



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

5. Disease history	5.1.	Age at onset	Age at which symptoms/signs first appeared	<ul style="list-style-type: none"> • Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined 	
	5.2.	Age at diagnosis	Age at which diagnosis was made	<ul style="list-style-type: none"> • Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined 	
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	http://www.orphadata.org/cgi-bin/inc/product1.inc.php
	6.2.	Genetic diagnosis	Genetic diagnosis retained by the specialised centre	International classification of mutations (HGVS) (strongly recommended – see link) / HGNC / OMIM code	http://www.hgvs.org
	6.3	Undiagnosed case	How the undiagnosed case is defined	<ul style="list-style-type: none"> • Phenotype (HPO) • Genotype (HGVS) 	
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"> • YES • NO 	
	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"> • YES • NO 	
	7.3.	Biological sample	Patient's biological sample available for research	<ul style="list-style-type: none"> • YES • NO 	If YES answer question 7.4
	7.4.	Link to a biobank	Biological sample stored in a biobank	<ul style="list-style-type: none"> • YES (if appropriate use link) • NO 	https://directory.bbmri-eric.eu
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"> • Disability profile / Score 	http://www.who.int/classifications/icf/whodasii/en/