

## Data Set for Rare Disease Patient Registries Recommended for European Cooperation\* (Version 3.0)

Item Group	Item No.	Item Concept	Question	Content Coding	Data Collection (one-time/repeatedly)	Comment
1. Pseudonym	1.1	Patient's pseudonym (PID)	Patient's pseudonym (as defined in the meta-dataset)	String	one-time	
2. Personal Information	2.4	Patient's date of birth	Patient's date of birth as recorded on the birth certificate	Date	one-time	
	2.5	Gender	Patient's gender	Female Male Undetermined Unknown (for the foetus)	one-time	
3. Family Information	3.1	Patient born from a relationship between related parties	Is the patient born from a relationship between related parties?	Yes No Unknown		
4. Vital status	4.1	Patient's vital status	Is the patient still alive?	Yes No Lost to follow-up Discharged from registry	repeatedly	Update of the data base at least once per year
	4.2	Patient's date of death	Patient's date of death	Date	one-time	Update of the data base at least once per year
	4.3	Death due to the rare disease	Is the death due to the rare disease the patient is suffering from?	Yes No Unknown		
5. Care Pathway	5.1	Patient's date of inclusion in the registry	Date at which the patient was included in the registry	Date		
6. Disease history	6.1	Age at onset	Age at which symptoms first appeared	Antenatal At birth XX year(s) and XX month(s) Undetermined		
	6.2	Age at diagnosis	Age at which the diagnosis was made	Antenatal At birth XX year(s) and XX month(s) Undetermined		
7. Diagnosis	7.2	Diagnosis of the rare disease	Diagnosis retained by the RD center	Alpha code		
8. Research	8.1	Agreement to be contacted for a protocol	Does the patient give permission to be contacted for a research protocol?	Yes No		
	8.2	Patient non-opposition to the reuse of data	Does the patient give permission for his/her data to be reused for other research purposes	Yes No		
	8.3	Patient having previously given a biological sample for research	Has the patient already given a biological sample for research?	Yes No		
	8.4	Patient having previously given a biological sample for molecular diagnosis	Has the patient already given a biological sample for molecular diagnosis?	Yes No		

\* This table is based on the French National Rare Diseases Minimum Data Set, RD MDS v1.08