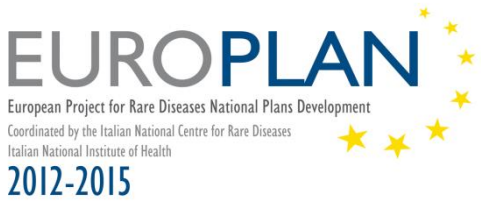

EUROPLAN NATIONAL CONFERENCES 2012-2015

CONTENT GUIDELINES FOR WORKSHOP 2 / THEME 2

DEFINITION, CODIFICATION,

INFORMATION AND TRAINING

Table of Contents

A. How to read and use these Content Guidelines

The EUROPLAN Content Guidelines cover 6 main Themes. For each Theme, these Content Guidelines cover all the core topics to be addressed in the Workshop dedicated to that Theme. These Guidelines include:

1st column – RESOURCES	**2nd column - TOPICS FOR DISCUSSION**
This column includes the background documents and relevant material that should be referred to in preparation for the discussion. They mainly include: • Specific articles of the EU Council Recommendation on an action in the field of rare diseases; • Specific items of the Commission Communication on Rare Diseases: Europe's challenges; • EUCERD Core Recommendations on Rare Disease Patient Registration and Data Collection; • EURORDIS-NORD-CORD Joint Declaration of 10 Key Principles for Rare Disease Patient Registries (2012); • Specific recommendations from the "EUROPLAN Recommendations"; • Extracts from the Synthesis Report of the 15 EUROPLAN National Conferences held in 2010; • Specific EUCERD Core Indicators; • Specific EUROPLAN Indicators. **NB: Full documents of the sources referenced above can be found in Section C**	**The topics for discussion are questions formulated to stimulate the discussion within the Workshop**. The conference organisers, with the help of their Advisor, will **select those questions that are relevant for the discussion in their countries**. As such, **not all listed questions need to be addressed in a mandatory way**. They rather represent a "menu" from which to pick the questions that address the most relevant topics in the country, having considered the level of advancement of the national policy on rare diseases in the country.

B. Guidelines for discussion for Workshop 2 – Theme: Definition, Codification, Information and Training

Important: HOW TO READ and USE THESE GUIDELINES.
For each Theme, the "Guidelines for discussion" cover all the core topics to be addressed by the Workshop dedicated to that Theme. These Guidelines include:

- **Resources** (1st column below): **background documents and relevant material that should be referred to and looked up in preparation of the discussion**. They mainly include the relevant parts or articles from the Council Recommendation and the EUROPLAN Recommendations, as well as extracts from the Final Report of the 2012 EUROPLAN National Conferences. **Please note that at the end of the section 1. Guidelines for Discussion, sections 2 to 4 and the Annex provide the referenced resources in full.** Moreover, **other resources that may be relevant in order to enrich the discussion of the Workshop are also referred to in this column**.
- **Topics for discussion** (2nd column): the topics for discussion are questions formulated to stimulate the discussion within the Workshop. The conference organisers, with the help of their Advisor, will select those topics that are more relevant for the discussion in their countries. As such, **not all listed questions need to be addressed in a mandatory way**. They rather represent a "menu" from which to pick up the questions that address the most stringent and relevant topics in the country, having considered the level of advancement of the national policy (policies) on rare diseases in the country.

RESOURCES	TOPICS for DISCUSSION
B.1 Definition of RD	
Council Recommendation on RD 2. Use for the purposes of Community-level policy work a common definition of rare disease as a disease affecting no more than 5 per 10 000 persons. **Commission Communication on RD** 4.1. The existing definition of rare diseases in the EU was adopted by the Community action programme on rare diseases 1999-2003 as those diseases presenting a prevalence not more than 5 per 10 000 persons in the European Union. The same definition is set out in Regulation (EC) 141/2000 and, accordingly used by the European Commission for the designation of orphan drugs. The EU will maintain the current definition. A more refined definition taking into account both prevalence and incidence will be developed using the Health Programme resources and taking into	• Is the **definition proposed by the Council Recommendation** (see left column) based on the definition of the Orphan Drugs Regulation 141/2000/EC, adopted in your country? What is the status of this definition? – When the EU definition is not in use, how are the characteristics of rare diseases defined? Please also assess the readiness to adopt the EU definition and the possible obstacles to its systematic use in the country. – When used but not recognised as "official definition", please discuss whether this status should be changed and officially acknowledged. • What mechanisms could be put in place to include diseases which do not strictly fall within the definition in National Plans for RD? How to ensure that **exceptions**

account the international dimension of the problem. **Final Report of EUROPLAN Conferences** (Area 2, page 35) • "It is generally agreed that one European definition is useful for facilitating cooperation and community actions. Conferences' participants broadly expressed the necessity and expectation to (continue to) use the European definition, especially in view of laying down legally binding legislation, as required by European policy documents."	to the definition are redressed or otherwise explained? *e.g. some rare diseases are at the "frontier" of the definition, e.g. thalassemia in some Mediterranean countries is not that rare, but it is in some other European countries, and as a result, Mediterranean countries may wish to include it within their NP for the purposes of synergy with other countries.* The national government needs to make it clear why a specific disorder that is not rare in that country is to be included in the National Plan for Rare Diseases and why a Government decides to allocate specific funding for those diseases. These exceptions should be explained in the Plan.
B.2 Codification of RD and traceability in the national health system	
Council Recommendation 2. Aim to ensure that rare diseases are adequately coded and traceable in all health information systems, encouraging an adequate recognition of the disease in the national healthcare and reimbursement systems based on the ICD while respecting national procedures. **Commission Communication on RD** 4.2. Classification and codification of rare diseases The international reference for classification of diseases and conditions is the International Classification of Diseases (ICD), coordinated by the World Health Organisation (WHO). The Commission will lead work with regard to rare diseases within the process of revising the existing ICD (International Classification of Diseases) in order to ensure a better codification and classification of rare diseases. For this purpose a working group on Classification and Codification of rare diseases will be created by the Commission. This working group could be appointed as Advisory Working Group by the WHO in the current ICD revision process. **EUROPLAN Recommendations** R 2.2 The use of a common EU inventory of rare diseases (Orphanet) is promoted in the national health care services and collaboration is carried out to keep it updated. R 2.3 Coding of rare diseases is promoted, encouraging their traceability in the national health system.	• Which are the most used **classification systems** in your country (ICD9, ICD10, SNOMED, ICDO for rare cancers, MIM, Orpha Code…) and for which purposes are they used: e.g. surveillance, reimbursement, provision of social support, etc.? • Is your country part of Orphanet – if so, would you like it to be? Why and why not? • The revision of the International Classification of Diseases (ICD) leading to 'ICD11' is planned to become operative in 2015 and will provide a tool to trace rare diseases in health information systems. Before ICD 11 comes into force, the Orphanet classification (Orpha Code) provides a code which is largely in line with the future ICD11. What needs to be done in your country for a swift adoption of the WHO-led system, the ICD-11, when ready in 2015? • What strategy could be put in place to ensure that all RD are coded and traceable in the health information and social systems? What initiatives are promoted at national level for the integrated use of administrative, demographic and health care data sources to improve the management of RD? • What procedure, based on the ICD system, could be put in place to facilitate the smooth recognition of a disease as 'rare disease' in the national healthcare and social systems and thus allow people living with RD to access to both healthcare

