



# **EUROPLAN PROJECT NATIONAL CONFERENCE ON RARE DISEASES**

## **WORKSHOP**

### **STANDARDS OF CARE FOR RARE DISEASES**

### **CENTRES OF EXPERTISE / EUROPEAN REFERENCE NETWORKS/ ORPHAN DRUGS AND PROVISION OF TREATMENTS**



**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:**

- **Identify appropriate centres of expertise throughout their national territory by the end of 2013, and consider supporting their creation.**
- **Foster the participation of centres of expertise in European reference networks respecting the national competences and rules with regard to their authorisation or recognition.**
- **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.**

# COUNCIL RECOMMENDATIONS

- **Support the use of information and communication technologies such as telemedicine where it is necessary to ensure distant access to the specific healthcare needed.**
- **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.**
- **Encourage centres of expertise to be based on a multidisciplinary approach to care when addressing rare diseases.”**



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

# EUROPLAN RECOMMENDATIONS

- Transparent mechanisms of designation of centres of expertise are established ensuring their quality, efficiency and long term sustainability
- Healthcare pathways are defined and adopted, based on best practices and expertise at national and international level
- A national directory of Centres of expertise operating in the national territory is compiled, kept updated and published at national level as well as in a common European Inventory (Orphanet)
- 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 networking is promoted
- Travelling of biological samples, radiologic images, other diagnostic materials, and e-tools for tele-expertise is supported

# EUROPLAN RECOMMENDATIONS

- A national framework is ensured on rare diseases screening options and policies
- Proper performance of newborn screenings prescribed in the country is monitored with appropriate indicators
- Accessibility to genetic counselling is promoted
- The quality of genetic testing and other diagnostic tests is ensured, including participation in external quality control schemes at national and international level
- Collaboration with the EU inventory of the national provision of testing for rare diseases (genetic, biochemical, enzymatic, tissue/cell identification, clinical) published both at national level and on Orpha.net is ensured

# EUROPLAN RECOMMENDATIONS

- Centers 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
- An ad hoc coding must be incorporated into the national health system to recognise and appropriately resource the special rehabilitation treatments necessary for people affected by rare diseases



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

# EUROPLAN INDICATORS FOR CENTRES OF EXPERTISE

ACTIONS	INDICATORS	TYPE	ANSWERS
<p>Improve the quality of health care by defining:</p> <ul style="list-style-type: none"> <li>-appropriate centres with experience on RD</li> <li>- pathways that reduce the diagnosis delay and facilitate the best care and treatments</li> </ul>	<p>Existence of a policy for establishing centres of expertise at the national/regional level</p>	<p><b>Process</b></p>	<p>Not existing, not clearly stated Existing, clearly stated, partly implemented  Existing, clearly stated and substantially implemented</p>
	<p>Number of centres of expertise adhering to the policy defined in the country</p>	<p><b>Outcomes</b></p>	<p>Number of reference centres</p>
	<p>Groups of rare diseases followed up in centres of expertise</p>	<p><b>Outcomes</b></p>	<p>Computation must be referred to the whole country  Covering all or most of rare diseases  Covering only some rare diseases</p>

# EUROPLAN INDICATORS FOR CENTRES OF EXPERTISE

ACTIONS	INDICATORS	TYPE	ANSWERS
<p><b>Improve the quality of health care by defining:</b></p> <ul style="list-style-type: none"> <li>-appropriate centres with experience on RD</li> </ul>	<p>Centres of expertise adhering to the standards defined by the Council Recommendations - paragraph d) of preamble</p>	<p><b>Outcomes</b></p>	<p>Percentage of centers of expertise adhered by the total of centers of expertise designed</p>
<ul style="list-style-type: none"> <li>- pathways that reduce the diagnosis delay and facilitate the best care and treatments</li> </ul>	<p>Participation of national or regional centres of expertise into European reference networks</p>	<p><b>Outcomes</b></p>	<p>Index based on number of centres of expertise cooperating with European Reference Networks by number of total of centres of expertise designed</p>

# EUROPLAN INDICATORS FOR DIAGNOSIS

ACTIONS	INDICATORS	TYPE	ANSWERS
Develop Screening Policies	Number of diseases included in the neonatal screening programme	Outcomes	Number of diseases
	Number of diseases included in the neonatal screening programme properly assessed	Outcomes	Index based on the number of disease tests assessed and included in the neonatal screening programme divided by the total number of diseases included in the neonatal screening programme
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	Outcomes	Number of validated RD laboratories divided by the total number of RD laboratories

