

## Regional Coordinator Centre for Rare Diseases of the Friuli Venezia Giulia Region-Italy (RCCRD), University Hospital of Udine, Italy

| <b>1. Respondent Profile</b>   |  |
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| 1.1 Please indicate the type of organisation on behalf of which you are responding to this consultation:   | Academic/public health and healthcare specialised institution/organisation (e.g. Institutes and University Departments of Public Health, Quality, Healthcare, Clinical Excellence) |
| Please indicate level:   | National level organisation  |
| Please indicate for what the administration is responsible:  |  |
| 1.1.1. Other (please specify):   |  |
| 1.2 Please indicate the name of your organisation or centre:   | Regional Coordinator Centre for Rare Diseases of the Friuli Venezia Giulia Region-Italy (RCCRD), University Hospital of Udine, Italy   |
| 1.3 Please indicate the country where your organisation/centre is located/has its headquarters or main representative office in  | IT   |
| 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 | 11   |
| 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  | bemi.bruno@aoud.sanita.fvg.it  |
| 1.6 Please provide us with a contact person (incl jobtitle and daytime phone number):  | Dr. Giulia Liva, secretary responsible: +390-0432-559914   |
| 1.7 Please provide additional contact details if needed:   |  |

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

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| 2.1 How would you describe your organisation's knowledge of CoE and HSHC?  | Very high   |
| 2.1.1 Space for further comments:  | The RCCRD directs the Rare Diseases Regional Registry, represents FVG Region at the technical table for Rare Diseases (RD) at the Ministry of Health, is involved in  |
| 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)                            | Highly specialised healthcare provision<br>Costs and economic evaluation<br>Priorities, description and characteristics of CoE and HSHC<br>Ethical analysis<br>Social aspects<br>Professional performance, clinical practice, quality and safety of specialized healthcare<br>Assessment/evaluation/certification of clinical practice and healthcare providers<br>Information system, coding |
| 2.2.1. Space for further comments:   |   |
| 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:  | The RCCRD provides genetic and biochemical diagnosis for RD at UE level, is involved in research projects on lysosomal diseases (pathophysiology and new  |
| 2.4. What specific field of healthcare services/specialities are most relevant for your centre/organisation's field of work?   | Medical speciality  |
| Please specify:  | Therapy for RD (enzyme, small molecules therapy)  |
| 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:  | The RCCRD is involved in the organization of regional and national network for RD, in Centre of Rreference certification, in epidemiological data management,   |
| 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 | Yes   |

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| 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:  | The RCCRD is involved in an EU projects on RD registry; collaborates with EU Centres on scientific programmes of RDs management;it's Laboratory is part of an EU net for Lysosomal Disorders. |
| 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  |   |
| 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

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

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| 3.1.2.1. Rare expertise/need of concentration of cases  | 5  |
| 3.1.2.2. Low prevalence/incidence/number of cases   | 4  |
| 3.1.2.3. Evaluated experiences of Member States   | 4  |
| 3.1.3. Based on high-quality, accessible and cost-effective healthcare  | 5  |
| 3.1.3.1. Evidence of the safety and favourable risk-benefit analysis  | 5  |
| 3.1.3.2. Feasibility and evidence of the value and potential positive outcome (clinical)  | 4  |
| 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:   | Evidence of scientific activity  |
| 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:   | Lysosomal Storage Disorders for their complexity and high cost of therapies. |

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

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| 3.2.1. Organisation and management                                   | 5 |
| 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 |

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| 3.2.5. Business continuity, contingency planning and response capacity   | 4  |
| 3.2.6. Information systems, technology and e-health tools and applications   | 5  |
| 3.2.7. Overall framework and capacity for research and training  | 5  |
| 3.2.8. Specific commitment of the management/direction of the centre/hospital to ensure a full and active participation in | 4  |
| 3.2.9. Do you recommend any additional option that would effectively address the issue?                                    | No |
| 3.2.9.1. Space for further comments:   |    |

| <b>3.3. Specific criteria regarding the areas of expertise</b>                            |   |
|---|---|
| 3.3.1. Competence, experience and good outcomes and care                                  | 5 |
| 3.3.2. Specific resources and organisation:   | 5 |
| 3.3.2.1. Human resources  | 4 |
| 3.3.2.2. Team/centre organisation   | 5 |
| 3.3.2.3. Structural conditions  | 5 |
| 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 |

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| 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                   | 5   |
| 3.3.6. Specific information systems   | 4   |
| 3.3.7. Do you recommend any additional criteria or option that would effectively address the issue? | No  |
| 3.3.7.1. Space for further comments:  | The RCCRD considers a priority the organization of an UE Registry of RDs as well as the creation of a EU network of Lysosomal Diseases Centre of Excellence |