

Warning! The rare diseases minimum data set is licensed under a Creative Commons Attribution-ShareAlike 4.0 International & 3.0 France License.
If you are a manufacturer, we allow the use of it when an interface is also created in the host application for sending data to the BNDMR.



RD MDS v1.11
French National Rare Diseases Minimum Data Set



Item group	Objective	Item #	Condition(s)	Status	Cardinality	Item	Definition	Coding			
1. Consent	Legal information	1.1		mandatory	1	Patient's non-opposition for data reuse	Was the patient duly informed that part of his/her de-identified data, will be used for public health purposes, and that he/she did not express his/her opposition?	Yes No			
		2. Patient identification	2.1	At least one identifier	optional(1)	1	Health national identifier	Patient identifier: unique national identifier allowing future connections with the French patient medical file (DMP).	String (automatically generated)		
2.2	optional		1		Patient's local hospital identifier	Local hospital identifier.	String				
3. Administrative information	Identification of RD patient	3.1		mandatory	1	Patient is affected with a rare disease	Is the patient affected with a rare disease?	Yes No			
		Type of patient	3.2		mandatory	1	foetus	Is the patient a foetus?	Yes No		
	3.a Personal information	Necessary information to identify the patient	3.a.3		mandatory	1	Patient's patronymic name (surname at birth)	Patient's patronymic name (surname at birth).	String		
			3.a.4		optional	1	Patient's commonly used last name	Patient's commonly used last name.	String		
			3.a.5		mandatory	1	Patient's first name	Patient's first name as specified on the birth certificate or identity card.	String		
			3.a.6		mandatory	1	Patient's date of birth	Patient's date of birth as recorded on the birth certificate.	Date		
			3.a.7		mandatory	1	Patient's gender	Patient's gender.	female male		
			3.a.8		mandatory	1	Country of birth	Patient's country of birth.	Country code		
			3.a.9	3.a.8 = France	mandatory	1	City of birth	Patient's city of birth.	City code		
			Information used to calculate distance between residence and hospital	3.a.10		mandatory	1	Country of residence	Patient's country of residence.	Country code	
				3.a.11	3.a.10 = France	mandatory	1	City of residence	Patient's city of residence.	City code	
			3.b Parents personal information (foetus)	Necessary information to identify the foetus	3.b.3		mandatory	1	Mother's patronymic name (surname at birth)	Mother's patronymic name (surname at birth).	String
					3.b.4		optional	1	Mother's commonly used last name	Mother's commonly used last name.	String
	3.b.5				mandatory	1	Mother's first name	Mother's first name as specified on the birth certificate or identity card.	String		
3.b.6		mandatory			1	Date of pregnancy	Start of pregnancy (dd/mm/yyyy)	Date			
3.b.7		mandatory			1	Multiple pregnancy	Pregnancy with at least 2 embryos	Yes No			
3.b.8		mandatory			1	foetus' gender	foetus' gender	female male unknown			
3.b.9		optional			1	foetus' first name	Foetus' first name if available	String			
Information used to calculate distance between residence and hospital	3.b.10				mandatory	1	Country of residence	Mother's country of residence.	Country code		
	3.b.11	3.b.10 = France			mandatory	1	City of residence	Mother's city of residence.	City code		