R 2.4 Cross-referencing rare diseases is carried out across the different classification systems in use in the country, ensuring coordination and coherence with European initiatives, such as reference to the Orpha-code. R 2.5 Collaboration with the ICD10 revision process is ensured and ICD-11 is adopted as soon as possible. Examples from the EUROPLAN Recommendations (page 30): • IMPROVE CODING RARE DISEASES ON EUROPEAN LEVEL: the Working Group (WG) of the Rare Disease Task Force on Rare Diseases • THE ITALIAN WORKING GROUP FOR CODING AND CLASSIFICATION OF RARE DISEASES **Final Report of EUROPLAN I National Conferences** (Area 2, pag 36) – "A change in attitude vis-à-vis disease inventorying is demanded, as the foundation for ICD11 acceptance. This is why it is recommended, while awaiting the release of ICD11, to introduce existing coding systems into clinical practice: Orpha Code, preferably, as ICD10 is deemed not satisfactory as for the coverage of RDs, or jointly, as a combined system. Such an early introduction would also help to promote a higher level of awareness and knowledge of RDs by healthcare professionals (in addition to the more obvious inventorying purposes)."	and social services? • Is the Orpha Code already used in the hospitals and for data collection?
B.3 Registries and databases	
Council Recommendation on RD 5.Consider supporting at all appropriate levels, including the Community level, on the one hand, specific disease information networks and, on the other hand, for epidemiological purposes, registries and databases, whilst being aware of an independent governance." **EUROPLAN Recommendations** R 2.7 Initiatives are promoted at national level for the integrated use of administrative, demographic and health care data sources to improve the	• Is there a policy for RD data collection and RD patient registration laid down in the National Plan or Strategy for RD? • Are official lists of RD compiled in your country? For what purpose(s)? • Is there an official governmental RD registry? And/or specific RD databases e.g. held by Centres of Expertise? • What legal framework or mechanisms do oversee the interactions among delocalised registries and central registries or databases, when the former are supposed to feed into a central registry or database? What mechanisms are in

management of rare diseases.
R 2.8 International, national and regional registries for specific rare diseases or groups of rare diseases are promoted and supported for research and public health purposes, including those held by academic researchers.
R 2.9 Collection and sharing of data from any valid sources, including Centres of Expertise, and their availability for public health purposes is promoted by public health authorities, in compliance with national laws.
R 2.10 Participation of existing national registries in European/International registries is fostered.
R 2.11 Instruments are identified for combining EU and national funding for registries.

EUCERD Recommendations on RD Patient Registration and Data Collection

http://www.eucerd.eu/?page_id=13

1. RD patient registries and data collections need to be internationally interoperable as much as possible and the procedures to collect and exchange data need to be harmonised and consistent, to allow pooling of data when it is necessary to reach sufficient statistically significant numbers for clinical research and public health purposes.

1.1 They should use international standards and nomenclature to code the tentative or final RD diagnosis. Either the OMIM code or the Orpha codes are recommended alongside any other coding system in operation in the MS health systems, such as ICD and SNOMED-CT, with a view to establishing a common semantic approach.

1.2 There should be adoption of a minimum common data set across RD that registries should collect, in collaboration with global initiatives, to allow the establishment of national and/or European RD population registries, which have the potential to collect data on all RD patients.

1.3 A minimum common data set should be defined, and supported with a semantic approach and Standard Operating Procedures. Interoperability (via means of mapping) of registry specific data sets towards this common data set should enable comparison across all RD and internationally.

1.4 For disease-specific registries, appropriate core data sets specific to the diseases or disease groups should be adopted. In the future, such disease-specific registries could fall under the remit of RD ERNs. Every effort should be made to incorporate

place or should be put in place to avoid overlaps?

- What measures do ensure the **interoperability** of different RD registries and the harmonisation of procedures to collect data and thus facilitate pooling of data?
- How to stimulate the harmonisation of procedures and technical tools, in particular the development of minimum data sets, for both registries and biorepositories?
- Are RD patient registries using international standards and nomenclature to code the RD diagnosis, even if tentative?
- What rules do ensure that **quality standards** of registries are consistently high?
- What system could ensure that **data directly reported by patients** are included along with data reported by clinicians?
- Please assess and discuss the **participation and involvement of patients** in rare disease patient registries, including establishing registries; defining content and purposes of the registries; resolving ethical and legal issues; authorising access and utilisation of data; creating partnerships with health professionals and industry representatives; contributing to the selection of data items collected; helping recruit patients for participation into the registry; preparing specific information for patients to be registered prior to their consent; motivating health professionals to input data, and directly entering data.
- What measures could promote the involvement of patients as well as other stakeholders in the design, analysis and governance of RD registries?
- Is the National Plan or Strategy on RD also facilitating **access and sharing of data** to control how data is shared and published in the public domain?
- What other measures do ensure that RD patient registries adhere to good practice guidelines in the field, including European and international ones, notably on consenting patients for participation in registries, regular feedback to registered patients and clinical teams, etc.?
- How to ensure, through appropriate funding mechanisms, the **long-term sustainability** of registries and databases? Do these registries and programmes receive government support? What space is there for private public partnerships

current disease-specific registry initiatives where quality can be assured.

[…]

2. All sources of data should be considered as sources of information for RD registries and data collections, to speed up the acquisition of knowledge and the development of clinical research.

[…]

2.4 Collection of data on RD should be delineated in the National RD plan/strategy.

2.5 A system to allow the collection of data directly reported by patients should be included along with systems for data reported by clinicians.

[…]

3. Collected data should be utilised for public health and research purposes.

[…]

3.4 Pooling of data across data collections and other resources, including internationally, should be encouraged to reach a critical mass for data analysis. According to the governance/oversight criteria, data should be made accessible to groups with legitimate questions such as researchers and policy/decision makers.

3.5 Access and sharing of data should be defined to control how data is shared and published in the public domain and this should be facilitated through the national RD plan/strategy.

4. Patient registries and data collections should adhere to good practice guidelines in the field. Specific to the current and future specificities of RD registries:

4.1 Involvement of stakeholders such as patients, policymakers, researchers and clinicians (and industry, where appropriate) in the design, analysis and governance of registries is important to address the complexity and scarcity of knowledge on RD.

[…]

6. Patient registries and data collections should be sustainable for the foreseeable timespan of the registries' utility.

Final Report of EUROPLAN I Conferences
(Area 2, page 37)

- "In some countries an explicit call has been made for laying down the legal

to fund these infrastructures?

- Do RD patient registries usually envisage exit strategies in their work plans? What provisions are necessary to make sure that this occurs on a regular basis?
- Discussion are ongoing on the creation of a **European Platform for Rare Disease Registration**, supported by the European Commission and aimed to provide common services and tools for the existing (and future) rare diseases registries in the European Union. What contribution could you country provide? How a European Platform may help optimise national resources devoted to rare disease regisitration?

ADDITIONAL QUESTIONS on "Registries" from WORKSHOP 3. Research on RD

- What initiatives and incentives are or should be in place to bring clinicians to actively participate in the collection of data?
- How to motivate the sharing and open access to pre-competitive resources such as databases, biobanks or knowledge bases?
- How to engage in **international initiatives** such as those promoted by the **IRDiRC** (the International Research Consortium for Rare Disease Research) in favour of harmonisation and interoperability of RD registries and thus promoting the creation and functioning of registries with larger geographical scope?
- Please explore the feasibility of a common central resource or platform for creating or reconfiguring registries and describing the content of existing registries and databases with the potential to collect data on all RD.

framework necessary to define competences and establish the coordination mechanisms among public institutions and administrations and/or private centres, especially when it is required that delocalised registries feed into central registries or databases. Such a legal structure is clearly important for countries where the healthcare system is decentralised."

