

## EURORDIS - The European Organisation for Rare Diseases, France

1. Respondent Profile	
1.1 Please indicate the type of organisation on behalf of which you are responding to this consultation:	Patient organisation/association
Please indicate level:	European Union umbrella organisation
Please indicate Member States representation:	Pan European
Please indicate for what the administration is responsible:	
1.1.1. Other (please specify):	EURORDIS represents the voice of an estimated 30 million citizens affected by a RD in the EU, covering overall more than 4000 RDs. EURORDIS currently has 544 members, 477 of which are from 24 EU countries, 2 EFTA countries (Iceland and Norway), one accessing country (Croatia) and 42 other countries around the world. EURORDIS includes 32 National Alliances of Rare Diseases and 36 disease-specific European Federations.
1.2 Please indicate the name of your organisation or centre:	EURORDIS - The European Organisation for Rare Diseases
1.3 Please indicate the country where your organisation/centre is located/has its headquarters or main representative office in Europe:	FR
1.4 Please indicate the number of EU Member States and EEA countries (Norway, Iceland, Lichtenstein) and accessing country (Croatia) in which your organisation conducts business/is represented:	27
1.5 If need be, can we contact you by e-mail to obtain further information on your submission?	Yes
1.5.1 Please provide an e-mail address where we can contact you:	yann.lecam@eurordis.org

1.6 Please provide us with a contact person (incl jobtitle and daytime phone number):	Yann Le Cam, Chief Executive Officer and Vice-Chair of the European Union Committee of Experts on Rare Diseases - Tel: + 33 1 56535211
1.7 Please provide additional contact details if needed:	Flaminia Macchia, European Public Affairs Director flaminia.macchia@eurordis.org Ariane Weinman, European Public Affairs Manager ariane.weinman@eurordis.org

## 2. Involvement of your organisation in the matter of centres of excellence/reference (COE) and healthcare networks in highly specialised healthcare (HSHC).

2.1 How would you describe your organisation's knowledge of CoE and HSHC?	Very high
2.1.1 Space for further comments:	Participation in HLG on Health Services & Medical Care & its report on agreed principles on ERN; recommendations of RDTF/EUCERD; shaping of the CBHC Directive; workshop on national measures EUROPLAN
2.2. What aspects or domains related to the topic of CoE and HSHC would correspond to your organisation's key knowledge? (cross any that applies)	<ul style="list-style-type: none"> <li>Priorities, description and characteristics of CoE and HSHC</li> <li>Ethical analysis</li> <li>Management and organisational aspects of highly specialized healthcare</li> <li>Social aspects</li> <li>Professional performance, clinical practice, quality and safety of specialized healthcare</li> <li>Assessment/evaluation/certification of clinical practice and healthcare providers</li> </ul>

2.2.1. Space for further comments:	Priorities&characteristics CoE;EUnetHTA Stakeholders Forum;EUPATI;Summer School drug development&Regulatory processes;EMA Scient.Committees; analysis of ethical issues&specialised Social Services JARD
2.3 Is highly specialised healthcare a priority in your organisation's strategies and work plans?	Very high
2.3.1 Space for further comments:	Both in the Strategy 2019-2015 and in all the annual EURORDIS workplans. Participation in EU Public Health Policy projects and supporting national policy, capacity-building activities, inforamtion.
2.4. What specific field of healthcare services/specialities are most relevant for your centre/organisation's field of work?	Laboratories and central services (e.g. pathology, genetics, biochemistry, pharmacy, microbiology etc.)
Please specify:	geneticmultifactorial need concentration expertise
2.5. Has your organisation/centre been directly involved in the design or assessment of professional standards and criteria related with highly specialised healthcare?	Frequently
2.5.1 Please describe your role in such actions/projects:	Research projects funded by FP5, FP6 & FP7, including pilots reference networks on rare diseases; EURORDIS developed projects under the 1st & 2nd EU Public Health Programmes, including RAPSODY project
2.6. Has your organisation been involved in projects/activities supported by the Commission in relation with HSHC or professional and technical criteria/standards in highly specialised healthcare?	Yes