Item group	Objective	Item #	Condition(s)	Status	Cardinality	Item	Definition	Coding
4. Family information ⁽¹⁾	Family studies	4.1		mandatory	1	Propositus	First patient recorded in the hospital for a given family.	Yes No
		4.2	4.1 = No	mandatory	1	Propositus identifier	First patient's identifier (IdMR) recorded in the hospital for a given family.	Propositus identifier
		4.3	4.1 = No	mandatory	1	Parental relationship with the propositus	Describes the relationship between a patient of the family and the propositus.	Brother Sister [...] Maternal grandfather Maternal grandmother
		4.4		optional	1	Inbreeding (consanguinity)	Is the patient born from a relationship between related parties?	Yes No Unknown
5. Vital status	Necessary information for survival and prevalence studies	5.1		mandatory	1	Patient's vital status	Is the patient deceased?	Yes No
		5.2	5.1 = Yes	mandatory	1	Patient's date of death	Patient's date of death	Date
	Necessary information for survival studies	5.3	5.1 = Yes	mandatory	1	Death due to the rare disease	Was patient's death due to the rare disease?	Yes No Unknown
		5.4	5.1 = Yes et 5.3 = No	optional	1	Main cause of death	If death not due to the rare disease, what was the main cause of death?	ICD10 code
6. Care pathway (multiple)	Necessary information to appreciate the attractiveness of Rare Disease centres in the health professionals community and the care pathway	6.1		mandatory	multiple	Patient referred by	The patient may have been referred to the RD centre by a health care professional. This assesses the attractiveness of the RD centre for professionals referring patients.	come by himself patient association general practitioner paediatrician (non-hospital practice) paediatrician (hospital practice) centre of maternal and child health geneticist gynaecologist/obstetrician other specialist support centre centre of prenatal screening or multidisciplinary centre of prenatal diagnosis Reference centre Competence centre other
		6.2		mandatory	1	Patient's date of inclusion in the RD reference centre	Date at which the patient was recorded in the reference RD centre.	Date
	Necessary information for access rights to patient's file	6.3		mandatory	1	Name of the physician in charge of the patient	Name of the physician in charge of the patient in the current care unit	Physician identifier
	Necessary information for centres activity report (and care pathway)	6.4		mandatory	1	RD centre(s) of the patient	Name of the RD centre(s)	RD centre identifier
		6.5		mandatory	1	Not included in the label terms	The healthcare is provided to a patient whose disease is not included in the label terms (this patient and associated activities will not be recorded in annual reports of the expert centre)	Yes No

Item group	Objective	Item #	Condition(s)	Status	Cardinality	Item	Definition	Coding
7. Care activities (multiple)	Necessary information for centres' annual activity report and incidence studies (and studies on patients follow-up)	7.1		mandatory	1	Date of the performed rare disease care	Date at which the declared rare diseased patient care was performed.	Date
		7.2		mandatory	1	Context of the rare disease care activity	Context indicating the type of care performed. (Useful information for the RD centre annual report).	consultation multidisciplinary consultation day hospital traditional hospital advice on medical record during consultation expert advice on medical record multidisciplinary meeting on medical record advice given in another ward teleconsultation other context
		7.3		mandatory	multiple	Objectives of the rare disease care activity	Objectives of the declared care activity.	diagnosis disease management setup follow-up care genetic counselling prenatal diagnosis preimplantation emergency support medical act research protocol patient education child/adult transition consult
		7.4		mandatory	multiple	Occupation of the personnel performing the declared care activity	Type of personnel performing the declared activity.	social worker dietician occupational therapist physiotherapist psychologist physical therapist genetic counsellor nurse speech therapist specialist teacher physician other professional
		7.5		optional	multiple	Personnel performing the declared activity	Name of the professional performing the activity.	Professional identifier (Patronymic name, first name, national identifier)
		7.6		mandatory	1	RD Centre for which the activity is declared	Name of RD Centre for which the activity was performed	RD site identifier
		7.7		optional	1	Location of medical consultation	Service/Hospital where the activity was performed if different from patient's RD Centre of care	Country code or city code (if country = France)