- "In order to ensure registries sustainability, another challenge to face is the availability of resources. One first recommended step is to optimise resources by reducing fragmentation and overlapping. Focus should be on a better use and better information on existing knowledge and information sources. ORPHANET could be instrumental in doing this. Optimisation of resources may include adopting mechanisms to extract the already established information hidden under the global (all diseases) information.
- Government resources are generally considered indispensable by all Conferences' participants to sustain the functioning of RD registries, even though in some countries this is current practice (Germany, Sweden, Italy, for instance) and in others this is not happening (Greece).
- In two projects run in Bulgaria, the involvement of all stakeholders proved to be key to the success and to the long-term duration of RD registries. This is especially recommended as it offers diversification of funding sources (government, academia, industry, patient organisations,...).
- The differentiation of registries' outcomes also emerged as a decisive factor: as far as possible, registries should be "multi-purposes", i.e. used for public health planning, research and international cooperation...".

EURORDIS-NORD-CORD Joint Declaration 10 Key Principles for RD Patient Registries
(Full text here, with 10 Key Principles below explained in more detail:
http://download.eurordis.org/documents/pdf/EURORDIS_NORD_CORD_JointDec_Registries_FINAL.pdf)

1. Patient Registries should be recognised as a global priority in the field of Rare Diseases.
2. Rare Disease Patient Registries should encompass the widest geographic scope possible.
3. Rare Disease Patient Registries should be centred on a disease or group of diseases

rather than a therapeutic intervention. 4. Interoperability and harmonization between Rare Disease Patient Registries should be consistently pursued. 5. A minimum set of Common Data Elements should be consistently used in all Rare Disease Patient Registries. 6. Rare Disease Patient Registries data should be linked with corresponding biobank data. 7. Rare Disease Patient Registries should include data directly reported by patients along with data reported by healthcare professionals. 8. Public-Private Partnerships should be encouraged to ensure sustainability of Rare Disease Patient Registries. 9. Patients should be equally involved with other stakeholders in the governance of Rare Disease Patient Registries. 10. Rare Disease Patient Registries should serve as key instruments for building and empowering patient communities.	
B.4 Information on available care for RDs in general, for different audiences	
Council Recommendation on RD 5.Consider supporting at all appropriate levels, including the Community level, on the one hand, specific disease information networks and, on the other hand, for epidemiological purposes, registries and databases, whilst being aware of an independent governance." **Commission Communication on RD** 4.4. Disease information networks Priorities for action regarding the existing (or future) specific disease information networks are: – to guarantee the exchange of information via existing European information networks; – to promote better classification of particular diseases; – to develop strategies and mechanisms for exchanging information between stakeholders; – to develop comparable epidemiological data at EU level; – and to support an exchange of best practices and develop measures for patient groups.	• What are the existing information sources in your country? Are they of good quality? Please consider whether: – there is a national official website for RD in the country; – there are help lines, whether they are known to the public (*see also next topic*); – there are initiatives of centres of expertise and/or patient organisations or programmes to stimulate the development of information and educational material for patients or specific publics (teachers, social workers, etc.); – if existing resources at European level, ORPHANET and EURORDIS, are used: information on diseases, specialised centres and patient groups, ongoing research projects, clinical trials – there are initiatives to raise awareness on RD such as the Rare Disease Day.

EUROPLAN Recommendations R 5.1 The use of international global information websites and data repositories for rare diseases is promoted. R 5.2 Access to knowledge repositories and to expert advice for health professionals is established. **Final Report of EUROPLAN Conferences** (Area 5, page 49) – "Access to both national and international information websites and data repositories is widely promoted and their financial support encouraged. The unique role of ORPHANET has been highlighted everywhere, as well as the necessity to increase knowledge and awareness among the general public on this and other data repositories, which are crucial for patients and families. – Government-supported campaigns have been mentioned as a solution (e.g. France, Bulgaria) to raise public awareness on RDs in general, and to make information services and repositories available at national level better known by the public at large. – National awareness-raising initiatives are called for in many countries. Apart from the consolidated Rare Disease Day, proposals include organisation and participation to events, seminars and conferences, better media coverage on generic press, targeted articles on specialised press and medical journals, dissemination of information through professional societies and patients associations, informative brochures and leaflets to distribute locally (centres of primary care, pharmacies, etc.). – Regarding the information sources available to both patients and professionals, the information provided is not homogeneous (it varies depending on the region, on the disease or the group of diseases) and that quality is not always validated. Proposals include mapping out available information country-wide; the introduction of validation/verification systems; identifying control mechanisms for information provided on the web. It could be the responsibility of the committee in charge of the NP to assess the evaluation needs and the standardisation mechanisms of information sources."	• How are **specific disease** (or group of diseases) **information networks** organised? • How is information exchanged amongst stakeholders and what level of involvement do they have? • How are their European/international activities promoted and supported? • How are they financially supported? Do they receive public funding? • Please discuss about **mapping out information resources** on rare diseases available in your country, as well as the possibility of introducing **validation/verification systems** to ensure that the quality of that information is consistently good.

B.5 Help Lines	
EUROPLAN Recommendations R 6.8 Interactive information and support services for patients are promoted (such as help lines, e-tools etc). **Commission Communication on RD** 5.2. Access to specialised social services Centres of expertise may also have an essential role in developing or facilitating specialised social services which will improve the quality of life of people living with a rare disease. Help Lines, Respite care services and Therapeutic Recreation Programmes, have been supported and need to be sustainable to pursue their goals: awareness-raising, exchange of best practices and standards, pooling resources using Health Programme and the Disability Action Plans. **Final Report of EUROPLAN I Conferences** (Area 6, page 54) – "Help lines as well as other interactive information and support services for patients should be included in the provisions of a NP on RD. – National help lines should be created which are based on toll free numbers and linked to the European free toll number, when the latter is set up. – Setting up quality help lines is essential for the patients benefiting from the service and for the credibility of the service itself. Quality criteria for help lines are established in the framework and in the follow-up of the EC funded Rapsody project, led by EURORDIS. It was recalled that, amongst other, an important requirement for creating a help line is the prior and regular monitoring of the information needs of patients and their families. – It was suggested that, where this is not the case yet, the country's umbrella organisation such the country's National Alliance of RD associations may be useful in consolidating help line services and perhaps acting as a signposting service."	• What kind of **help lines** (all diseases) exist in your country to assist RD patients and healthcare professionals? • How to develop or consolidate existing patient-run help line services for RD? • How to improve the service offered? How to improve their visibility esp. for patients? • How are help lines financed? By private initiative or patient associations? Is there any government funding? • How to ensure their long-term sustainability? • What national measures can be promoted to establish the **116 European number for RD Help Lines**? With the "116 number" the European Commission seeks to identify services of social value in Europe that could benefit from single European free phone numbers starting with 116. *NB:* An application has been made by EURORDIS to the European Commission, but it will then require the support of at least 14 Member States. Once the 116 number has been assigned by the Commission, national procedures are necessary to make the free number work in each EU country. Please discuss about how to support at national level the adoption of the official EU-wide number for RD Help Lines.

