

Core Data Set – Swiss Rare Disease Registry (SRDR)

	Variable	Description	Mandatory
Patient Information			
<i>1. Personal Information</i>			
	Patient ID	Clinic-specific code in Switzerland	X
	Date of Birth	dd.mm.yyyy	X
	Sex	-Female -Male -Other -Unknown	X
	First name		X
	Last name		X
	Family name at birth		
	Street name		X
	Street number		
	Zip code		X
	City		X
	Country		X
	Phone		
	E-mail		
	Correspondence Language	-German -French -Italian -English	X
<i>2. Legal Representative</i>			
	Relationship	Parents, legal custodian etc.	
	First name		
	Last name		
	Street name		
	Street number		
	Zip code		
	City		
	Country		
	Phone		
	E-mail		
<i>3. Informed Consent</i>			
	Information date	Date on which patient has been informed by the e.g., the clinic	

X = entry mandatory; O = entry mandatory if a specific condition is met

	Variable	Description	Mandatory
	Consent status	-Consent signed -Consent refused -No signature/no refusal within 6 weeks -Consent withdrawn	X
	Date of signature/ refusal/ withdrawal	dd.mm.yyyy	X
	Level of withdrawal	-stop medical data collection -stop being contacted for questionnaire surveys or nested studies -keep minimal data set only -unknown	O
4. Patient Status			
	Vital status	-Alive -Dead -Lost to follow-up -Unknown	X
	Date of death	dd.mm.yyyy	
Clinical Information			
1. Classification			
	ORPHAcode		X
	Name of diagnosis	Information is automatically fetched from Orphanet	
	ORPHA-Link	Information is automatically fetched from Orphanet	
	ORPHAcode status	Information is automatically fetched from Orphanet	
	Classification level	Information is automatically fetched from Orphanet	
	Type of disorder	Information is automatically fetched from Orphanet	
2. Diagnosis Information			
	Date of diagnosis	-dd.mm.yyyy -unknown	X
	Date valid until	dd.mm.yyyy if a diagnosis is not valid anymore, date until diagnosis was valid	
	Type of diagnosis	-confirmed -suspected	X
	Diagnostic method	-Molecular genetics -Biochemistry -Histology -Clinical -Imaging -EEG -Newborn Screening -Prenatal -Cytology	X

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	Variable	Description	Mandatory
		-Antibody Status -Other -Unknown	
	Specify other diagnostic method		O
	How many genes or chromosomes are affected	-1 -2 -3 -unknown	O
	Affected gene or chromosome, diagnosed mutation(s)		O
	Used nomenclature		
	Mutation carrier	-Symptomatic -Presymptomatic	O
3. Disease History			
	Disease-related symptoms (past or current)	-yes -no -unknown	X
	Age at first occurrence of symptoms	-Antenatal -Newborn (0-4 weeks) -First year of life (> 4 Weeks - 1 yr.) -Toddler (1 - 5 yrs.) -School child (6 - 12 yrs.) -Adolescent (13 - 17 yrs.) -Adult (> 18 yrs.) -unknown	O
	Year of first symptoms occurrence		
	Month of first symptoms occurrence	-January -February -March -April -May -June -July -August -September -October -November -December	
	Date of first contact with specialized center	dd.mm.yyy	
	Remarks		

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Registered in another Registry			
	Type of the registry	-National registry -International registry -Other -Unknown	
	Other type of registry	specify	
	Name of registry		
	Patient's registry ID		
	Biosample available for research	-Yes -No -Unknown	
	Name of biobank	Link or name of the biobank where the biological sample is stored	
Treating Institution			
	Name of treating clinic/hospital/ private practice	automatically generated	
	Name of treating department	automatically generated	

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