EUROPLAN
European Project for Rare Diseases National Plans Development

Recommendations for the Development of National Plans or Strategies for Rare Diseases

Domenica Taruscio

EUROPLAN Leader
National Centre for Rare Diseases
Italian National Institute of Health, Rome

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AREAS OF THE COUNCIL AND EUROPLAN RECOMMENDATIONS

1. PLANS AND STRATEGIES IN THE FIELD OF RARE DISEASES
2. ADEQUATE DEFINITION, CODIFICATION AND INVENTORYING
3. RESEARCH ON RARE DISEASES
4. CENTRES OF EXPERTISE AND EUROPEAN REFERENCE NETWORKS FOR RARE DISEASES
5. GATHERING THE EXPERTISE ON RARE DISEASES AT EUROPEAN LEVEL
6. EMPOWERMENT OF PATIENT ORGANISATIONS
7. SUSTAINABILITY

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EUROPLAN Recommendations

reflect the EU Council Recommendation structure:

AREA 1. Plans and strategies in the field of rare diseases

AREA 2. Adequate definition, codification and inventorying

AREA 3. Research on rare diseases

AREA 4. Centres of expertise and european reference networks for rare diseases

AREA 5. Gathering the expertise on rare diseases at european level

AREA 6. Empowerment of patient organisations

AREA 7. Sustainability

Primary prevention of congenital anomalies (EUROCAT/EUROPLAN Recommendations to be approved by EUCERD)

EUROPLAN indicators follow the same structure

Recommendations and indicators

EUROPLAN Recommendations

AREA 1. Plans and strategies in the field of rare diseases

AREA 2. Adequate definition, codification and inventorying

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AREA 5. Gathering the expertise on rare diseases at european level

AREA 6. Empowerment of patient organisations

AREA 7. Sustainability

EUROPLAN Indicators

follow same organization

AREA 1. Plans..

AREA 2. Definition…

AREA 3. Research…

AREA 4. CoE and ERN

AREA 5. Gathering …

AREA 6.Empowerment

AREA 7. Sustainability

EUROPLAN DEFINITION OF A NATIONAL PLAN OR STRATEGY

a set of integrated and comprehensive health and social policy actions for rare diseases, to be developed and implemented at national level, characterized by identified objectives to be achieved within a specified timeframe.

The allocation of appropriate **resources** (human, financial, infrastructural)and its **monitoring and evaluation** are of special value to ensure the efficacy of the plan or strategy.

Council Recommendation (2009/C 151/02)

1. Establish and implement plans or strategies for rare diseases at the appropriate level or explore appropriate measures for rare diseases in other public health strategies, in order to aim to ensure that patients with rare diseases have access to high-quality care, including diagnostics, treatments, habilitation for those living with the disease and, if possible, effective orphan drugs, and in particular:

(a) 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;

b) take action to integrate current and future initiatives at local, regional and national levels into their plans or strategies for a comprehensive approach;

(c) define a limited number of priority actions within their plans or strategies, with objectives and follow-up mechanisms;

(d) take note of the development of guidelines and recommendations for the elaboration of national action for rare diseases by relevant authorities at national level in the framework of the ongoing European project for rare diseases national plans development (EUROPLAN) selected for funding over the period 2008-2011 in the first programme of Community action in the field of public health

EUROPLAN Recommendations on Area 1: Plans or Strategies in the field of rare diseases

R 1.1 Patients with rare diseases deserve dedicated public health policies to meet their specific needs.

R 1.2 Initiatives are taken to raise awareness about the dimension of the problem and to create joint responsibility.

R 1.3 **A mechanism (e.g. interdisciplinary panel, committee)** including relevant stakeholders is established to assist the development and implementation of the National Plan or Strategy.

R 1.4 A **situation analysis** is carried out including:

- ❑ **An inventory of existing healthcare resources, services, clinical and basic research activities and policies** directly addressing rare diseases as well as those from which rare disease patients may benefit.
- ❑ **Unfulfilled needs** of patients are assessed.
- ❑ **Available resources** for improving health and social care of people affected by rare diseases at national level are evaluated.
- ❑ **European collaboration and the European documents** in the field of rare diseases are taken into account in the development of the National Plan or Strategy.

EUROPLAN Recommendations on Area 1: Plans or Strategies in the field of rare diseases

R 1.5 The National Plan or Strategy is elaborated with well described objectives and actions. The general objectives of a National Plan or Strategy are based on the general overarching values of universality, access to good quality care, equity and solidarity.

R 1.6 The policy decisions of the National Plan or Strategy are integrated i.e. structured maximizing synergies and avoiding duplications with existing functions and structures of the health care system of the country.

R 1.7 The policy decisions of the National Plan or Strategy are comprehensive, addressing not only health care needs, but also social needs.

R 1.8 Specific areas for action are indicated, with priority given to those of the Council Recommendations, taking into account the major needs identified in the member state.

R 1.9 Appropriate resources are allocated to ensure the feasibility of the actions in the planned time.

R 1.10 Information on the National Plan or Strategy is made accessible to the public and it is disseminated to patients' groups, health professionals' societies, general public and media, making the plan known also at European level.

EUROPLAN Recommendations on Area 1: Plans or Strategies in the field of rare diseases

R 1.11 **Measures are taken to ensure the sustainability**, transfer and integration of the actions foreseen by the national plan or strategy into the general health system of the country.

R 1.12 **The National Plan or Strategy has a duration of three to five years**. An intermediate deadline is established, after which, an evaluation process is undertaken and corrective measures are adopted. For longer time scales or no defined time frame, a 2- to 3-year cyclic evaluation and adaptation process is adopted, if needed.

R 1.13 **The National Plan or Strategy is monitored** and assessed at regular intervals using, as far as possible, **EUROPLAN indicators**.

