

List and description of the registry elements for the ERN-LUNG BReATHeREGISTRY

Group	Number of Common Data Element in EU RD Platform	English Designation	English Definition	Value Domain (list or prose)	Example value	Comments
1. General Questions		Who entered the data?	Indicates whether the patient, a related person a member of the care team entered the data	Patient / Relative / Care Team Member/ Other	patient	
		Agreement to be contacted to share the data with the disease-specific registries if feasible	Prerequisite: email address is given and kept together with identifying data	Yes / No / not answered	Not answered = no	Email Address necessary to clarify the issue and seek patient informed consent for the disease specific registry
		Country of residence	Indicates the country of residence	Three-digit ISO 3166-1 Code		Three-digit ISO 3166-1 Code https://www.iso.org/obp/ui/#search
		How did you learn about the ERN-LUNG population Registry?	Indicates the way how users came to know about the population registry	Care team / patient organization / patient / internet / other	Patient organization	Free text could be enabled

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1. General Questions		Patient regularly cared for by one of the member HCPs of ERN-LUNG	Indicates whether this is an inside patient or not	Yes / No / not known	Yes	It is possible that a patient has more than one YES in case he has had an EXABO and/or Tele consultation and /or cross-border consultation
		Patient has utilized ERN-LUNG Cross-border online expert advice EXABO	Indicates whether this patient has utilized the cross- border online services of ERN- LUNG called EXABO	Yes / No / Don't know	Yes	It is possible that a patient has more than one YES in case he has had an EXABO and/or Tele-consultation and /or cross-border consultation
		Time needed from first contact to recommendation in EXABO	Number of days needed to receive the final report	Days	3	
		If EXABO has been utilized, how satisfied were you by these services?	Indicates the degree of satisfaction on a rating scale	Very good / good / OK / poor / very poor	good	
		Cross-border teleconsultation	Indicates whether this patient has utilized cross- border teleconsultation	Yes / No / Don't know	Yes	
		Cross-border referral	Indicates whether this patient has utilized cross- border referral	Yes / No / Don't know	Yes	

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2. Pseudonym	1.1.	Pseudonym	Patient's pseudonym	String	GSZ56AS	
3. Personal Information	2.1.	Date of birth	Patient's date of birth	Date (dd/mm/yyyy)	16/05/1977	
	2.2.	Sex	Patient's sex at birth	Female / Male / Undetermined / Foetus (Unknown)	Female	
4. Patient Status	3.1.	Patient's status	Patient alive or dead	Alive / Dead / Lost in follow-up / Opted-out	Alive	If dead then answer question 3.2
	3.2.	Date of death	Patient's date of death	Date (dd/mm/yyyy)/-	-	
5. Care Pathway	4.1.	First contact with specialised centre	Date of first contact with specialised centre	Date (dd/mm/yyyy)	23/09/2016	
6. Disease history	5.1.	Age at onset	Age at which symptoms/signs first appeared	Antenatal / At birth / Date (dd/mm/yyyy) / Undetermined	Undetermined	
	5.2.	Age at diagnosis	Age at which diagnosis was made	Antenatal / At birth / Date (dd/mm/yyyy) / Undetermined	17/07/2007	

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7. Diagnosis	6.1.	Diagnosis of the rare disease	Diagnosis retained by the specialised centre	Orpha code (strongly recommended - see link) / Alpha code / ICD-9 code / ICD-9-CM code / ICD-10 code	244	http://www.orphadata.org/cgi-bin/inc/product1.inc.php
		Name of the rare disease	The name of the diagnosis has to be given here			Ideally the name and the orpha code should match; if the person entering the data knows the diagnosis but not the orpha code this data set should still be accepted http://www.orphadata.org/cgi-bin/inc/product1.inc.php
	6.2.	Genetic diagnosis	Genetic diagnosis retained by the specialised centre	International classification of mutations (HGVS) (strongly recommended – see link) / HGNC / OMIM code		http://www.hgvs.org
	6.3.	Undiagnosed case	How the undiagnosed case is defined?	Phenotype (HPO), Genotype (HGVS)	HGVS	

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8. Research	7.1.	Agreement to be contacted for research purposes	Patient's permission exists for being contacted for research purposes	Yes / No	Yes	
	7.2.	Consent to the reuse of data	Patient's consent exists for his/her data to be reused for other research purposes	Yes / No	Yes	
	7.3.	Biological sample	Patient's biological sample available for research	Yes / No	Yes	If YES answer question 7.4
	7.4.	Link to a biobank	Biological sample stored in a biobank	yes / No (if appropriate use link)		https://directory.bbmri-eric.eu
9. Disability	8.1.	Classification of functioning/disability	Patient's disability profile according to International Classification of Functioning and Disability (ICF)	Disability profile / Score		http://www.who.int/classifications/icf/whodasii/en/