B.6 Training healthcare professionals to recognise and code RD	
EUROPLAN Recommendations R 2.6 Healthcare professionals are appropriately trained in recognising and coding rare diseases. **Final Report of EUROPLAN I Conferences** (Area 2, page 36) – "In general, healthcare professionals do not have sufficient awareness of RDs in general, of the ORPHANET classification, nor are they aware of the lack of a proper codification of RDs within the ICD10. It was stressed in more than one country (e.g. Bulgaria, Greece, Spain) that the problem lies with the lack of familiarity with RDs by general practitioners or family doctors. – The introduction and the use of appropriate IT tools may support training efforts. An example is the protocol developed in Spain that allows family doctors to check if a disease is rare or not and provide support to identify criteria for decision-making. – Undergraduate and postgraduate courses are proposed for improving RD knowledge among professionals, which include opportunities to familiarise with the coding systems."	• What level of awareness and knowledge do healthcare professionals have of the RD classification and codification? What can be done to improve it? • Due to the changes in the coding of RDs, which will appear in ICD11, what specific actions can be envisaged to prepare and train healthcare professionals in view of the introduction of ICD11?
B.7 Training healthcare professionals	
EUROPLAN Recommendations R 5.4 The curriculum of the medical degree course includes an education package on rare diseases and on the relevant, specific provisions in the healthcare services. R 5.5 Training of medical doctors (general practitioners and specialists), scientists and new healthcare professionals in the field of rare diseases is supported. R 5.6 Continuing education programmes on rare diseases are made available for health professionals. **Final Report of EUROPLAN Conferences** (Area 5, pages 49-51) – "Largely inspired by the two-hour module included in the French undergraduate	• How are healthcare professionals trained in your country? In particular: • Please discuss about the possibility of introducing, if not existing, an ad hoc **educational package on rare diseases in the curriculum of the medical degree course** and on relevant provisions in the healthcare systems. • Please discuss in particular of the development of training modules aiming to provide comprehensive care to people affected by RDs and including specific protocols for paediatricians to recognise relevant symptoms of RDs • Please discuss about the possibility of introducing **specialist training** on rare

medical courses, the inclusion of an educational package in the curriculum of the medical degree course is demanded to be adopted as quickly as possible in other European countries. – In two countries, Spain and Greece, there was a specific demand for creating a specialisation in the medical career in Clinical Genetics, which is currently inexistent. – Specific proposals include innovative trainings using virtual universities (see www.edubolirare.ro), online trainings and virtual channels. Virtual programmes would have to be approved by expert centres and other institutions involved in RDs. A public policy support and adequate IT platforms and tools are, of course, preliminary steps to the implementation of such training modalities. – Systems to evaluate the quality and effectiveness of trainings should be set up, supported by outcome indicators. – Trainings of professionals should be systematically introduced and organised. Specifically targeted trainings should be provided, with different degrees of knowledge, depending on whether addressed to family doctors or specialists. – In particular, it is widely recognised that a general training, broadly encompassing all disease areas, should be assured for general practitioners and paediatricians. This is necessary to create awareness on possible rare diagnoses and to train them to deal on a day-to-day basis with rare patients. – Postgraduate training is required to bring a more in-depth knowledge of RDs to specialists, with an additional component of management, so to move from the knowledge of the disease to the knowledge of the patient. – Training is necessary for paramedical professionals as well. Centres of Expertise should be the elective places where such training is provided, according to the outcomes of many Conferences, or at least where it is coordinated. Sharing experiences and resources by creating exchanges among Centres of Expertise would help to maximise the efforts. – Training opportunities need to be developed for professional figures, such as the assistant of persons with severe disabilities, or other professional profiles whose role contributes to improve the quality of life of people living with RDs. – Information and training resources should be made available on a widely accessible directory possibly validated and/or managed by a competent body. They could be supported by the ORPHANET portal. – Adequate competence on RDs (general, for GPs, and more targeted for	diseases for medical doctors in fields relevant to diagnosis of RDs (e.g. genetics, oncology, immunology, neurology, paediatrics), including post-graduate trainings. • Please discuss about the training opportunities that need to be provided to **paramedical specialists** and to professional figures, such as e.g. people assisting patients with severe disabilities. • Please discuss about measures and incentives for medical training of **young doctors and scientists** in the field of RD. • What measures can be taken to ensure adequate **lifelong training** of medical doctors and other healthcare professionals, including paramedical specialists? • Please discuss of supporting measures and tools such as virtual universities, online training programmes or channels, etc. Specifically, how could such IT infrastructures and programmes be supported?

specialists) should be achieved by guaranteeing lifelong training of medical doctors and other healthcare professionals, including paramedical specialists. Continuing education initiatives are essential for primary care providers. Continuous training modules should aim at providing comprehensive care to people affected by RDs and should include specific protocols for paediatricians to recognise relevant symptoms of RDs. – Cooperation with patient organisations is desirable. This involves encouraging cooperation and information exchanges between health professionals and patient associations, so to enhance patient-doctor communication (Spain, Italy)."	

C. Background Documents

C.1 Council Recommendation of 8 June 2009 on an action in the field of rare diseases (2009/C 151/02)

Whereas
[…]
(4) Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products provides that a medicinal product shall be designated as an 'orphan medicinal product' when intended for the diagnosis, prevention or treatment of a life-threatening or chronically debilitating condition affecting not more than 5 in 10 000 persons in the Community when the application is made.
[…]
(10) According to the Orphanet database, of the thousands of known rare diseases for which a clinical identification is possible, only 250 of them have a code in the existing International Classification of Diseases (ICD) (10th version). An appropriate classification and codification of all rare diseases is necessary in order to give them the necessary visibility and recognition in national health systems.
(11) In 2007 the World Health Organisation (WHO) launched the process of revision of the 10th version of the ICD in order to adopt the new, 11th version of this classification at the World Health Assembly in 2014. The WHO has appointed the Chair of the EU Rare Diseases Task Force as the Chair of the Topic Advisory Group on Rare Diseases in order to contribute to this process of revision, providing proposals for codification and classification of rare diseases.
(12) The implementation of a common identification of rare diseases by all the Member States would strongly reinforce the contribution of the EU in this topic advisory group and would facilitate cooperation at Community level in the field of rare diseases.

(The Council of the EU) hereby recommends that Member States:
[…]
"II. ADEQUATE DEFINITION, CODIFICATION AND INVENTORYING OF RARE DISEASES

2. Use for the purposes of Community-level policy work a common definition of rare disease as a disease affecting no more than 5 per 10 000 persons.

3. Aim to ensure that rare diseases are adequately coded and traceable in all health information systems, encouraging an adequate recognition of the disease in the national healthcare and reimbursement systems based on the ICD while respecting national procedures.

4. Contribute actively to the development of the EU easily accessible and dynamic inventory of rare diseases based on the Orphanet network and other existing networks as referred to in the Commission Communication on rare diseases.

5. Consider supporting at all appropriate levels, including the Community level, on the one hand, specific disease information networks and, on the other hand, for epidemiological purposes, registries and databases, whilst being aware of an independent governance."

http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF

C.2 Commission Communication on Rare Diseases: Europe's challenges COM(2008)679

("Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's challenges", 11/11/2008, COM(2008)679)

3.1. Improving Recognition and Visibility on Rare Diseases

The key to improving overall strategies for rare diseases is to ensure that they are recognised, so that all the other linked actions can follow appropriately. To improve diagnosis and care in the field of rare diseases, appropriate identification needs to be accompanied by accurate information, provided and disseminated in inventory and repertory formats adapted to the needs of professionals and of affected persons. This will contribute to tackling some of the main causes of neglecting the issue of rare diseases. The Commission therefore aims to put in place a thorough coding and classification system at European level, which will provide the framework for better sharing knowledge and understanding rare diseases as a scientific and public health issue across the EU.