R 1.14 **The implementation of the actions and their achievements are assessed**.

R 1.15 **The most appropriate evaluation of a National Plan or Strategy is by an external body** and takes into account also patients' and citizens' views. Patients needs are assessed at the beginning and the end of the plan implementation using the same methodology. Evaluation Reports are made public.

Council Recommendation (2009/C 151/02)

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.

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.

EUROPLAN Recommendations on area 2:
Adequate definition, coding and inventorying of 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.

Council Recommendation (2009/C 151/02)

6. Identify ongoing research and research resources in the national and Community frameworks in order to establish the state of the art, assess the research landscape in the area of rare diseases, and improve the coordination of Community, national and regional programmes for rare diseases research.

7. Identify needs and priorities for basic, clinical, translational and social research in the field of rare diseases and modes of fostering them, and promote interdisciplinary cooperative approaches to be complementarily addressed through national and Community programmes.

8. Foster the participation of national researchers in research projects on rare diseases funded at all appropriate levels, including the Community level.

9. Include in their plans or strategies provisions aimed at fostering research in the field of rare diseases.

10. Facilitate, together with the Commission, the development of research cooperation with third countries active in research on rare diseases and more generally with regard to the exchange of information and the sharing of expertise.

EUROPLAN Recommendations on Area 3: Research on Rare diseases

R 3.1 Dedicated national research programs for rare diseases (basic, translational, clinical, public health and social research) are established and supported with dedicated funds,preferably for a long period. Research projects on rare diseases should be made identifiable and traceable within broader national research programs.

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 3.3 National networks are promoted to foster research on rare diseases. Special attention is given to clinical and translational research in order to facilitate the application of new knowledge into rare disease treatment. Compilation and updating of a directory of teams carrying out research on rare diseases should be endorsed when feasible.

R 3.4 Proper initiatives are developed to foster participation in cooperative international research initiatives on rare diseases, including the EU framework program and E-RARE. The national funding of these initiatives should be increased considerably.

EUROPLAN Recommendations on Area 3: Research on Rare diseases

R 3.5 Specific technological platforms and infrastructures for rare disease research, including clinical research, are established and supported and the creation of public-private partnership is explored.

R 3.6 Multi-centre national and trans-national studies are promoted, in order to reach a critical mass of patients for clinical trials and to exploit international expertise.

R 3.7 Specific programs are launched for funding and/or recruitment of young scientists on rare diseases research projects.

R 3.8 The assessment of already existing drugs in new combinations and in new indications is supported since it may be a cost-effective way to improve treatment for patients with rare diseases.

Council Recommendation (2009/C 151/02)

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.

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.

EUROPLAN recommendations on Area 4:

Centres of Expertise and European Reference Networks for 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.

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.

EUROPLAN recommendations on Area 4:

Centres of Expertise and European Reference Networks for rare diseases.

R 4.7 A national framework is ensured on rare diseases screening options and policies.

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 iseases.

Council Recommendation (2009/C 151/02)

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;

(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.

EUROPLAN Recommendations on Area 5:
Gathering the expertise on rare diseases at European level

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 to 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.

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.

EUROPLAN Recommendations on Area 5:
Gathering the expertise on rare diseases at European level

R 5.8 Collaboration is ensured in the European evaluation of the existing screening programs.

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.

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.

EUROPLAN Recommendations on Area 5:
Gathering the expertise on rare diseases at European level

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 on-going 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 as planned in the Joint Action.

6. EMPOWERMENT OF PATIENT ORGANISATIONS

Council Recommendation (2009/C 151/02)	
18. Consult patients and patients' representatives on the policies in the field of rare diseases and facilitate patient access to updated information on rare diseases.	*patient organisations, such as awareness-raising, capacity-building and training, exchange of information and best practices, networking and outreach to very isolated patients.*
19. Promote the activities performed by	

EUROPLAN Recommendations on Area 6: Empowerment of patients' organisations

R 6.1 Advocacy of patients' needs by patients' associations is recognised as an important element in defining policies on rare diseases; the organisation of a national umbrella organisation that represents the interests of all rare diseases patients is encouraged.

R 6.2 The patients' organisations are involved in decisions making processes in the field of rare diseases.

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 6.5 Specialised social services are supported for people living with a chronically debilitating rare disease and their family carers.

R 6.6 Specialised social services are established to facilitate integration of patients at schools and workplaces.

EUROPLAN Recommendations on Area 6: Empowerment of patients' organisations

R 6.7 A directory of centres providing specialised social services, including those offered by patients' associations, is compiled, kept updated and communicated to national, regional and patients' websites and included in the Rapsody network.

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.)

R 6.10 The activities aiming at patients' empowerment carried out by patients' associations are facilitated.

7. SUSTAINABILITY

Council Recommendation (2009/C 151/02)

20. Together with the Commission, aim to ensure, through appropriate funding and cooperation mechanisms, the long-term sustainability of infrastructures developed in the field of information, research and healthcare for rare diseases.

EUROPLAN Recommendations on Area 7: Sustainability

R 7.1 The National Plan or Strategy on rare diseases is supported combining national (regular and ad hoc) and European funds, according to the country health system and decision-making processes.

R 7.2 Possibilities for European funding are exploited for those parts of National Plans or Strategies which are in the scope of the European Social Fund and European Regional Development Fund.

R 7.3 The cooperation with other member states is envisaged when cross-border health care is needed, in order to address the need for sustainability of common European infrastructures, share costs and maximise the efficacy of initiatives.

R 7.4 Participation in the debate on enhanced EU governance is ensured, in order to find agreed and improved mechanisms for the governance of the healthcare, information and research initiatives requiring transnational collaboration.

R 7.5 Agreements for coordinated projects, including long-term sustainability of common infrastructures, are pursued.