



## EUROPEAN PLATFORM ON RARE DISEASE REGISTRATION (EU RD Platform)

### SET OF COMMON DATA ELEMENTS FOR RARE DISEASES REGISTRATION

| GROUP                   | ELEMENT N° | ELEMENT NAME                          | ELEMENT DESCRIPTION                           | CODING                                                                                                                 | COMMENT                                                                                                     |
|-------------------------|------------|---------------------------------------|-----------------------------------------------|------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------|
| 1. Pseudonym            | 1.1.       | Pseudonym                             | Patient's pseudonym                           | <ul style="list-style-type: none"> <li>String</li> </ul>                                                               | <a href="https://eu-rd-platform.jrc.ec.europa.eu/spider">https://eu-rd-platform.jrc.ec.europa.eu/spider</a> |
| 2. Personal information | 2.1.       | Date of birth                         | Patient's date of birth                       | <ul style="list-style-type: none"> <li>Date (dd/mm/yyyy)</li> </ul>                                                    |                                                                                                             |
|                         | 2.2.       | Sex                                   | Patient's sex at birth                        | <ul style="list-style-type: none"> <li>Female</li> <li>Male</li> <li>Undetermined</li> <li>Foetus (Unknown)</li> </ul> |                                                                                                             |
| 3. Patient Status       | 3.1.       | Patient's status                      | Patient alive or dead                         | <ul style="list-style-type: none"> <li>Alive</li> <li>Dead</li> <li>Lost in follow-up</li> <li>Opted-out</li> </ul>    | If dead then answer question 3.2                                                                            |
|                         | 3.2.       | Date of death                         | Patient's date of death                       | <ul style="list-style-type: none"> <li>Date (dd/mm/yyyy)</li> </ul>                                                    |                                                                                                             |
| 4. Care pathway         | 4.1.       | First contact with specialised centre | Date of first contact with specialised centre | <ul style="list-style-type: none"> <li>Date (dd/mm/yyyy)</li> </ul>                                                    |                                                                                                             |

|                    |      |                                                 |                                                                                                            |                                                                                                                                        |                                                                                                                           |
|--------------------|------|-------------------------------------------------|------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------|
| 5. Disease history | 5.1. | Age at onset                                    | Age at which symptoms/signs first appeared                                                                 | <ul style="list-style-type: none"> <li>• Antenatal</li> <li>• At birth</li> <li>• Date (dd/mm/yyyy)</li> <li>• Undetermined</li> </ul> |                                                                                                                           |
|                    | 5.2. | Age at diagnosis                                | Age at which diagnosis was made                                                                            | <ul style="list-style-type: none"> <li>• Antenatal</li> <li>• At birth</li> <li>• Date (dd/mm/yyyy)</li> <li>• Undetermined</li> </ul> |                                                                                                                           |
| 6 Diagnosis        | 6.1. | Diagnosis of the rare disease                   | Diagnosis retained by the specialised centre                                                               | Orpha code (strongly recommended – see link) / Alpha code/ ICD-9 code/ ICD-9-CM code / ICD-10 code                                     | <a href="http://www.orphadata.org/cgi-bin/inc/product1.inc.php">http://www.orphadata.org/cgi-bin/inc/product1.inc.php</a> |
|                    | 6.2. | Genetic diagnosis                               | Genetic diagnosis retained by the specialised centre                                                       | International classification of mutations (HGVS) (strongly recommended – see link) / HGNC / OMIM code                                  | <a href="http://www.hgvs.org">http://www.hgvs.org</a>                                                                     |
|                    | 6.3  | Undiagnosed case                                | How the undiagnosed case is defined                                                                        | <ul style="list-style-type: none"> <li>• Phenotype (HPO)</li> <li>• Genotype (HGVS)</li> </ul>                                         |                                                                                                                           |
| 7. Research        | 7.1. | Agreement to be contacted for research purposes | Patient's permission exists for being contacted for research purposes                                      | <ul style="list-style-type: none"> <li>• YES</li> <li>• NO</li> </ul>                                                                  |                                                                                                                           |
|                    | 7.2. | Consent to the reuse of data                    | Patient's consent exists for his/her data to be reused for other research purposes                         | <ul style="list-style-type: none"> <li>• YES</li> <li>• NO</li> </ul>                                                                  |                                                                                                                           |
|                    | 7.3. | Biological sample                               | Patient's biological sample available for research                                                         | <ul style="list-style-type: none"> <li>• YES</li> <li>• NO</li> </ul>                                                                  | If YES answer question 7.4                                                                                                |
|                    | 7.4. | Link to a biobank                               | Biological sample stored in a biobank                                                                      | <ul style="list-style-type: none"> <li>• YES (if appropriate use link)</li> <li>• NO</li> </ul>                                        | <a href="https://directory.bbmri-eric.eu">https://directory.bbmri-eric.eu</a>                                             |
| 8.Disability       | 8.1. | Classification of functioning/disability        | Patient's disability profile according to International Classification of Functioning and Disability (ICF) | <ul style="list-style-type: none"> <li>• Disability profile / Score</li> </ul>                                                         | <a href="http://www.who.int/classifications/icf/whodasii/en/">http://www.who.int/classifications/icf/whodasii/en/</a>     |