EUCERD
European Union Committee of Experts on Rare Diseases

EUCERD RECOMMENDATIONS ON CORE INDICATORS FOR RARE DISEASE NATIONAL PLANS/STRATEGIES

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INTRODUCTION

1. BACKGROUND TO THE RECOMMENDATIONS

Council Recommendation. The Council of the European Union, in its Recommendation of 8 June 2009[1] on an action in the field of rare diseases, recommends that "Member States elaborate and adopt a plan or strategy as soon as possible, preferably by the end of 2013 at the latest, aimed at guiding and structuring relevant actions in the field of rare diseases within the framework of their health and social systems".

EUROPLAN Indicators. In order to support this process, the programme of Community action in the field of Public Health co-funded "EUROPLAN" (the European Project for Rare Disease National Plans Development), a three-year project (2008-2011) coordinated by the Istituto Superiore di Sanità (ISS) - Italian National Centre for Rare Diseases. The aim of EUROPLAN was to develop tools and carry out activities that would help EU countries to establish and implement national plans or strategies in the field of rare diseases. One of the tools that have been created is the "Report on indicators for monitoring the implementation and evaluating the impact of National Plans or Strategies for rare diseases"[2] .

The development and use of indicators is an integral part of planning and designing health and social services, as they can be used as management tools, as well as for monitoring the implementation of National Plans for Rare Diseases.

Elaborated in 2010, the "EUROPLAN Indicators" are a set of 59 indicators for health and social planning monitoring, intended for policy makers and planners. While all indicators were considered relevant, from an operational perspective, they were quite numerous and difficult to handle. Accordingly, within the subsequent "EUCERD Joint Action: Working for Rare Diseases (2012-2015)", work was planned to derive a selection of core indicators based on their usefulness and feasibility within the activities of its Work Package 4 "EUROPLAN 2012-2015" coordinated by the ISS.

These Recommendations deals with indicators for monitoring health and social planning and does not intend to focus on health indicators for rare diseases, aimed to provide information on the health status of populations and the impact of health policies on this status. For this purpose, the Rare Diseases Task Force-Working Group on Indicators (RDTF-WG) published in 2010 a Report "Health Indicators for Rare Diseases I – Conceptual Framework and Development of Indicators from Existing Sources"[3] that proposes a list of sources and types of health indicators in the field of rare diseases.

[1] Council Recommendation of 8 June 2009 on an action in the field of rare diseases (2009/C 151/02).
[2] The document may be downloaded from the EUROPLAN website (www.europlanproject.eu).
[3] Rare Disease Task Force, "Health Indicators for Rare Diseases I – Conceptual Framework and Development of Indicators from Existing Sources", Final Report – April 2010, http://www.eucerd.eu/?post_type=document&p=1211

2. METHODOLOGY

The Recommendations listed here define "Core Indicators" for monitoring Rare Disease National Plans/Strategies. They were derived from the initial set of 59 Indicators from EUROPLAN I project, organised as process and outcome indicators. The current selection of Core Indicators is the result of two parallel efforts. One was a Delphi Method organised by ISS, where representatives of all of the EU Ministries of Health[4] and experts (e.g. epidemiologists, health services planning experts, rare disease experts) classified the indicators according to their usefulness and feasibility. The other methodology consisted of an appraisal of the indicators by 9 EUROPLAN-EURORDIS Advisors, on behalf of their National Alliances of patients' associations, and representatives of the Ministries of Health, under the auspices of EURORDIS. They rated the full list of EUROPLAN I indicators based on usefulness for patients and carers, feasibility and political usefulness.

The results from the two methodologies were compared and merged to select the final list, which comprises the highest scoring indicators.

The outline of the present document was presented at the 7th EUCERD meeting (Luxemburg, 31th January 2013), and at the EUCERD Joint Action WP4/EUROPLAN Workshop (Rome, 25th March 2013). The EUCERD received the post-workshop draft for their input over then days before the document was finalised and then submitted to EUCERD members 4 weeks in advance of their 8th EUCERD meeting on 5-6 June 2013 for adoption, where the present recommendations were adopted.

3. TARGET GROUPS FOR THESE RECOMMENDATIONS

The target groups of these Recommendations are the EU Member States and the European Commission.

Additionally, their dissemination is intended to other target audiences, such as EC initiatives (e.g. other projects and joint actions, the Cross-Border Healthcare Expert Group, EUnetHTA, EPAAC), Centres of Expertise in the field of rare diseases, healthcare providers, rare disease experts and existing rare disease network co-ordinators and partners, patient organisations.

[4] With the exception of the Germany Ministry for Health which deemed it too early for their country to be involved in this kind of process, with regards to the development of their national plan.