[…]

4.2. Classification and codification of rare diseases

The international reference for classification of diseases and conditions is the International Classification of Diseases (ICD), coordinated by the World Health Organisation (WHO). The Commission will lead work with regard to rare diseases within the process of revising the existing ICD (International Classification of Diseases) in order to ensure a better codification and classification of rare diseases. For this purpose a working group on Classification and Codification of rare diseases will be created by the Commission. This working group could be appointed as Advisory Working Group by the WHO in the current ICD revision process.

4.3. Dissemination of knowledge and information on rare diseases

One key element for improving diagnosis and care in the field of rare diseases is to provide and disseminate accurate information in a format adapted to the needs of professionals and of affected persons. The establishment of an EU dynamic inventory of rare diseases will contribute to tackle some of the main causes of neglecting the issue of rare diseases including the ignorance of which diseases are rare. The Commission will ensure that this information continues to be available at European level, building in particular on the Orphanet5 database, supported through Community programmes.

4.4. Disease information networks

Priorities for action regarding the existing (or future) specific disease information networks are:
– to guarantee the exchange of information via existing European information networks;
– to promote better classification of particular diseases;

– to develop strategies and mechanisms for exchanging information between stakeholders;
– to develop comparable epidemiological data at EU level;
– and to support an exchange of best practices and develop measures for patient groups.

[…]

5.2. Access to specialised social services

Centres of expertise may also have an essential role in developing or facilitating specialised social services which will improve the quality of life of people living with a rare disease. Help Lines, Respite care services and Therapeutic Recreation Programmes, have been supported and need to be sustainable to pursue their goals: awareness-raising, exchange of best practices and standards, pooling resources using Health Programme and the Disability Action Plans.

5.7. e-Health

eHealth can contribute in a number of different ways to this area, in particular through:

– Electronic online-services developed by Orphanet and by other EU funded projects, are a clear demonstration of how Information and Communication Technology (ICT) can contribute to putting patients in contact with other patients and developing patient communities, to sharing databases between research groups, to collecting data for clinical research, to registering patients willing to participate in clinical research, and to submitting cases to experts which improve the quality of diagnoses and treatment;

[…]

http://ec.europa.eu/health/ph_threats/non_com/docs/rare_com_en.pdf

C.3 EUCERD Core Recommendations on Rare Disease Patient Registration and Data Collection

http://www.eucerd.eu/wp-content/uploads/2013/06/EUCERD_Recommendations_RDRegistryDataCollection_adopted.pdf

1. RD patient registries and data collections need to be internationally interoperable as much as possible and the procedures to collect and exchange data need to be harmonised and consistent, to allow pooling of data when it is necessary to reach sufficient statistically significant numbers for clinical research and public health purposes.

1.1 They should use international standards and nomenclature to code the tentative or final RD diagnosis. Either the OMIM code or the Orpha codes are recommended alongside any other coding system in operation in the MS health systems, such as ICD and SNOMED-CT, with a view to establishing a common semantic approach.

1.2 There should be adoption of a minimum common data set across RD that registries should collect, in collaboration with global initiatives, to allow the establishment of national and/or European RD population registries, which have the potential to collect data on all RD patients.

1.3 A minimum common data set should be defined, and supported with a semantic approach and Standard Operating Procedures. Interoperability (via means of mapping) of registry specific data sets towards this common data set should enable comparison across all RD and internationally.

1.4 For disease-specific registries, appropriate core data sets specific to the diseases or disease groups should be adopted. In the future, such disease-specific registries could fall under the remit of RD ERNs. Every effort should be made to incorporate current disease-specific registry initiatives where quality can be assured.

1.5 To avoid duplication and to support Cross-Border Healthcare, the possible benefits of using a global or European RD patient identifier (possibly incorporating the current health identifier) should be investigated to provide a way to link information, samples and research data, and to ensure a quick and secure means of data sharing and protection.

1.6 For countries with regional organisation of healthcare, where multiple registries exist, overlap and duplication between the regional and national registries, should be avoided.

2. All sources of data should be considered as sources of information for RD registries and data collections, to speed up the acquisition of knowledge and the development of clinical research.

2.1 As with all registries, registries for RD should establish clear purposes and objectives of the data collection: the type of data collection should be suited to the need, and the data captured should be appropriate to the proposed use of the data, both in terms of scope and level of detail.

2.2 RD Centres of Expertise, where they exist, should contribute to a registry(ies). Other experts in the field should also contribute to the registry(ies).

2.3 (Electronic) health records from any sector of healthcare delivery are a valuable source for core data collection. Automatic data acquisition from these sources should be envisaged to ease the data collection process.

2.4 Collection of data on RD should be delineated in the National RD plan/strategy.

2.5 A system to allow the collection of data directly reported by patients should be included along with systems for data reported by clinicians.

3. Collected data should be utilised for public health and research purposes.

3.1 RD data collected should be used to support policy development at local, regional, national and international level.

3.2 RD data collected should, where possible, facilitate clinical and epidemiological research and the monitoring of care provision and therapeutic interventions, including off-label use of approved drugs and existing medications.

3.3 RD data collected should, where possible, be used to provide information for multi-centre and multi-national clinical trial feasibility studies.

3.4 Pooling of data across data collections and other resources, including internationally, should be encouraged to reach a critical mass for data analysis. According to the governance/oversight criteria, data should be made accessible to groups with legitimate questions such as researchers and policy/decision makers.

3.5 Access and sharing of data should be defined to control how data is shared and published in the public domain and this should be facilitated through the national RD plan/strategy.

4. Patient registries and data collections should adhere to good practice guidelines in the field.

Specific to the current and future specificities of RD registries:

4.1 Involvement of stakeholders such as patients, policymakers, researchers and clinicians (and industry, where appropriate) in the design, analysis and governance of registries is important to address the complexity and scarcity of knowledge on RD.

4.2 Representatives of all stakeholders should be invited to provide best possible expert support through an advisory board or committee to ensure appropriate information flow and knowledge exchange into and from the registry, and they should define a sustainability and exit strategy for the registry. Where appropriate, representatives from industry should also provide input.

4.3 This multi-stakeholder model for registry governance should apply not only at a national level but also at the European level and/or pan-European Platform repository of RD registries.

4.4 The process for consenting patients for participation in a RD registry should take into account the wider European and international context to ensure that patients are well informed of this dimension and the consent process is in line with the legal requirements at European and International level.

4.5 Patients already in a RD registry may be required to go through an additional consenting step to ensure compatibility with such systems.

4.6 RD registries should have a system to provide regular feedback to registered patients and their clinical teams, recognising their specific role in the success of registries in this field.

5. Existing and future patient registries and data collections should be adaptable to serve regulatory purposes, where required.

5.1 For the monitoring of therapeutic interventions for RD, a strategy between industry, academia and regulators should be agreed to ensure that data collection is expanded as necessary, and in time embedded in disease-specific registries to serve, for example, the requirements for post-marketing surveillance, and to support development of new therapies. Data access needs to be compliant with agreed guidelines established by the registry.

5.2 As quality assurance is crucial, it is a priority for existing RD registries to explore their capacity to adapt to collect data for regulatory purposes.

5.3 There should be an early dialogue on the type of registry required (and what data is required for regulatory purposes), and/or whether a registry exists for the condition targeted, with all stakeholders, in order to optimise the registration of patients and the generation of knowledge for RD for which a therapeutic intervention is being developed. Collection of data regarding off-label use of approved drugs and existing medications should be encouraged.