# EUROPLAN INDICATORS FOR ACCESS TO ORPHAN DESIGNATED DRUGS (ODD)

ACTIONS	INDICATORS	TYPE	ANSWERS
<p>Ensure the mechanism that facilitates ODD access and the reimbursement of their cost to patients after they got the Market Authorisation by EMA</p>	<p>Number of ODD market authorisations by EMA and placed in the market in the country</p>	<p>Outcomes</p>	<p>Index based on Number of ODD placed in the market by total of ODD approved by the EMA</p>
	<p>Time between the date of a ODD market authorisation by EMA and its actual date of placement in the market for the country</p>	<p>Outcomes</p>	<p>Average days since the date of market authorisation by EMA until the official date of placement in the market in the country</p>
	<p>Time from the placement in the market in the country to the positive decision for reimbursement by public funds</p>	<p>Outcomes</p>	<p>Average days since the date of placement in the market until the reimbursement decision date in the country</p>
	<p>Number of ODD reimbursed 100%</p>	<p>Outcomes</p>	<p>Number ranging 0 to 1,000</p>

# EUROPLAN INDICATORS FOR ACCESS TO ORPHAN DESIGNATED DRUGS (ODD)

ACTIONS	INDICATORS	TYPE	ANSWERS
To develop mechanisms to accelerate ODD availability	Existence of a governmental programme for compassionate use for Rare Diseases	Outcomes	No Yes In process



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**FURTHER GUIDELINES FOR DISCUSSION**

# IDENTIFICATION OF NATIONAL / REGIONAL CoE ALL THROUGH THE NATIONAL TERRITORY BY 2013

- How to ensure that all patient living with a rare disease have access to a CoE in your country or abroad, and support the CoE creation where necessary?
- How to best apply in your country the criteria identified in the report of the EC Rare Disease Task Force, which will possibly become part of the EC Cross-Border Healthcare Directive, art.15, for the designation of CoE?
- In particular, how to make sure that CoE are –as much as possible- expert of the specific RD in both the clinical and the research field?
- What are the best structures and solutions suitable to be a CoE in your country? Do they depend on the disease (or group of diseases)? Are they regional or national?

# SUSTAINABILITY OF CENTRES OF EXPERTISE

- How to ensure, through appropriate funding mechanisms, the long-term sustainability of healthcare infrastructures, in particular Centres of Expertise?

# PARTICIPATION IN EUROPEAN REFERENCE NETWORKS

- How to foster the participation of Centres of Expertise to European Reference Networks?
- How to support the mobility of patients and/or professionals beyond the national borders?

# HOW TO SHORTEN THE ROUTE TO DIAGNOSIS

- How to support the mobility of expertise and knowledge to facilitate the treatment of patients in their proximity (including mobility of information through ICT)?
- How to map, network and support the laboratories at national level?
- Specific area: Screening and genetic testing
  - How to organise DNA and samples exchanges and reimbursement at European and international level?
  - How to support the development of European guidelines on diagnostic tests and population screening?
  - What mechanisms to develop in order to support common protocols and recommendations such as European reference opinions on diagnostic tools, medical care, education and social care?

# HOW TO OFFER SUITABLE CARE & ORGANISE HEALTH CARE PATHWAYS

- How to develop a system based on the adoption of clinical pathways for the provision of care for RD patients: provide funding for multidisciplinary consultations, cover patient and families transportation costs?
- How to link medical expertise of the specialised centres to local medical, paramedical and social care?
- How to establish good cooperation with relevant experts within the country or from abroad when necessary through European reference networks, with the aim to adopt common healthcare pathways based on the best evidence and expertise?

# MULTIDISCIPLINARY APPROACHES

- How to ensure multidisciplinary approaches in Centres of Expertise?
- How to ensure integration between medical and social levels?
  - Specific area: Social counselling

# EVALUATION OF CENTRES OF EXPERTISE

- How to envisage a system for the evaluation of Centres of Expertise?
- Would it be based on clinical outcomes or patient satisfaction, or both?

# ACCESS OF RD PATIENTS TO ORPHAN DRUGS PRICING AND REIMBURSEMENT

- How to improve and speed up national procedures for pricing and reimbursement of OD?
- In particular, what mechanisms to put in place to use the “clinical added value of orphan drugs” report developed at the EU level (EMA) to base the national decision on pricing and reimbursement in order to minimise delays in access to OD?
- How to promote a national policy on conditional pricing and reimbursement, based on the EU Pharma Recommendation “Improving access to orphan drugs”?
- Access to orphan drugs through Centres of Expertise
- Participating to the EU-level collaboration on the assessment of the clinical added value of orphan drugs at the EMEA

# COMPASSIONATE USE AND TEMPORARY APPROVAL OF ORPHAN DRUGS

- How to foster access to OD through compassionate use ?
- Can drugs be prescribed off label and reimbursed when the evidence of a benefit for the patients exists?
- What measures can be studied and put in place to provide treatments other than medicinal products when the evidence of a benefit for the patient exists?

# FUTURE OF ORPHAN DRUGS

- Number of OD on the market and number of patients treated.
- Obstacles to the availability of OD approved in the EU within the timeframe requested by the regulations (180 days).



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**PROPOSALS FROM THE AUDIENCE**