RECOMMENDATIONS OF THE EUCERD TO THE EUROPEAN COMMISSION AND THE MEMBER STATES

MISSION, VISION AND SCOPE

1. The **overall objective** of the Core Indicators for Rare Disease National Plans and Strategies (hereafter "**Core Indicators**") is to capture relevant data and information on the process of planning, implementing and monitoring of these plans and strategies. The Core Indicators are therefore instrumental for the decision-making process related to the adoption, assessment and further development of public policies for rare diseases.

2. EU Member States should use these Core Indicators to collect data on an annual basis.

3. The data collected shall be used

    a) By Member States as a supporting instrument to follow up the policy initiatives integrated in their National Plans and National Strategies for Rare Diseases, adopted in response to the Council Recommendation of 8 June 2009 on an action in the field of rare diseases. Naturally, this process does not prevent Member States using additional indicators to monitor and evaluate their plans.
    b) For the annual reporting at Member States level to elaborate the "Report on the State of the Art on Rare Disease Activities in Europe of the European Union Committee of Experts on Rare Diseases", published on a yearly basis by the EUCERD. The EUCERD will incorporate these Core Indicators in the report's elaboration guidelines for Member States.
    c) Moreover, the data collected may be used to further inform the Implementation Report that the European Commission is due to prepare before the end of 2013, as stipulated in the afore-mentioned Council Recommendation.

4. Following the adoption of these Recommendations, the EUCERD may revise the List of Core Indicators and propose, where appropriate, amendments. This revision will rely upon the experience gained in EU Member States with the adoption and implementation of National Plans and Strategies on Rare Diseases, as commended by the Council Recommendation.

List of Core Indicators

The following list of the Core indicators is proposed to **all EU Member States so as to monitor their National Plans or Strategies on Rare Diseases**.

BACKGROUND INDICATORS *(PREPARATION OF THE PLAN/STRATEGY)*

1. Existence of regulations/laws, or equivalent official national decisions that support the establishment and development of a Rare Diseases (RD) plan
2. Existence of a RD advisory committee
3. Permanent and official patients' representation in plan development, monitoring and assessment
4. Adoption of the EU RD definition

CONTENT INDICATORS

Centres of Expertise

5. Existence of a national policy for establishing Centres of Expertise on RD
6. Number of national and regional Centres of Expertise adhering to the national policy
7. Participation of national or regional Centres of Expertise in European Reference Networks

Information

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

Knowledge, classification/coding, registries and research

10. Existence of a national policy on rare disease clinical practice guideline development and implementation
11. Type of classification/coding used by the health care system
12. Existence of a national policy on registries or data collection on RD
13. Existence of RD research programmes and/or projects in the country
14. Participation in European and international research initiatives

Therapies

15. 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)
16. Existence of a governmental system for compassionate use of medicinal products

Social services

17. Existence of programmes to support the integration of RD patients in their daily life

FINANCIAL SUPPORT INDICATORS *(IMPLEMENTATION OF THE PLAN/STRATEGY)*

18. Existence of a policy/decision to ensure long-term sustainability of the RD plan/strategy
19. Amount of public funds allocated to the RD plan/strategy
20. Specific public funds allocated for RD research
21. Public funds specifically allocated for RD research actions/projects per year since the plan started

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)*					
1. Existence of Regulations/Laws, or equivalent official national decisions that support the establishment and development of a Rare Diseases (RD) plan	1	This Indicator refers to the fact that National Plans/Strategies for Rare Diseases should be devised/regulated at national level in accordance with the Council Recommendation on RD, relevant Recommendations of the EUCERD e.g. those on Centres of Expertise and European Reference Networks, as well as relevant legislation (Regulation EC n° 141/2000 on Orphan Medicinal Products, Directive EU/2011/24 on Cross Border Healthcare, etc.). The National Plan or Strategy is adopted via binding legislative acts, the exact nature or level of which may vary (regulation, laws, or other types of decisions). They may be established at the appropriate level of governance (federal vs. federated state level) depending on the country's system of government. It is therefore embedded in a legislative or operational framework.	Process	YES	**YES**, existing, fully embedded in a regulation/law/official national decision
					YES, existing, partly embedded
				In progress /in development	
				NO	
2. Existence of a RD advisory committee	1	The Expert Advisory Committee refers to the existence of a coordination mechanism that oversees the development and implementation of the National Plan/Strategy for Rare Diseases. This body is composed of representatives of all relevant stakeholders, including patient representatives, national government, industry, treating physicians, payers, academia, etc.	Process	YES	**YES**, exists and meets regularly and includes all relevant stakeholders
					YES, exists but partly functioning and includes all relevant stakeholders
					YES, exists and meets regularly but does not include all relevant stakeholders
					YES, exists but partly functioning and does not include all relevant stakeholders
				NO	
3. Permanent and official patients'	6	Patients are officially represented at all stages of plan	Process	YES	**YES**, at all stages
					YES, but only as observers

