

Supplementary Table 1. Representation of JRC common data elements in ERKReg.

Group	Element	Element Name	Field name in ERKReg	ERKReg coding
1 Pseudonym	1.1	Pseudonym	pat_id	string
2 Personal Information	2.1	Date of birth	birthdate	yyyy/mm
	2.2	Sex	sex	M=Male, F=Female
3 Patient's status	3.1	Date of status change	cr_term_date	dd/mm/yyyy
		Status type (if not currently followed)	term_reason	blank= alive and followed in registry 1=loss of follow-up 2=patient death 3=Transition to adult unit 4=Transition to other ERKReg center
	3.2	Date of death	death_date	dd/mm/yyyy
4 Care Pathway	4.1	First contact with specialized center	first_visit_date	dd/mm/yyyy
5 Disease History	5.1	Age of onset	first_symptoms_date	yyyy/mm

	5.2	Age at diagnosis	dia_date	dd/mm/yyyy
6 Diagnosis	6.1	Diagnosis of the rare disease	diagnosis diagnosis2nd	Orpha Code Orpha Code
	6.2	Genetic diagnosis	gen_diagnosis	0= No, 1= Yes, 9= Result pending HGVS classification system
	6.3	Undiagnosed case	dia_est if no: signs and symptoms compatible with symptoms_group	0= No, 1= Yes Orpha Code
7 Research	7.1	Agreement to be contacted for research purposes	informed_consent_date consent_research	dd/mm/yyyy 0=No, 1=Yes
	7.2	Consent to the reuse of data	informed_consent_date consent_registry	dd/mm/yyyy 0=No, 1=Yes
	7.3	Biological sample	Not implemented in ERKReg	
	7.4	Link to a databank	Not implemented in ERKReg	
8 Disability status	8.1	Disability score	Not implemented in ERKReg	