6. Patient registries and data collections should be sustainable for the foreseeable timespan of the registries' utility.

6.1 Local, regional, national and European structures contributing to or overseeing data collection should all be supported financially to carry out this role in a sustainable way so that financial responsibility for registries is shared proportionately between stakeholders, MS and the EC and defined in the appropriate funding programmes.

6.2 Public-private partnerships for RD registries should be considered where applicable as a long-term model for optimisation of resources, sustainability and co-creation of knowledge.

6.3 All registries and data collections should have in place an exit strategy in its work plan, including contingency planning for the data in the event that the registry is terminated. There should also be a procedure outlined for succession planning for registry continuation.

C.4 EURORDIS-NORD-CORD Joint Declaration of 10 Key Principles for Rare Disease Patient Registries (2012)

(http://download.eurordis.org/documents/pdf/EURORDIS_NORD_CORD_JointDec_Registries_FINAL.pdf)

1. **Patient Registries should be recognised as a global priority in the field of Rare Diseases.**
2. **Rare Disease Patient Registries should encompass the widest geographic scope possible.**
3. **Rare Disease Patient Registries should be centred on a disease or group of diseases rather than a therapeutic intervention.**
4. **Interoperability and harmonization between Rare Disease Patient Registries should be consistently pursued.**
5. **A minimum set of Common Data Elements should be consistently used in all Rare Disease Patient Registries.**
6. **Rare Disease Patient Registries data should be linked with corresponding biobank data.**
7. **Rare Disease Patient Registries should include data directly reported by patients along with data reported by healthcare professionals**
8. **Public-Private Partnerships should be encouraged to ensure sustainability of Rare Disease Patient Registries.**
9. **Patients should be equally involved with other stakeholders in the governance of Rare Disease Patient Registries.**
10. **Rare Disease Patient Registries should serve as key instruments for building and empowering patient communities.**

On behalf of an estimated 60 million people living with rare diseases in Europe and North America, the European Organisation for Rare Diseases (EURORDIS), the National Organization for Rare Disorders (NORD) and the Canadian Organization for Rare Disorders (CORD), jointly submit the following declaration on common principles regarding Rare Disease Patient Registries.

EURORDIS, NORD and CORD, along with the patients they represent in Europe and in North America, recognize that Rare Disease Patient Registries constitute key instruments for increasing knowledge on rare diseases, supporting fundamental clinical and epidemiological research, and post-marketing surveillance of orphan drugs and treatments used off-label. Furthermore, and of great importance for patients and their families, they can be instrumental in supporting health and social services planning. Rare Disease Patient Registries are powerful, cost-effective instruments to improve the overall quality of care, quality of life and survival of patients.

EURORDIS, NORD and CORD also recognize that patient involvement is a key element in the successful establishment and long-term maintenance of Rare Disease Patient Registries and many patient groups are already very active and capable in this role. On behalf of rare disease patients and their representatives in Europe and in North America, we would like to jointly put forward the following common reflections and principles regarding patient registries. These common reflections and principles may serve as a reference to all other stakeholders when shaping policies and taking actions in the field of Rare Disease Patient Registries.

A Patient Registry can be defined as an organized system that uses observational study methods to collect uniform data (clinical and other) to evaluate specified outcomes for a population defined by a particular disease, condition, or exposure, and that serves a predetermined scientific, clinical, or policy purpose(s)[1]. The following principles refer to this definition.

[1] Gliklich RE, Dreyer NA, eds. Registries for Evaluating Patient Outcomes: A User's Guide. 2nd ed. Rockville, MD: Agency for Healthcare Research and Quality. September 2010. http://www.effectivehealthcare.ahrq.gov/ehc/products/74/531/Registries%202nd%20ed%20final%20to%20Eisenberg%209-15-10.pdf.

1. **Patient Registries should be recognised as a global priority in the field of Rare Diseases**.

Rare Disease Patient Registries represent a fundamental research effort upon which a number of critical activities are based. They constitute key instruments for increasing knowledge on rare diseases, by pooling data for epidemiological research, clinical research, and real-life post-marketing observational studies[2].

They broadly support health and social service planning by playing a pivotal role in healthcare organization. In particular, Centres of Expertise/Excellence and the European and International networks that connect them centralize patient data patient registries which can be used as an evidence base to shape regional, national and international health policy and standards of care.

It has also been demonstrated that Patient Registries are a major determinant for successful translational research in the field rare diseases. Where well-implemented registries and active patient organizations exist, the likelihood for developing a treatment for the disease in question is increased[3]. Furthermore, the consistent longitudinal collection of patient data facilitates the creation of standards of care and dramatically improves patient outcomes and life expectancy even in the absence of new therapies. The compelling arguments for Rare Disease Patient Registries as indispensable infrastructure tools for translating basic and clinical research into therapeutic solutions have elevated their status to a major priority for all stakeholders - a building block of any sound rare disease policy.

2. **Rare Disease Patient Registries should encompass the widest geographic scope possible.**

Due to the low individual prevalence and the scarcity of information related to each rare disease, collaboration and maximum use of limited resources is particularly meaningful for rare diseases. This is especially true for very rare diseases where no single institution, and in many cases no single country, has a sufficient number of patients to conduct fundamental, clinical and translational research. In fact, geographic dispersion of patients continues to make recruitment for clinical trials difficult, often aggravated by the dearth of scientific and medical knowledge and relevant endpoints for study designs.

The International Rare Diseases Research Consortium (IRDiRC)[4], launched in April 2011, fosters international collaboration in research on RD. Canada, Europe and the United States have fully committed to this endeavour agreeing on the principle that maximizing scarce resources and coordinating research efforts are key elements for success in the rare disease field. IRDiRC advocates that the worldwide sharing of information, data and samples gathered by robust and harmonised Rare Disease Patient Registries will boost research at all levels and ultimately favor therapy development.

3. **Rare Disease Patient Registries should be centred on a disease or group of disease rather than a therapeutic intervention.**

Treatment-specific registries, frequently funded by industry, are required by regulators to monitor the effectiveness and side-effects of treatments approved under exceptional circumstances. However, because treatment-specific registries must be re-created for each product, limitations in their completeness, quality, and cost-effectiveness have been demonstrated. Consensus is growing around the opinion that disease-centric patient registries provide a more comprehensive and collaborative approach to rare disease patient data collection by aligning stakeholder efforts, avoiding fragmentation of patient populations and dissipation of resources, and ultimately addressing regulatory and payer requirements with greater accuracy.

4. **Interoperability and harmonization between Rare Disease Patient Registries should be consistently pursued.**

Centres of Expertise/Excellence and the international networks that connect them play a pivotal role in capturing data of patients treated at their facilities and centralizing them in Rare Disease Patient Registries. Nevertheless, no uniform, accepted standards currently govern the collection, organization, or availability of data collected by Rare Disease Patient Registries which

[2] EURORDIS Position on Rare Disease Research. http://www.eurordis.org/publication/eurordis-position-rd-research
[3] Orphanet. Report on Rare Disease Research, Its Determinants in Europe and the Way Forward, May 2011. http://asso.orpha.net/RDPlatform/upload/file/RDPlatform_final_report.pdf
[4] International Rare Disease Research Consortium. http://ec.europa.eu/research/health/medical-research/rare-diseases/irdirc_en.html

may even vary within the same disease group or health system. Moreover, registry custodians frequently hold proprietary views on their data or face legal limitations on data-sharing as a result of patient consent restrictions and privacy protection or conflicting national legislations. These data-sharing barriers create a compelling argument for developing globally accepted definitions, classifications, ontologies[5,6], data standards and favourable and congruent policies and resources facilitating data sharing and pooling. Ideally, standard operating procedures and common resources or platforms for centralizing new or existing registries should be developed.