representation in plan development, monitoring and assessment		development and governance, including its monitoring and evaluation.			**YES**, but only consulted before the final document is approved
				NO	
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					
CENTRES OF EXPERTISE					
5. Existence of a national policy for establishing Centres of Expertise on RD	4	This policy defines a strategy to identify and designate centres of expertise, aiming to improve the quality of health care by defining appropriate centres with experience on RD as well as pathways that reduce the diagnosis delay and facilitate both care and treatment for RD patients.	Process	YES	**YES**, existing, fully implemented
					YES, existing, partly implemented
				In progress/in development	
				NO	
6. Number of national and regional Centres of Expertise adhering to the national policy	4	Member States identify and appoint Centres of Expertise (CEs) throughout their national territory, and consider supporting their creation. The Centres of Expertise should adhere to the national policy. It is to be remembered that the EUCERD adopted the "EUCERD Recommendations on Quality Criteria for Centres of Expertise" which are "intended to help EU Member States in their reflections or policy developments concerning national plans	Outcomes	*Number*	*Number of CEs complying with the national policy*
					Number of CEs / million inhabitants

		and strategies for rare diseases when addressing the issue of organisation of healthcare pathways at national and European level". This indicator therefore also aims to count the number of Centres of Expertise that are compliant with the EUCERD recommendations.			*Number of CEs fulfilling EUCERD criteria*
7. Participation of national or regional centres of expertise in European Reference Networks	4	The information on the integration of national Centres of Expertise in European Reference Networks (ERNs) is essential to obtain the broader picture of RD care across Europe and enables the diffusion of expertise across the EU, regardless of the size/population of each country. According to the "EUCERD Recommendations on European Reference Networks for Rare Diseases", different forms of affiliation to an RD ERN (association, collaboration) should be allowed to ensure inclusivity." Therefore this indicator aims to differentiate between full and associated membership of RD Centres of Expertise to RD ERNs. However, it should be taken into account that it will take some time before ERNs are established. Therefore it should be expected that this Indicator will provide meaningful information only a few years after the adoption of these Recommendations.	Outcomes	*Number of CEs participating in ERNs as full members* *Number of CEs participating in ERNs as associated members*	
INFORMATION					
8. NP/NS support to the development of/participation in an information system on RD	2	This indicator refers to the existence of a functional, RD-specific information system that is comprehensive and nation-wide (such as Orphanet). This indicator includes the participation in the Orphanet Joint Action and eventually the production of information packages in national language(s).	Process	YES	**YES**, national **YES,** regional/s
				NO	
				Participation in	**YES**, participates in Orphanet

				the Orphanet Joint Action	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					
10. Existence of a national policy for developing ,adapting and implementing clinical practice guidelines	2	The indicator checks the existence of a policy for developing, adapting and implementing clinical practice guidelines (CPGs) for diseases/groups of diseases *("Adapting" refers to adaption of supra-nationally based clinical guidelines to the local context)*.The cumulative production of protocols and clinical guidelines is an instrument for equity of access to care by rare disease patients across the European Union.	Process	YES	**YES**, a policy exists for developing CPGs
					YES, a policy exists for adapting CPGs
					YES, a policy exists for implementing CPGs
				NO	
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	Process	*Type of coding system used*	ICD-9
					ICD-10
					OMIM

		constitute one relevant tool for Health Technology Assessment.			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	
13. Existence of a RD research programmes/projects in the Country	3	This indicator aims to describe the status of RD research in the country, most notably whether a dedicated programme exists, or whether RD research is carried out by individual projects within the general research programme.	Process	YES	**YES**, specific research PROGRAMME
					YES, specific PROJECTS for RD within general research programme
				NO	
14. Participation in European and international research initiatives	3	Participation of national research agencies in international research initiatives (such as E-RARE – www.e-rare.eu, and IRDiRC – www.irdirc.org) is important to foster research on rare diseases a global level, by pooling resources and coordinating national research programmes to overcome the fragmentation of research on RD.	Process	YES	**YES**, E-RARE
					YES, IRDIRC
					YES, others (specify)
				NO	
THERAPIES					
15. Number of Orphan Medical Products (OMPs) with a European Union marketing authorisation and available in the country (i.e. priced and reimbursed or directly	5	The actual availability of OMPs in the national market is essential to illustrate patients' access to treatment in their country. Moreover, with patient access to OMPs differing across Member States, the success of cross border healthcare depends	Outcomes	*Number*	

