EURORDIS
RARE DISEASES EUROPE

CENTRES OF EXPERTISE AND EUROPEAN REFERENCE NETWORKS FOR RARE DISEASES

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- 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:

11. Identify appropriate centres of expertise throughout their national territory by the end of 2013, and consider supporting their creation.
12. Foster the participation of centres of expertise in European reference networks respecting the national competences and rules with regard to their authorisation or recognition.
13. Organise healthcare pathways for patients suffering from rare diseases through the establishment of cooperation with relevant experts and exchange of professionals and expertise within the country or from abroad when necessary.
14. Support the use of information and communication technologies such as telemedicine where it is necessary to ensure distant access to the specific healthcare needed.

EU COUNCIL RECOMMENDATION

15. Include, in their plans or strategies, the necessary conditions for the diffusion and mobility of expertise and knowledge in order to facilitate the treatment of patients in their proximity.

16. Encourage centres of expertise to be based on a multidisciplinary approach to care when addressing rare diseases.

17. Gather national expertise on rare diseases and support the pooling of that expertise with European counterparts in order to support:

    (a) the sharing of best practices on diagnostic tools and medical care as well as education and social care in the field of rare diseases;

    (b) adequate education and training for all health professionals to make them aware of the existence of these diseases and of resources available for their care;

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EU COUNCIL RECOMMENDATION

(c) the development of medical training in fields relevant to the diagnosis and management of rare diseases, such as genetics, immunology, neurology, oncology or paediatrics;

(d) the development of European guidelines on diagnostic tests or population screening, while respecting national decisions and competences;

(e) the sharing Member States′ assessment reports on the therapeutic or clinical added value of orphan drugs at Community level where the relevant knowledge and expertise is gathered, in order to minimise delays in access to orphan drugs for rare disease patients.”

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

EUCERD
European Union Committee of Experts on Rare Diseases

EUCERD CORE INDICATORS

N°5. Existence of a national policy for establishing Centres of Expertise (CE) on RD

Short definition =
Existence of a strategy to identify and designate RD CEs at national/regional level

N°6. Number of national and regional Centres of Expertise adhering to the national policy

N° of CEs complying with national policy and n° CEs in line with EUCERD Recommendations on Quality Criteria for CEs that help MS designate their CEs

EUCERD CORE INDICATORS

N°7. Participation of national or regional Centres of Expertise in European Reference Networks (ERNs)

Indicator counts the CEs integrated in ERNs, differentiating between full and associated members

N°10. Existence of a national policy on rare disease clinical practice guidelines development and implementation

Whether a national policy exists on developing, adapting and implementing clinical practice guidelines with the aim to ensure equal access to care for all RD patients

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

EUROPLAN
European Project for Rare Diseases National Plans Development

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 3.2 Specific provisions are included in the National Plans or Strategies to promote appropriate collaborations between Centres of Expertise and/or other structures of the health system and health and research authorities in order to improve knowledge on different aspects of rare diseases.
- R 4.1 Well defined mechanisms of designation of centres of expertise are established and their quality is assured, efficiency and long term sustainability.
- R 4.2 Healthcare pathways are defined and adopted, based on best practices and expertise at national and international level.

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EUROPLAN RECOMMENDATIONS

- R 4.3 Cross-border healthcare should be promoted, where appropriate. In that case, centres able to provide quality diagnosis and care are identified in neighbouring or other countries, where patients or biological samples can be referred to, and cooperation and networking is promoted.
- R 4.4 A national directory of Centres of expertise is compiled and made publicly available.
- R 4.5 Travelling of biological samples, radiologic images, other diagnostic materials, and e-tools for tele-expertise are promoted.
- R 4.6 Centres of expertise provide proper training to paramedical specialists; paramedical good practices are coordinated, in order to serve the specific rehabilitation needs of rare diseases patients.
- R 4.7 A national framework is ensured on rare diseases screening options and policies.