5. **A minimum set of Common Data Elements should be consistently used in all Rare Disease Patient Registries.**

A pillar for the systematic, coordinated approach to Rare Disease Patient Registries would be the definition of minimum set of Common Data Elements (CDEs) and corresponding validated standards and ontologies globally endorsed by all stakeholders. The consistent use of CDEs would facilitate the standardization of data (ensuring that data are defined and entered in the same way, use the same standards, and the same vocabularies), harmonization (allowing data to be more easily exchanged and compared), and interoperability (enabling common strategies for quality assurance and data security). Lastly, the definition of CDEs will allow greater opportunities for meta-analysis across diseases providing evidence for public health and social planning. The NIH Office of Rare Disease Research[7] and EPIRARE[8] are currently establishing such CDEs for North America and Europe.

6. **Rare Disease Patient Registries data should be linked with corresponding biobank data.**

Biobanks are collections of human biomaterials and represent an essential tool for fundamental and translational research. The high value of biological samples only increases when coupled with well-documented, associated data housed in a patient registry. The development of a system that assigns a unique global identifier to each patient is recommended to facilitate data linkage and avoid duplicate entries and waste of precious biomaterial. Engagement of patients and patient organizations is instrumental for the development of networks between registries and biobanks.

7. **Rare Disease Patient Registries should include data directly reported by patients *along* with data reported by healthcare professionals.**

Many patient organizations in Europe and North America are actively and successfully collecting clinical and non-clinical patient data. Most stakeholders in the rare disease community recognized that patients and their caregivers are best placed to report on their health-related quality of life, satisfaction with and utility of care and treatment. Much progress has been made in creating regulatory standards[9,10] to validate this type of data reported by patients and caregivers, which are also of significant benefit to patients' management of their own outcomes.

Out of necessity, patient groups further proceeded to collect data beyond perceived outcomes and collect post-marketing treatment outcomes, off-label drug use outcomes and even natural history data. By complementing clinician-reported data in Rare Disease Patient Registries, patients can contribute to improving their robustness, comprehensiveness and quality. Continued creation of easily accessible and validated standards, platforms and scientific guidance to ensure the high quality collection of patient entered clinical data should be encouraged and guaranteed.

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[5] Disease ontology refers to a consistent, reusable and sustainable set of descriptions that defines human disease terms, phenotypic/genotypic characteristics and related medical vocabulary. Common disease ontologies are needed to ensure both shared understanding between people and interoperability between information systems about diseases. Common ontologies are particularly important for rare diseases as exiting vocabulary (disease definition, diagnosis, phenotype/genotype) describing many of them is still incomplete and inconsistent.

[6] Rath A, Olry A, Dhombres F, Brandt MM, Urbero B, Ayme S (2012) Representation of rare diseases in health information systems: The Orphanet approach to serve a wide range of end users. Hum Mutat 33:803-8. http://onlinelibrary.wiley.com/doi/10.1002/humu.22078/pdf

[7] NIH Office of Rare Disease Research. Common Data Elements. http://www.grdr.info/files/ORDR_CDE_10_2_2012.xls

[8] European Platform for Rare Disease Registries (EPIRARE). http://www.epirare.eu/

[9] US Food and Drug Administration. http://www.fda.gov/downloads/Drugs/GuidanceComplianceRegulatoryInformation/Guidances/UCM193282.pdf

[10] European Medicines Agency. http://www.ema.europa.eu/ema/pages/includes/document/open_document.jsp?webContentId=WC500003637

8. **Public-private partnerships should be encouraged to ensure sustainability of Rare Disease Patient Registries.**

In context of the current economic climate, the need for the optimal sharing of resources is an imperative. Different scenarios are being proposed to provide financial sustainability to registries and their networks, and the most promising rely on the collaboration amongst all the stakeholders[11,12]. This collaborative approach has been recognized as a requirement to: avoid duplication of efforts and take advantage of economies of scale; foster improved quality and robustness of data collected; to unify patient data especially for diseases where several treatments exist, and best sustain registries as long-term endeavours. With both governments and private groups showing interest in patient registries, public-private partnerships are a promising collaborative scheme. Patient groups can be instrumental facilitators of public-private partnerships driving the common goals of all stakeholders through a patient-centred approach and assuring optimal efficiency and transparency. Regulatory bodies can strongly encourage such collaboration in this pre-competitive space. The nature of potential public-private partnerships, the issues to consider when establishing such a partnership, and best practices enhancing the success of such efforts should be investigated in a prompt and transparent manner.

9. **Patients should be equally involved with other stakeholders in the governance of Rare Disease Patient Registries.**

Patient involvement is a key element in the successful establishment of registries and many patient groups are already very active in this role. Patients should be involved at all levels of development, management and maintenance in order to best represent patient needs, increase awareness among all stakeholders of the existence of the registry and, ultimately, improving the quality and quantity of data collected through a patient-centred approach. Patient groups are willing and able to be involved in initiating the establishment of registries; defining content and purposes of the registries; resolving ethical and legal issues; authorising access and utilisation of data; creating partnerships with health professionals and industry representatives; contributing to the selection of data items collected (in particular on the impact of the disease on their daily life); helping to recruit patients for participation into the registry; preparing specific information for patients to be registered prior to their consent; motivating health professionals to input data, and directly entering data. This essential role of the patients should be reflected in the governance of the registry.

10. **Rare Disease Patient Registries should serve as key instruments to build and empower patient communities.**

Registries can be instrumental in building patient communities around a disease, a cluster of diseases or even common clinical features or common underlying causes. Registries thus become the aggregation point around which an organised patient community can be built where none exists. The creation of a patient registry can facilitate the congregation of patients and their families as they engage directly into the development of the very databases in which their data will be entered. Registries thus become the medical home for patients scattered internationally and empower patients with data available to share with health care professionals, clinical researchers and drug developers.

[11] NIH/FDA Workshop on Natural History Studies of Rare Diseases https://events-support.com/events/Natural_History_Studies
[12] EUCERD Workshop of Public-Private Partnerships for RD Registries.

C.5 EUROPLAN Recommendations

EUROPLAN recommendations on area 2: Adequate definition, coding and inventorying of rare diseases

R 2.1 The European definition of rare diseases is adopted in order to facilitate transnational cooperation and community level actions (e.g.: collaboration in diagnosis and health care; registry activities).

R 2.2 The use of a common EU inventory of rare diseases (Orphanet) is promoted in the national health care services and collaboration is carried out to keep it updated.

R 2.3 Coding of rare diseases is promoted, encouraging their traceability in the national health system.

R 2.4 Cross-referencing rare diseases is carried out across the different classification systems in use in the country, ensuring coordination and coherence with European initiatives, such as reference to the Orpha-code.

R 2.5 Collaboration with the ICD10 revision process is ensured and ICD-11 is adopted as soon as possible.

R 2.6 Healthcare professionals are appropriately trained in recognizing and coding rare diseases.

R 2.7 Initiatives are promoted at national level for the integrated use of administrative, demographic and health care data sources to improve the management of rare diseases.

R 2.8 International, national and regional registries for specific rare diseases or groups of rare diseases are promoted and supported for research and public health purposes, including those held by academic researchers.

