



**DEFINITION,  
CODIFICATION,  
INVENTORYING  
OF RARE DISEASES  
&  
REGISTRIES**




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- Relevant extracts from the **RECOMMENDATION of the Council of the European Union** on “an action in the field of rare diseases”.
- Relevant extracts from the **RECOMMENDATION of the EUCERD** - European Union Committee of Experts on Rare Diseases – on ‘**Core Indicators**’ for planning, implementing and monitoring national Rare Disease Plan or Strategy.

The EUCERD brought together the 28 EU Member States plus Norway, Iceland and Switzerland, and stakeholders from patients’ organisations, academia and industry.

- Relevant extracts from the **RECOMMENDATION of EUROPLAN** – EU co-funded project aimed at developing and implementing national Rare Disease Plan or Strategy.
- Relevant extracts from **EUROPLAN Indicators**.



**Relevant extracts from the**  
**RECOMMENDATION**  
**of the EU COUNCIL**  
**on an action in the field of rare diseases**  
**(2009/C 151/02)**  
**8 June 2009**

# EU COUNCIL RECOMMENDATION

“HEREBY RECOMMENDS that Member States:

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.

# EU COUNCIL RECOMMENDATION

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.”

Relevant extracts from the  
**EUCERD CORE INDICATORS for**  
**RD National Plans / Strategies**



# EUCERD CORE INDICATORS

## N°4. Adoption of the EU RD definition

Short Definition =  
Definition as laid down in OD Regulation EC 141/2000 &  
Cross Border Health Care Directive 2011/24/EU:  
"no more than 5 patients per 10 000 persons"

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

Existence of a nation-wide, comprehensive RD specific information system.  
The participation in Orphanet is also included.

# EUCERD CORE INDICATORS

## N°9. Existence of Help lines for RD

Refers to Help Lines for professionals only, for patients only and for both. Supported by private, public funding, or both.

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

Type of coding system(s) used in view of better RD management and ultimately harmonising RD nomenclature

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

Public support for their development and sustainability



**Relevant extracts from the**  
**EUROPLAN RECOMMENDATIONS**  
**for the development of RD National Plans/  
Strategies**



# EUROPLAN RECOMMENDATIONS

- 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

# EUROPLAN RECOMMENDATIONS

- 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 recognising 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.

# EUROPLAN RECOMMENDATIONS

- 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.
- R 5.1 The use of international global information websites and data repositories for rare diseases is promoted.
- R 5.15 All information on centres of expertise, good practice guidelines, medical laboratory activities, clinical trials, registries and availability of drugs, collected at national level, is also published on Orphanet.

**Relevant extracts from the**  
**EUROPLAN INDICATORS**  
**for the development of RD National Plans/  
Strategies**



# 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>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Yes</li> <li><input type="checkbox"/> No</li> <li><input type="checkbox"/> EU definition modified with an additional definition</li> </ul>
<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>	<ul style="list-style-type: none"> <li><input type="checkbox"/> ICD-9</li> <li><input type="checkbox"/> ICD-10</li> <li><input type="checkbox"/> OMIM</li> <li><input type="checkbox"/> SNOMED</li> <li><input type="checkbox"/> ORPHAN</li> <li><input type="checkbox"/> MESH</li> <li><input type="checkbox"/> Others</li> </ul>

# 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><b>Process</b></p>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Not existing, not clearly stated</li> <li><input type="checkbox"/> Existing, clearly stated, partly implemented and enforced</li> <li><input type="checkbox"/> Existing, clearly stated and substantially implemented and enforced</li> </ul>
<p>Defining a surveillance system based on a patient outcomes registry</p>	<p>Registering activity</p>	<p><b>Process</b></p>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Centralised RD registry</li> <li><input type="checkbox"/> Multiple RD registries but well coordinated and standardised</li> <li><input type="checkbox"/> Multiple RD registries not standardised</li> <li><input type="checkbox"/> No registry at all</li> </ul>
	<p>Number of diseases included</p>	<p><b>Outcome</b></p>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Number ranging from 1 to 20</li> </ul>