took several readings and checking of the documents.

  a. First, a decision was taken regarding inclusion or exclusion of NP/S from the formal review. Three documents did not undergo further analysis since they did not meet the defined criteria for the analysis of the national plans: one was a review document, one was a general health plan and the third one was a situation analysis. The remaining 18 NP/S were reviewed according to the Language used. A total of 8 NP/S were written both in English and in the native language, two NP/S were written only in English, one of them was from a non-Anglophone Country. The 10 NP/S available in English were included for review. Out the remaining 8 NP/S in native languages, 3 were included because they were written in a language known to the reviewer, whilst 5 were excluded because of the language barrier. The total number of NP/S reviewed was therefore 13.
  b. At this point, the included NP/S were reviewed and comprised eight Plans and five Strategies. The detailed results are found in the enclosed Annex 4.

The findings presented were further clarified by providing some additional information. The general consideration is that there is still a need to clarify some of the planning elements. Following are some examples.

a. There is no clear understanding of the different types of plans. A strategic plan is different to a tactical plan or an operational plan. Each should have a number of characteristics that should be present if a MS decides to undertake the adoption of one or the other type of plan.

b. The *situation analysis*, which is the first step in the planning process, is missing in most NP/S. Some NP/S, and these were considered positively in the review, included some information about the Country situation but not a concrete situation analysis. There were NP/S with no information regarding the Country situation.
c. Most NP/S have goals and/or general objectives, and both are difficult to monitor or evaluate, and have not specific objectives, even when these are labelled as such. Specific objectives should possess appropriate characteristics that allows for monitoring and evaluating the plan, both in terms of process and results.
d. Most NP/S have no operational plan and few of them have an execution calendar. These were considered positively.
e. Most NP/S have no budget and few have an indication of the allocation of funds for the specific activities.
f. There is no plan for monitoring and evaluation activities though in a few cases both activities are mentioned.
g. Issues relating to sustainability should be further clarified. Is, for example, sustainability only related to funding or does it includes other aspects, such as technical or organisational sustainability. Because of this, the review of this area was postponed until further clarification is available.

3. EUROPLAN National Conferences – Key priorities and way forward

V. Bottarelli (EURORDIS)

The objective of this presentation regards National Conferences on RD held across Europe under the sponsorship of national patient organisations. In total 25 National Conferences have been planned across Europe and several of them have already taken place at the time of the workshop. .

Since its inception in 2008, the EUROPLAN National Conferences (NC) promote and accompany the development and adoption of comprehensive NP/S for RDs, addressing the unmet needs of RD patients and integrating the current European policies, recommendations and legal framework in this field. The NC are organised according to the same philosophy, objectives and format, and they follow a common set of content guidelines, although the content can be adapted to each MS national context. The process followed in the organisation of NC is detailed in Annex 5.

The NC main themes, each covered in a specific workshop, are: 1) methodology and Governance of a NP; 2) Definition and inventorying (information and training); 3) Research on RD; 4) Care: Centres of Expertise and European Reference Networks; 5) Orphan drugs; and 6) Social services for RD.

The discussions during the NC have provided important information regarding some of the MS pending problems and some of the possible solutions within each thematic area. A synthesis is herewith reported comprising three main areas as follows.

A. Areas with enough guidance but additional support might be needed through exchange of best practices across EU Member states

1. Implementation and monitoring of NP

- Exchange of best practices across countries, focusing on specific solutions adopted in other countries

2. Centres of Expertise and Healthcare Pathways

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

3. *European Reference Networks*
   - Identification of ERNs by therapeutic areas
   - Exchange of best practices
   - Capacity building /information activities to support ERN establishment and MS in CE designation role

4. *Registries, Databases*
   - Advisory role to JRC and Exchange of best practices
   - Including in broader context of information flow, eHealth for RD – how to integrate, for example, Electronic Health Records while preserving specific RD needs?

5. *Research on RD*
   - Exchange of best practices (e.g. experiences of specific RD calls or programmes)
   - Sharing roadmap and funding priorities amongst IRDiRC members

B. Areas where there is need for new guidance to be provided by EUCERD (CERD)

1. *Codification of RD*
   - Technical EUCERD Recommendations on use of using OrphaCode alongside existing systems of codification at national level?
   - Exchange of best practices on implementation?