R 2.9 Collection and sharing of data from any valid sources, including Centres of Expertise, and their availability for public health purposes is promoted by public health authorities, in compliance with national laws.

R 2.10 Participation of existing national registries in European/International registries is fostered.

R 2.11 Instruments are identified for combining EU and national funding for registries.

EUROPLAN recommendations on Area 6: Patient Empowerment

[...]

R 6.3 Valid information on rare diseases is produced and made available at national level in a format adapted to the needs of patients and their families.

R 6.4 National information of interest to patients is communicated to EURORDIS for publication in its website.

[...]

R 6.8 Interactive information and support services for patients are promoted (such as help lines, e-tools etc).

R 6.9 Information and education material is developed for specific professional groups dealing with rare diseases patients (e.g. teachers, social workers, etc.).

http://www.europlanproject.eu/_newsite_986987/_down/results/2008-2011_2.EUROPLANGuidance.pdf

C.6 EUCERD Core Indicators

http://www.eucerd.eu/wp-content/uploads/2013/06/EUCERD_Recommendations_Indicators_adopted.pdf

NB: Out of the 21 EUCERD core indicators, please find below selected indicators for this specific theme.

4. Adoption of the EU RD definition

8. NP/NS support to the development of/participation in a comprehensive national and/or regional RD information system

9. Existence of Help lines for RD

11. Type of classification/coding used by the health care system

12. Existence of a national policy on registries or data collection on RD

Core Indicators – Definitions and associated answers

INDICATOR	**AREA OF COUNCIL REC.** (2009/ C151/02)	**INDICATOR DESCRIPTION**	**TYPE OF INDICATOR**	***SHORT* ANSWER**	***DETAILED* ANSWER** *(multiple answers are possible, if needed)*
BACKGROUND INDICATORS *(PREPARATION OF THE PLAN/STRATEGY)*					
4. Adoption of the EU RD definition	2	The EU defines "rare diseases" as those with a prevalence of no more than 5 patients per 10.000 persons. This definition is laid down in Regulation EC n° 141/2000 on Orphan Medicinal Products, Directive 2011/24/EU on Cross Border Healthcare as well as in the Council Recommendation on an action in the field of rare diseases of 8 June 2009.	Process	YES	**YES**, the NP/NS measures are applied using the EU definition
					YES, but the NP/NS measures are applied using a different definition
				NO	*Please, specify the definition used in the NP/NS*
CONTENT INDICATORS					
INFORMATION					
8. NP/NS support to the development of/participation in an	2	This indicator refers to the existence of a functional, RD-specific information system that is comprehensive and nation-wide (such as Orphanet).	Process	YES	**YES**, national

information system on RD		This indicator includes the participation in the Orphanet Joint Action and eventually the production of information packages in national language(s).			**YES,** regional/s
				NO	
				Participation in the Orphanet Joint Action	**YES**, participates in Orphanet JA and produces information in national language(s)
					YES, participates in Orphanet JA but does not produce information in national language(s)
					NO
9. Existence of Help lines for RD	2 & 6	The availability of help lines is fundamental for the diffusion of information and expertise on rare diseases. They have an important role in orienting patients towards a solution to the issues that directly or indirectly affect him/her as a result of the condition and are the only service that can offer social, psychological and information solutions to all of these needs. Professionals (including those working in emergency departments) may learn about resources and pathways to diagnose their patients or receive important information regarding the management of patients with a rare disease. This indicator aims to account for the national help lines on rare diseases, either aimed at patients or professionals (or both), including those not publicly funded.	Process	YES, supported by public funding	**YES**, only for professionals
					YES, only for patients
					YES, for both professionals and patients
				YES, supported by private funding	**YES**, only for professionals
					YES, only for patients
					YES, for both professionals and patients
				YES, supported by public funding and by private funding	**YES**, only for professionals
					YES, only for patients
					YES, for both professionals and patients
				NO	

KNOWLEDGE, CLASSIFICATION/CODING, REGISTRIES AND RESEARCH					
11. Type of classification/coding used by the health care system	2	The adoption and the daily use of an internationally recognised, comprehensive, health care codification system is important for RD management and would encourage the harmonisation of disease nomenclature worldwide. This enables budgetary and management decisions to have a more solid basis and would constitute one relevant tool for Health Technology Assessment.	Process	*Type of coding system used*	ICD-9
					ICD-10
					OMIM
					SNOMED
					MESH
					ICD-O
					Others
				ORPHA Code is used in addition to national coding system	YES
					NO
12. Existence of a national policy on registry and data collection on RD	2 & 3	This indicator collects information on Member States' support, at all appropriate levels, to rare diseases registries and databases for epidemiological, public health and research purposes, as well as on the role ensured by public authorities for the coordination and sustainability of data collection.	Process	YES	**YES**, for national/centralised registry and data collection
					YES, for regional registry and data collection
				NO	

C.7 EUROPLAN Indicators

http://www.europlanproject.eu/_newsite_986989/Resources/docs/2008-2011_3.EuroplanIndicators.pdf

Area to be explored	Aims	Actions		Indicators	Type of indicator	Answers
2. Adequate definition, codification and inventorying of rare diseases	Use a common definition	To officially adopt the EC RD definition (No more than 5 cases / 10,000 inhabitants)	**2.1**	**Adoption of the EC RD definition**	Process	• *Yes* • *not* • *EU definition modified with an additional definition*
	Ensure that RD are adequately coded and traceable in the health care information system	To include the best RD diseases classification currently existing into the public health care related services	**2.2**	**Type of classification used by the health care system**	Process	• *ICD-9* • *ICD-10* • *OMIM* • *SNOMED* • *ORPHAN* • *MESH* • *Others*
			2.3	**Developing policies for recognising RD by the care information systems**	Process	• *Not existing, not clearly stated* • *Existing, clearly stated, partly implemented and enforced* • *Existing, clearly stated and substantially implemented and enforced*
	Support registries for better epidemiological knowledge	Defining a surveillance system based on a patient outcomes registry	**2.4**	**Registering activity**	Process	• *Centralized RD registry* • *Multiple RD registries but well coordinated and standardised* • *Multiple RD registries not standardised* • *No registry at all*
			2.5	**Number of diseases included**	Outcomes	*Number ranging from 1 to 20*

Area to be explored	Aims	Actions		Indicators	Type of indicator	Answers
5. Gathering the expertise on Rare Diseases at European level	Improving education and training	Existence of a information sites for professionals provided by the plan/strategy	**5.1**	**Existence of a comprehensive national and/or regional RD information system supported by the government**	Process	• *Yes, covers most RD* • *Yes, covers only some RD* • *Not formal decisions* have *been taken*
			5.2	**Help lines for professionals**	Process	• *Yes, covers most RD* • *Yes, covers only some RD* • *Not formal decisions have been taken*
			5.3	**Clinical guidelines**	Outcomes	*Number ranging between 0 to 30*
		Promoting training activities and awareness educational campaigns among professionals	**5.4**	**Number of such activities** (training and awareness educational) **promoted by the plan/strategy**	Outcomes	*Number ranging between 0 to 30*
Area to be explored	**Aims**	**Actions**		**Indicators**	**Type of indicator**	**Answers**
6. Empowerment of Patients	Establishment of a mechanism that ensures that patients are empowered to directly contribute to shaping healthcare policies that affect their lives	Building - supporting the existence of comprehensive help line and information sites for patients provided by the plan/strategy	**6.10**	**Availability of Help line for RD**	Process	• *Own help line* • *Referred RD help lines* • *Not formal decisions have been taken*