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EUROPLAN RECOMMENDATIONS

- R 4.8 Proper performance of newborn screenings prescribed in the country is monitored with appropriate indicators.
- R 4.9 Accessibility to genetic counselling is promoted.
- R 4.10 The quality of genetic testing and other diagnostic tests is ensured, including participation in external quality control schemes at national and international level.
- R 4.11 A national inventory of medical laboratories providing testing for rare disease is compiled and made publicly available.
- R 4.12 The adoption of an ad hoc coding is promoted, when appropriate, to recognize and appropriately resource and reimburse the special rehabilitation treatments necessary for rare diseases.

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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.
- R 5.3 Information on how to establish or join a European Reference Network is made available for health professionals.
- 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.

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EUROPLAN RECOMMENDATIONS

- R 5.6 Continuing education programmes on rare diseases are made available for health professionals.
- R 5.7 The exchange and sharing of expertise and knowledge between centres within the country and abroad is promoted.
- R 5.8 Collaboration is ensured in the European evaluation of the existing screening programmes.
- R 5.9 The development and adoption of good practice guidelines for rare diseases is promoted. The guidelines are made publicly available and disseminated as of the reach targeted health professionals.
- R 5.10 Dissemination of the information about treatment for rare diseases is ensured in the most effective way, to avoid delays of treatment accessibility.

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EUROPLAN RECOMMENDATIONS

- R 5.11 Participation is ensured in common mechanisms, when available, defining conditions for the off-label use of approved medicinal products for application to rare diseases; for facilitating the use of drugs still under clinical trial; for compassionate provision of orphan drugs.
- R 5.12 An inventory of orphan drugs accessible at national level, including reimbursement status, is compiled and made publicly available.
- R 5.13 Patients' access to authorised treatment for rare disease including reimbursement status, is recorded at national and/or EU level.
- R 5.14 The list of ongoing clinical trials on Orphan Medicinal Products included in the European database for clinical trials on Orphan Medicinal Products (EUDRA) is made public at national level.
- 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
European Project for Rare Diseases National Plans Development

EUROPLAN INDICATORS

ACTIONS	INDICATORS	TYPE	ANSWERS
Improve the quality of health care by defining: - appropriate centres with experience on RD - pathways that reduce the diagnosis delay and facilitate the best care and treatments	Existence of a policy for establishing centres of expertise at the national/regional level	Process	□ Not existing, not clearly stated □ Existing, clearly stated, partly implemented □ Existing, clearly stated and substantially implemented
	Number of centres of expertise adhering to the policy defined in the country	Outcome	Number of reference centres
	Groups of rare diseases followed up in centres of expertise	Outcome	Computation must be referred to the whole country Covering all or most of rare diseases Covering only some rare diseases

EUROPLAN INDICATORS

ACTIONS	INDICATORS	TYPE	ANSWERS
Improve the quality of health care by defining: -appropriate centres with experience on RD	Centres of expertise adhering to the standards defined by the Council Recommendations - paragraph d) of preamble	Outcome	Percentage of centers of expertise adhered by the total of centers of expertise designed
- pathways that reduce the diagnosis delay and facilitate the best care and treatments	Participation of national or regional centres of expertise into European Reference Networks	Outcome	Index based on number of centres of expertise cooperating with European Reference Networks by number of total of centres of expertise designed

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EUROPLAN INDICATORS

ACTIONS	INDICATORS	TYPE	ANSWERS
Ensure quality of RD diagnosis laboratory	Existence of a public directory/ies of both genetic tests on Rare Diseases	Process	☐ Yes ☐ No ☐ Under discussion
	Proportion of laboratories having at least one diagnostic test validated by an external quality control	Outcome	Number of validated RD laboratories divided by the total number of RD laboratories

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EUROPLAN INDICATORS

ACTIONS	INDICATORS	TYPE	ANSWERS
Existence of information sites for professionals provided by the plan/strategy	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 ☐ No formal decisions have been taken
	Help lines for professionals	Process	☐ Yes, covers most RD ☐ Yes, covers only some RD ☐ No formal decisions have been taken
	Clinical guidelines	Outcome	Number ranging between 0 to 30
Promoting training activities & awareness educational campaigns among professionals	Number of such as activities (*training & awareness educational*) promoted by the plan/strategy	Outcome	Number ranging between 0 to 30