Item group	Objective	Item #	Condition(s)	Status	Cardinality	Item	Definition	Coding
Diagnosis (multiple)	8. Disease history Necessary information to calculate the diagnosis delay and diagnosis wavering	8.1		mandatory	1	Age at onset of first signs	Age at which first symptoms appeared?	antenatal at birth postnatal undetermined
		8.2	8.1 = postnatal	mandatory	1	Specification of the age at onset of first signs (months)	Age at which first symptoms appeared?	Numeric (months)
		8.3		mandatory	1	Assessment of the diagnosis at first admission in the rare disease centre	Was patient's diagnosis appropriate when admitted to the RD centre?	missing inappropriate appropriate
		8.4		mandatory	1	Age at diagnosis	Age at diagnosis?	antenatal at birth postnatal undetermined post-mortem
		8.5	8.4 = postnatal	mandatory	1	Specification of the age at diagnosis (months)	Age at diagnosis (postnatal)?	Numeric (months)
	9. Diagnosis Necessary information for all epidemiological studies or identification of patients for clinical trials	9.1		mandatory	1	Current status of the diagnosis	What is the current status of the diagnosis?	ongoing likely confirmed unknown
		9.2	9.1 = likely or confirmed	mandatory	1	Patient's rare disease diagnosis	Patient's rare disease diagnosis assessed by the rare disease centre	Orpha Code (Diseases)
		9.3		optional	multiple	Additional signs associated with the rare disease	Phenotypic Diagnosis of patient, assessed in RD Centre	HPO - ICD 10 - Orpha code (Groups)
		9.4		optional	multiple	Unusual signs associated with the rare disease	Unusual/Atypical signs associated to the diagnosis, assessed in RD Centre	HPO - ICD 10 code
		9.5		optional	1	Sporadic or familial case	Is the case isolated or familial at the time of observation (assessed by the RD professional)?	Sporadic Familial
	10. Diagnosis confirmation Necessary information to appreciate methods used for diagnosis	10.1		mandatory	multiple	Method(s) used for diagnosis investigation.	What was the method used to confirm or to investigate the diagnosis?	clinical genetic testing biochemical biological imaging Functional exploration pathological anatomy other
		10.2	10.1=genetic testing	mandatory	multiple	Method used to assess the diagnosis (if applicable)	Specify the method on which the diagnosis is based.	chromosomal ACPA targeted sequencing New generation sequencing other methods
		10.3		optional	multiple	Genes	Which gene(s) are associated to the rare disease diagnosis?	Code HGNC
		10.4		optional	1	Other genetic description	Mutations or other genetic description ?	String
		10.5		optional	1	Subject seemingly healthy	Is the carrier of the mutation seemingly healthy?	Yes No
	11. Treatment Assessment of the use of specific orphan medicinal products or inclusion of patients in research protocols	11.1		optional	1	Is a treatment specific to the rare disease in progress?	Is a treatment specific to the rare disease in progress? (Nota bene : the so-called "comfort" treatments are not considered here).	Yes No
		11.2	11.1 = Yes	optional	multiple	Ongoing treatment for the rare disease	Name of the ongoing treatment specific to the rare disease. Only rare disease treatments are considered here.	ATC Code (orphan designation)

Item group	Objective	Item #	Condition(s)	Status	Cardinality	Item	Definition	Coding
12. Ante and neo-natal ⁽¹⁾	Necessary information for national neonatal studies	12.1		optional	1	Medically assisted procreation	Has a medical assistance program for procreation been used?	Yes No
		12.2		optional	1	Presence of antenatal malformation	Does the patient have a prenatal malformation?	No Unique Multiple
		12.3		optional	1	Full-term birth?	Was the patient born at full-term?	Yes No
		12.4	12.3 = No	optional	1	Term (if premature birth)	Specification of the term in case of premature birth.	Numeric (weeks)
		12.5		optional	1	Height at birth	Patient's height at birth.	Numeric (cm)
		12.6		optional	1	Weight at birth	Patient's weight at birth.	Numeric (g)
		12.7		optional	1	Head circumference at birth	Patient's head circumference at birth.	Numeric (cm)
		12.8	3.2 = Yes et 5.1 = Yes	optional	1	Foetopathology	Has a foetopathology examination been performed ?	Yes No
13. Research ⁽¹⁾	General information regarding research	13.1		optional	1	Patient involved in a protocol	Is the patient currently involved in a research protocol (cohort, ...)?	Yes No
		13.2		optional	1	Agreement to be contacted for a protocol	Does the patient give permission to be contacted for a research protocol?	Yes No
		13.3		optional	1	Patient having previously given a biological sample for RD research	Did the patient already give a biological sample for research?	Yes No
		13.4		optional	1	Patient having previously given a biological sample for molecular diagnosis	Did the patient already give a biological sample for molecular diagnosis?	Yes No

(1) : Those item groups in brown are optional. If at least one item of the group has been filled, then the status of all the group items must be respected (optional/mandatory).



This work is licensed under the Creative Commons Attribution-ShareAlike 4.0 International & 3.0 France License.

To view a copy of this license, please visit :

<https://creativecommons.org/licenses/by-sa/3.0/fr/>

<http://creativecommons.org/licenses/by-sa/4.0/>