2. *Comprehensive information system for RD*
   - Technical EUCERD Recommendation – common principles

3. *Good practices guidelines*
   - Technical EUCERD Recommendations on Good Practice Guidelines? Principles, methods, gathering of experience at EU level, sharing of work between MS

   Take into account the tools produced by the Rare-Bestpractices project, funded by the EC (www.rarebestpractices.eu) (added by Domenica)

4. *Genetic testing, next generation sequencing*
   - Technical Recommendations /guidance on how to implement and use Next Generation Sequencing technology for RD

5. *Access to treatment*
   - 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

6. *Social Policy*
   - 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 People Living with Rare Diseases
   - Support to Orphanet for indexing the functional consequences of rare diseases with the Orphanet Disability Thesaurus

C. Area for further development

These are the main issues to be considered for further development:

1. There is a need to think to the next phase of National Plans to reach an integrated, comprehensive and long term strategy.
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 continue 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

4. EUROPLAN Debrief Sessions: identifying strengths and areas for support and development

A.E. Gentile (EUROPLAN Coordinating Team), V. Hedley (EJA Team)

This presentation reports on the process of carrying out Debriefing Sessions (DS) following National Conferences on RDs and highlights the strengths and areas in need of support that emerge from these DS across Europe. DS started in February 2013 and, to date, 12 DS have already been held in the following Countries: Slovak Republic (28 February, 2013), Romania (25 May 2013), Finland (21 September 2013), Poland (28 September 2013), Hungary (25 October 2013), Lithuania (14 November 2013), Cyprus (15 November 2013), Luxemburg (20 November 2013), Serbia (7 December 2013), Italy (28 January 2014), Croatia (28 February 2014), Belgium (28 February 2014).

Standard Operating Procedures (SOPs) have been established beforehand, and agreed upon, and are used to guide a standardised approach to hold DS across Countries in Europe. A National Alliance allied with EURORDIS, organises each NC and, at the end of the conference, the DS takes place. Participants to the DS are:

- 1/2 MoH representatives plus key persons involved in elaboration/implementation of NP
- 1/2 EUROPLAN Coordinating Team member/s
- 1 EJA Coordination representative
- 1 EURORDIS Advisor

The process is detailed in Annex 6. Meanwhile, the following is an account of some of the main findings deriving from the DS already processed.

The needs expressed during the DS are categorised approximately as Preliminary and Complementary: the first are generally the issues most prominently discussed during the DS, and tend to be the areas in which support seems most welcome by the national team (and often, appears most feasible to provide); the latter have tended to involve issues which, although often very important, were perhaps not the focus of the DS.

1. Preliminary needs

The area that has obtained the highest number of expressions of needs is the area of Registries with 12/12. That is, every MS expressed the need for support in setting up and maintaining

Registries. In decreasing order we find: CEs (8/12), Social Services (6/12), Research (6/12), Orphacode (5/12), ERNs (5/12), Guidelines (3/12), OMPs (3/12) and other areas follow with smaller numbers.

2. Complementary needs

While for the Area of Registries all MS have been unanimous in giving it the highest priority, the other areas may appear both in Preliminary as well as in the Complementary needs section. Following is, in descending order, the result of the expression of needs in the Complementary category: Lab Quality Control (8/12), Training of Health Professionals (8/12), Helplines (7/12), External Evaluation (7/12), Orphacode (6/12), Guidelines (6/12), Sustainability (6/12), Screening Policies (6/12), Social Services (5/12), and so on.

The DS, with its emerging needs, is the first step in Capacity Building, the goal of which is to increase the quality and quantity of work that an individual/organisation is able to do. It is an on-going evidence driven process of strengthening the abilities of individuals, organizations, and/or systems to perform core functions sustainably, and to continue to improve and develop over time.

The main mandate of WP4 is to develop and carry out Capacity Building activities to meet MS needs, expressed also during the DS. Now a significant number of DS have been realized and the trends identified give us good picture of MS needs.

5. Use of Core Indicators in Member States: study results

R.M. Ferrelli, M. De Santis (*EUROPLAN Coordinating Team*)

A pilot qualitative study on the use of the 21 Core Indicators was conducted in 5 MS: Bulgaria, Croatia, Italy, Romania and Spain. The selection of the Countries was done with purposive sampling, according to activities carried out in former and successful collaboration with the Italian National Centre for Rare Diseases.

The descriptive study was structured in two components:

1. A survey on the use of the indicators in selected EU Member States.
2. An exploratory collection of lessons to take into account for strengthening indicators potential to orient policies for rare diseases

The 21 core indicators were entered in the web-based survey system Survey Monkey (www.surveymonkey.com). The representatives of the Ministries of Health, directly involved in planning, monitoring and evaluating National Plans for Rare Diseases in the five selected Members States, were invited by E-mail to fill-in the questionnaire. Data were analysed by means of descriptive univariate analysis.
The second component of the study included open-answer questions about:

- positive aspects highlighted while using the indicators
- problems faced while using the indicators
- opportunity of integrating the indicators

The survey was carried out in March and April 2014 using a detailed questionnaire to key informants in each country and the results are compiled in Annex 7.