supplied by the national health system)		on the harmonisation of access to diagnosis and treatment. Therefore, quantifying the drugs that are available in each country, either in ambulatory or in-hospital regimens, is also important to bridge the existing gap between Member States.			
16. Existence of a governmental system for compassionate use of medicinal products	5	The indicator aims to identify whether a system exists to provide medicines to rare diseases patients prior to approval of new drugs (so-called compassionate use).. The existence of such programmes is relevant for the assessment of overall RD care.	Process	YES	
				In progress /in development	
				NO	
SOCIAL SERVICES					
17. Existence of programmes to support in their daily life RD patients integration	6	Rare Diseases often lead to disability and a need for continuous care. Specialised Social Services are instrumental in providing patients with a full, rewarding life. Their existence and number demonstrate the political commitment of Member States to this mission. Examples of social services to integrate patients in their daily life and support their psychological and educational development are: a) educational support for patients, relatives and caregivers; b) individual support at school, for both pupils with rare diseases and teachers, including disease-specific good practices; c) activities aimed to foster higher education for people with rare diseases; d) supporting mechanisms to participate in work life for people with disabilities.	Process	YES	**YES**, people living with RD can access general programmes for persons with a disability *(Please, specify -see examples in the indicator description-: a, b, c, d, others)*
					YES, there exist specific actions to enable real access for people living with RD to general social/ disability programmes (e.g. training, guidelines for social workers, etc.) *(Please, specify -see examples in the indicator description-: a, b, c, d, others)*
					YES, there exist specific programmes for people living with RD *(Please, specify -see examples in the indicator description-: a, b, c, d, others)*
				In progress /in development	*(Please, specify -see examples in the indicator description-: a, b, c, d, others)*
				NO	

FINANCIAL SUPPORT INDICATORS *(IMPLEMENTATION OF THE PLAN/STRATEGY)*					
18. Existence of a policy/decision to ensure long-term funding and/or sustainability of the measures in the RD plan/strategy	7	The indicator verifies whether the financial commitment for rare disease care and treatment is clearly defined in a budget decision that supports the implementation of the National Plan/Strategy actions.	Process	YES	**YES**, a policy/decision to ensure long-term sustainability
					YES a budget exists for the plan
				In progress /in development	
				NO	
19. Amount of public funds allocated to the RD plan/strategy	7	The indicator is the overall budget (in EUR) allocated per year to the National Plan/Strategy (excluding reimbursement of care and cost of standard care, excluding cost of orphan drugs). As with the previous indicator, this indicator aims to ensure that RD actions include appropriate provisions to ensure their sustainability over time. Efficient and effective actions for rare diseases depend on integrating scarce and scattered resources both nationally and within a common European effort.	Outcomes	*Number*	Value
					Value / million inhabitants
					Value available partially: only for funds allocated exclusively to National Plan (N/A for funds allocated in the general budget)
					N/A: it is incorporated in the general budget
20. Specific public funds allocated for RD research	3	This indicator aims to identify the policy decision(s) to allocate a portion of the national research budget specifically to RD research.	Process	YES	
				In progress /in development	
				NO	
21. Public funds specifically allocated for RD research actions/projects per year since the plan started	3	This indicator verifies the total amount of public funds (in EUR) allocated to RD research projects or programmes	Outcomes	*Number*	Value
					Value available partially: only for funds allocated exclusively to National Plan (N/A for funds allocated in the general budget)
					N/A: it is incorporated in the general research funds

KEY DOCUMENTS

- **Directive (EC 2011/24/EU) of the European Parliament and of the Council on the application of patients' rights in cross-border health care**

  http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:L:2011:088:0045:0065:EN:PDF

- **Commission Communication, Rare Diseases Europe's Challenge**

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

- **EC. (2009). COUNCIL RECOMMENDATION of 8 June 2009 on an Action in the Field of Rare Diseases**

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

- **EUROPLAN RECOMMENDATIONS. Guidance document containing the recommendations for the designing of the National Plans or Strategies for rare diseases**

  http://www.europlanproject.eu/_newsite_986987/Resources/docs/2008-2011_2.EUROPLANRecommendations.pdf

- **EUROPLAN REPORT ON INDICATORS for monitoring the implementation and evaluating the impact of National Plan or Strategy for rare diseases**

  http://www.europlanproject.eu/_newsite_986987/Resources/docs/2008-2011_3.EUROPLANIndicators.pdf

- **RDTF Report: Health indicators for rare diseases I – Conceptual framework and development of indicators from existing sources - April 2010**

  http://www.eucerd.eu/?post_type=document&p=1211