



EUROPLAN PROJECT NATIONAL CONFERENCE ON RARE DISEASES

WORKSHOP

ADEQUATE DEFINITION, CODIFICATION AND INVENTORYING OF RARE DISEASES

(with additional questions on information and training)



**RELEVANT EXTRACT FROM THE
EUROPEAN COUNCIL RECOMMENDATIONS
ON ACTIONS IN THE FIELD OF RARE DISEASES**

LUXEMBOURG, 9 June 2009

COUNCIL RECOMMENDATIONS

“HEREBY RECOMMENDS that Member States:

- **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.**
- **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.**

COUNCIL RECOMMENDATIONS

- **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.**
- **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.”**



**RELEVANT EXTRACT FROM THE
SPECIFIC EUROPLAN RECOMMENDATIONS
FOR THE DEVELOPMENT OF NATIONAL PLANS
FOR RARE DISEASES**

EUROPLAN RECOMMENDATIONS

- 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; registration activities)
- 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
- Coding of rare diseases is promoted, encouraging their traceability in the national health system
- Cross-referencing rare diseases is carried out across the different classification systems adopted in the country, ensuring coordination and coherence with European initiatives, such as the reference to the Orpha-code

EUROPLAN RECOMMENDATIONS

- The collaboration in the ICD10 revision process is ensured and ICD-11 is adopted as soon as it becomes available
- Healthcare professionals are appropriately trained in coding rare diseases
- 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
- National/regional registries of rare diseases are promoted and supported for public health purposes under the management of the health authorities or other stakeholders. Public/academia partnerships for the establishment and maintenance of registries are stimulated
- Participation of existing registries and centres of expertise in European registries is fostered

EUROPLAN RECOMMENDATIONS

- Collaboration at European level is ensured for assessing and agreeing on ethical and legal issues (ownership, intellectual property, personal data protection, etc.), scope and use of European/national registries and funding aspects, which can hamper the functioning of many European registries
- Instruments are identified for combining EU and national funding for registries



**RELEVANT EXTRACT FROM THE
EUROPLAN INDICATORS
TO EVALUATE THE ACHIEVEMENTS OF RD INITIATIVES**

EUROPLAN INDICATORS

ACTIONS	INDICATORS	TYPE	ANSWERS
<p>To officially adopt the EC RD definition (no more than 5 cases/10,000 inhabitants)</p>	<p>Adoption of the EC RD definition</p>	<p>Process</p>	<p>Yes No EU definition modified with an additional definition</p>
<p>To include the best Rare Diseases classification currently existing into the public health care related services</p>	<p>Type of classification used by the health care system</p>	<p>Process</p>	<p>ICD-9 ICD-10 OMIM SNOMED ORPHAN MESH Others</p>

EUROPLAN INDICATORS

ACTIONS	INDICATORS	TYPE	ANSWERS
<p>To include the best Rare Diseases classification currently existing into the public health care related services</p>	<p>Developing policies for recognising RD by the care information systems</p>	<p>Process</p>	<p>Not existing, not clearly stated</p> <p>Existing, clearly stated, partly implemented and enforced</p> <p>Existing, clearly stated and substantially implemented and enforced</p>
<p>Defining a surveillance system based on a patient outcomes registry</p>	<p>Registering activity</p>	<p>Process</p>	<p>Centralised RD registry</p> <p>Multiple RD registries but well coordinated and standardised</p> <p>Multiple RD registries not standardised</p> <p>No registry at all</p>
	<p>Number of diseases included</p>	<p>Outcomes</p>	<p>Number ranging from 1 to 20</p>



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FURTHER GUIDELINES FOR DISCUSSION
(including questions on information and training)

DEFINITION OF RARE DISEASES

- Is the EU official definition (RD are those affecting up to 5 out of 10 000 person) used in your country?
- Are there alternative or more specific definitions used instead or in addition?

CLASSIFICATION AND TRACEABILITY OF RDs IN THE NATIONAL HEALTH SYSTEM

- What classification system is used in your country? ICD9, ICD10, SNOMED, OMIM, ORPHAN...
- For which purpose is (are) the classification system(s) used, e.g. surveillance, reimbursement, provision of social support, etc.
- Is your country prepared to adopt the WHO-led system, the ICD-11, recommended by the EU in the Council Recommendation on RD, when ready (2014)?
- What level of awareness and knowledge do healthcare professionals have of the RD classification and codification? What can be done to improve it?

INVENTORIES, REGISTRIES AND LISTS

- Are there official lists of RD in your country? Is there an official governmental RD registry? And/or specific RD databases e.g. held by Centres of Expertise? Are there RD surveillance projects or programmes (e.g. sentinel programmes, surveys)?
- What kind of initiatives should be taken or reinforced in your country?
- Do these registries and programmes receive government support?
- How to ensure, through appropriate funding mechanisms, the long-term sustainability of registries and databases?
- Does your country participate to the development of a EU inventory of RD as recommended in the Council Recommendations on RD?

HOW TO IMPROVE ACCESS TO QUALITY INFORMATION ON RD

- How are these information sources and initiatives publicised?
- How to make sure that they reach out to the target audiences? And to the wider public?

HOW TO IMPROVE INFORMATION ON AVAILABLE CARE FOR RARE DISEASES

- What are the existing information sources in the country? Are they of good quality? Do they receive public funding or Patients Org. funding?
 - Is there a national official website for RD in the country?
 - Are there help lines for both patients and healthcare professionals? Are they known to the public?
 - Are there initiatives of centres of expertise and/or patient organisations or programme to stimulate the development of information and educational material for patients or specific publics (teacher, social workers, etc.)?
 - Are existing resources at EU level, Orphanet and Eurordis used: (information on diseases, specialised centres and patient groups, ongoing research projects, clinical trials)?
 - Are there initiatives to raise awareness on RD such as a RD Day?

HOW TO ENSURE ADEQUATE TRAINING OF HEALTHCARE PROFESSIONALS ON RARE DISEASES

- Training for healthcare professionals may include :
 - Training to make them aware of the existence of RDs and of resources available for their care;
 - Medical training in fields relevant to diagnosis of RDs (e.g. genetics, oncology, immunology, neurology, paediatrics);
 - Medical training of young doctors and scientists in the field of RD;
 - Exchange and sharing of expertise between centres within the country.

HOW TO ENSURE ADEQUATE TRAINING OF HEALTHCARE PROFESSIONALS ON RARE DISEASES

- How to ensure that existing and validated international guidelines are used at national level to guide diagnosis and treatment of RD?
- What mechanisms can be put in place to support the exchange of expertise at EU level and the adequate training for all healthcare professionals?
- Is your country supporting the participation of national experts in developing international guidelines to guide diagnosis and treatment of RD at national level?



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PROPOSALS FROM THE AUDIENCE