This is a summary of the areas under study and an overview of the results.

a. Areas covered by the survey

1- Background indicators

- Existence of Regulations/Laws, or equivalent official national decisions supporting the establishment and development of RD plan.
- Existence of a RD Advisory Committee

- Permanent and official patients' representation in plan development, monitoring and assessment
- Adoption of the EUCERD definition

2- *Content indicators: CoE & Information*
- Existence of National policy for establishing CoE on RD
- Number of National and Regional Centres of Expertise adhering to the national policy
- Participation of national or regional centres of expertise in European Reference Networks
- NP/NS support to the development/participation in an information system on RD
- Existence of Help lines for RD

3- *Content Indicators: Knowledge, Classification/coding, Registries, Research*
- Existence of a national policy for developing, adapting and implementing clinical practice guidelines
- Type of classification/coding used in the health care system
- Existence of a national policy on Registry and data collection on RD
- Existence of a RD research programmes/projects in the Country
- Participation in European & international research initiative

4- *Content indicators: Therapies & Social Services*
- Number of Orphan Medical Products (OMPs) with a European Union Marketing authorisation and available in the Country (i.e. Priced and reimbursed or directly supplied by the National Health System
- Existence of a governmental system for compassionate use of medicinal products
- Existence of programmes to support in their daily life RD patient organisations

5- *Financial support indicators*
- Existence of a policy/decision to ensure long term funding and/or sustainability of the measures in RD plan/strategy
- Amount of public funds allocated to RD plan/strategy
- Specific public funds allocated to RD research
- Public funds especially allocated for RD research/projects per year since the plan started

b. **Concerning the open answer questions the following positive aspects and difficulties while using the indicators were highlighted:**

1. *Positive aspects while using indicators*
- Collection of important information about NP on RD and easiness to answer
- Excellent opportunity to share knowledge and comparability among countries
- Political usefulness
- Chance to adapt national RD policies to best examples available and recommended at EU level
- Tool to timely follow up and report on national RD activities
- Possibility to follow up the progress
- Focus on relevant issues regarding development and implementation of NP which are common to the 28 EU MSs;
- Harmonisation of monitoring procedures & criteria and assessment of common RD policies in the 28 EU MSs
- Usefulness to capture and describe the situation as far as the general measure adopted by countries are concerned

2. *Difficulties faced while using indicators*
- Quantitative indicators may not reflect qualitative improvements
- Comparing MS different rules and regulations to make clear the different organisation of the countries through short answers can be very difficult

- Indicator on participation in ERNs is difficult to use (ERNs for RD are not yet established).
- Low visibility of what is done for RD patients in programmes to support in their daily life integration (in place in all Countries)
- Difficulty in collecting information from the regional level

In relation to the opportunity of integrating the indicators, the participants to the survey believe that increasing their number would entail a return to the "starting line". However, they did not exclude a possible future integration with EUCERD criteria on centres of expertise for RD and RD reference networks, and they also suggested to include information on newborn screening policies and on the genetic diagnosis and genetic counselling policies.
In conclusion, the pilot study on the usability of the core indicators highlighted their usefulness in giving a snapshot of the main areas of concern for national planning for rare diseases. Moreover, a synthetic representation allows for defining common policies at European level; e.g. the situation about RD coding, while showing heterogeneity in the system adopted for coding rare diseases in the Country, allows for identifying a system that is likely to be used in most, hopefully all, of the MS.
The core indicators represent an excellent opportunity to share knowledge and comparability among MS. However, it is important to acknowledge the strengths and weaknesses of the single tools and be aware that quantitative indicators may not reflect qualitative substantial aspects of the issue they are measuring, that may be studied with appropriate and different tools, according to the objectives that have been set for the matter under study.

D. On-site Qualitative Questionnaire to Workshop Participants

A Questionnaire to be delivered on-site was constructed in order to collect qualitative data useful to the process of translating MS expressed and derived needs into a capacity building operational plan, tackling needs through a set of prioritised actions.

Capacity building can be undertaken using different means, methods and instruments and each choice entails the need for different resources. Therefore, it is important not only to prioritise needs but, at the same time, it is essential to get a sense of which available actions meet the priority needs in the most effective and efficient way. This avoids unnecessary activities and wastes. In light of this, it was deemed necessary to further investigate the issue in a qualitative manner delivering the questionnaire to all participants to the Workshop coming from the majority of European MS.

The Questionnaire (Annex 8) comprised the priority areas (Registries, Centres of Expertise, European Reference Networks, Guidelines, Research, Social services, Orphacode) identified in the expressed MS needs, which derive from the DS.

E. General discussion

Chairpersons: D. Taruscio (*EUROPLAN Leader)*

Following the compilation of the questionnaires, it was felt more productive to undertake a discussion in plenary and therefore the small group work, given also the time limitation, was annulled. The information shared by the participants during the Tour de Table is summarized in Annex 8.

While on-site, a preliminary extemporaneous analysis of the answers evidenced some trends, confirming the main topics emerged during the DS and providing some general indication of possible actions to be taken (e.g. training activities on specific areas such as registries; implementation of coding).

The Workshop closed at 18:00