Proposal of Common Data Set. RD-CONNECT_WP2

RD-Connect CDE		Description/definition	Original CDE source	Variable structure/Size	Response values	Source of standardization (if applicable) & Required/Optional
IDENTIFICATION						
RDC_1	Participant global registry ID	Unique, common and interoperable identifier	RD-CONNECT	String	Length=10 Depending of the final development of this ID	RD-CONNECT Required
RDC_2	Participant local registry ID	Local registry's own identifier	All CDEs strategies	String	Length=10 According to the primary source	Required
RDC_3	First date of entry	Date when data about the participant was first reported into the local registry	WP3 RD-CONNECT	Date	4-digit Gregorian year 2-digit for month and 2 digit for day YYYYMMDD	Required
RDC_4	Record date	Date when the current registry record was updated or completed	NIH/NCATS/GRDR	Date	4-digit Gregorian year 2-digit for month and 2 digit for day YYYYMMDD	Required
RDC_5	Full name of registrar	Name of person entering the data in the registry	NIH/NCATS/GRDR	String	Length=60 Full surname, Name (Initials), Mid Name (Initials) An alternative could be to include the ORCID instead the full name	Optional

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GEOGRAPHICAL SETTING						
RDC_6	Country of participant's residence	Country where the participant usually lives	NIH/NCATS/ GRDR & EPIRARE	String	Select the 2-character country code from the International Organization for Standardization	ISO-3166 http://www.iso.org/iso/ country_codes.htm Required
RDC_7	Province/ Region/ State of participant's residence	Province/ Region/ State where the participant usually lives	NIH/NCATS/ GRDR	String		ISO-3166 http://www.iso.org/iso/ country_codes.htm Optional

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CONSENT						
RDC_8	Consent	Is consent given for international data sharing according to the national laws?	NIH/NCATS/ GRDR	Integer	1 – Yes 2 – No	Required

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SOCIO-DEMOGRAPHIC						
RDC_9	Foetus (if applicable)	Whether information is recorded for a foetus	National (France)	Integer	1-Yes 2-No	Optional
RDC_10	Participant date of birth	Date of participant's birth as reported in the national ID/passport or declared by the patient	EPIRARE	Date	4-digit Gregorian year 2-digit for month and 2 digit for day YYYYMMDD	Required
RDC_11	Vital status	Vital status of the participant (dead or alive)	NIH/NCATS/ GRDR & EPIRARE	Integer	1 – Yes 2 – No 3 –Unknown	Required
RDC_12	Participant date of death	Date of participant's death if available	EPIRARE	Date	4-digit Gregorian year	Optional
RDC_13	Principal cause of death	ICD-10 code and/or ORDO code corresponding to the principal cause of participant's death	National (France and Spain)	String	Length=5 (XXX.X)	http://apps.who.int/clas sifications/icd10/browse /2015/en Optional
RDC_14	Sex of participant	Participant's physical sex	NIH/NCATS/ GRDR & EPIRARE	Integer	1 - Male 2 - Female 3 - Undetermined 4 - Unknown	SNOMED-CT Required
RDC_15	Gender of participant	Participant's gender	Neuromics	Integer	1 - Male 2 - Female 3 - Other 4 - Transsexual 5 – Unknown	SNOMED-CT Optional
RDC_16	Ethnicity of participant	Ethnicity of participant (list)	WP2 RD-CONNECT	String	Length=5	Table H-1: Language Assistance Data Base

						(LADB), based on CDC Race/Ethnicity Codes and ISO Language Codes http://www.ahrq.gov/research/findings/final-reports/iomracereport/reldataaph1.html Optional
RDC_17	Country of birth	Country where the participant was born	NIH/NCATS/GRDR & EPIRARE	String	Select the 2-character country code from the International Organization for Standardization	ISO-3166 http://www.iso.org/iso/country_codes.htm Required
RDC_18	Province/ Region/ State of birth	Province/ Region/ State where the participant was born	NIH/NCATS/GRDR	String		ISO-3166 http://www.iso.org/iso/country_codes.htm Optional
RDC_19	City/Town/Village of birth	City/town/village where the participant was born	NIH/NCATS/ GRDR & EPIRARE	String	Length=15	Required

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colspan="7"						**DIAGNOSIS**
RDC_20	Diagnosis coding system or ontology	Name of coding system (SNOMED-CT, ICD10, ORPHA, OMIM, etc) used for the diagnosis	WP2 RD-CONNECT	Integer	1-ICD10 2-ORPHA 3-SNOMED-CT 4-OMIM	Required
RDC_21	Rare disease diagnosis	Diagnosis code from selected coding system or ontology	WP2 &WP3 RD-CONNECT	String	Length=10	Required
RDC_22	How diagnosis was made	The basis by which the diagnosis was made (Genetic laboratory analysis; Histology; History; Imaging - CT; Imaging - MRI; Imaging - PET; Physical examination; Other modality; Unknown)	WP2 RD-CONNECT	Integer	1-Clinical 2-Biochemical lab 3-Genetic 4-Histology 5- Imaging - CT; Imaging - MRI; Imaging - PET; 6-Other 7-Undiagnosed 8-Unknown	Multiple options can be selected Required
RDC_23	Diagnosis free text	Explanation about diagnosis or symptom in case of unknown diagnosis or insufficient	WP3 RD-CONNECT	String	Length=100 Free clinical description	Optional
RDC_24	Age at the date of diagnosis	Age current rare disease diagnosis was made	EPIRARE	String	Antenatal At birth 1 2 … 80	Required

					Not available	
RDC_25	Age at disease onset	Age when the first symptoms appeared	NIH/NCATS/ GRDR & Neuromics	Integer	Antenatal At birth 1 2 … 80 Not available	Required
RDC_26	Medical centre's name	Name of the medical centre sending data to the registry	National strategy (Italy)	String	Length=60	Optional
RDC_27	City of medical centre	City of the medical centre sending data to the registry	WP2 RD-CONNECT	String		ISO-3166 http://www.iso.org/iso/country_codes.htm Optional

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			FAMILY HISTORY			
RDC_28	In vitro fertilisation	Was the patient conceived as a result of in vitro fertilisation?	WP2 RD-CONNECT		Yes=1 No=2	Optional
RDC_29	Gestation at delivery	Number of gestation week	NIH/NCATS/GRDR	Integer	1 to 42	Optional
RDC_30	Rare disease family history	Which family members also are known to have the participant's rare disease?	NIH/NCATS/GRDR	String	Brother Sister […] Maternal grandfather Maternal grandmother Unknown Not provided	Multiple options can be selected Required
RDC_31	Consanguinity	Whether patient's parents are consanguineous	EPIRARE & Neuromics	Integer	1-Yes 2-No 3- Unknown	Optional