2.7. Do you have concrete examples based on your own organisation's experience or could you provide us with references or links to documents related with professional criteria and standards in highly specialised healthcare/CoE or HSHC (e.g. quality criteria, guidelines, consensus documents)?	Yes
2.7.1 Space for further comments:	EurordisCare3 on CoE&ERN; declaration of common principles on CoE&ERNS; The Voice of 12000 Patients; Good practices for collaboration between CoE & POs; Policy Fact Sheets & Position Paper on CoE&ERNS
2.13. What is the scope of the network?	
2.14. Which kind of network?	
2.14.1 Space for further comments:	
2.15. Would you be interested in applying to the process to be considered Centre of Excellence of the future European Reference Network? (1 = not interested at all, 5 = very interested)	
2.15.1 Space for further comments:	

### 3. Proposed criteria for ERN (scope, general and specific criteria)

#### 3.1 Criteria related with diseases or conditions in order to be considered under the scope of the ERN

3.1.1. Need of highly specialised healthcare	4
3.1.1.1. Complexity of the diagnosis and treatment	5
3.1.1.2. High cost of treatment and resources	3
3.1.1.3. Need of advanced/highly specialised medical equipment or infrastructures	4
3.1.2. Need of particular concentration of expertise and resources	5

3.1.2.1. Rare expertise/need of concentration of cases	5
3.1.2.2. Low prevalence/incidence/number of cases	5
3.1.2.3. Evaluated experiences of Member States	3
3.1.3. Based on high-quality, accessible and cost-effective healthcare	4
3.1.3.1. Evidence of the safety and favourable risk-benefit analysis	2
3.1.3.2. Feasibility and evidence of the value and potential positive outcome (clinical)	5
3.1.4. Do you recommend any additional criteria or option that would effectively address the issue?	Yes
3.1.4.1 Explain your proposal in free text:	Need for advanced/highly specialised HC professionals. Highly specialised professionals in the field of the disease or group of diseases concerned are necessary to gather scarce expertise within ERNs
3.1.5. Would you prioritise or suggest any concrete disease or group of diseases to be addressed by the future ERN according to the above criteria?	Yes
3.1.5.1 Explain your proposal in free text:	ERNs system organised through a step-wise approach aimed at establishing limited number of ERNs gathered by therapeutic areas covering in fine all RDs patients and delivering structured HC pathways

### 3.2. General criteria of the centres wishing to join a European Reference Network

3.2.1. Organisation and management	5
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3.2.2. Patients empowerment and centered care	5
3.2.3. Patient care, clinical tools and health technology assessment	5
3.2.4. Quality, patient safety and evaluation framework policies	5
3.2.5. Business continuity, contingency planning and response capacity	5
3.2.6. Information systems, technology and e-health tools and applications	5
3.2.7. Overall framework and capacity for research and training	4
3.2.8. Specific commitment of the management/direction of the centre/hospital to ensure a full and active participation in the ERN	5
3.2.9. Do you recommend any additional option that would effectively address the issue?	Yes
3.2.9.1. Space for further comments:	Involvement of POs & patient representatives in the governance/activities of the CoE; ability to contribute and implement good practice guidelines; ability to collect data for registries

### 3.3. Specific criteria regarding the areas of expertise

3.3.1. Competence, experience and good outcomes and care	4
3.3.2. Specific resources and organisation:	5
3.3.2.1. Human resources	5
3.3.2.2. Team/centre organisation	4

3.3.2.3. Structural conditions	4
3.3.2.4. Specific equipment	5
3.3.2.5. Presence and coordination with other required complementary units or services	5
3.3.3. Patient care pathways, protocols and clinical guidelines in the field of expertise	5
3.3.4. External coordination, care management and follow-up of patients	5
3.3.5. Research, training, health technology assessment in the field of expertise	3
3.3.6. Specific information systems	4
3.3.7. Do you recommend any additional criteria or option that would effectively address the issue?	Yes
3.3.7.1. Space for further comments:	Overall multi-disciplinarity, including clinical and social/supportive care, flexibility of the structure, interoperability, training & information, ability to gather critical mass of patients & data