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		Identity	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

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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

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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)

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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
		Necessary information for all epidemiological studies or identification of patients for clinical trials	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.	Product Code (orphan designation)

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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).

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