

# **ERN-PAEDCAN Partner - Paediatric Rare Tumours Network - European Registry (N°777336)**

## **D4.3 - List of variables that will be selected to link the PARTNER to the virtual consultation system**

(Version 30.06.2019)

## **List of variables that will be selected to link the PARTNER to the virtual consultation system**

A list of variables from the virtual consultation system for integration into the PARTNER data base was produced in order to use the actual data provided when seeking advice and to prevent parallel data transfer. A consensus was reached on June 7<sup>th</sup>, 2019:

1. Variables collected by the virtual consultation system should be identical with the variables used by the EUROPEAN PLATFORM ON RARE DISEASES REGISTRATION (EU RD Platform, see below).
2. Additionally, a notice should be send by the virtual consultation to the data base in case of events.



**EUROPEAN PLATFORM ON RARE DISEASES REGISTRATION (EU RD Platform)**

**SET OF COMMON DATA ELEMENTS FOR RARE DISEASES REGISTRATION**

GROUP	ELEMENT N°	ELEMENT NAME	ELEMENT DESCRIPTION	CODING	COMMENT
1. Pseudonym	1.1.	Pseudonym	Patient's pseudonym	<ul style="list-style-type: none"> <li>String</li> </ul>	The JRC is working on providing a pseudonymisation tool to the registries
2. Personal information	2.1.	Date of birth	Patient's date of birth	<ul style="list-style-type: none"> <li>Date (dd/mm/yyyy)</li> </ul>	
	2.2.	Sex	Patient's sex at birth	<ul style="list-style-type: none"> <li>Female</li> <li>Male</li> <li>Undetermined</li> <li>Foetus (Unknown)</li> </ul>	
3. Patient Status	3.1.	Patient's status	Patient alive or dead	<ul style="list-style-type: none"> <li>Alive</li> <li>Dead</li> <li>Lost in follow-up</li> <li>Opted-out</li> </ul>	If dead then answer question 3.2
	3.2.	Date of death	Patient's date of death	<ul style="list-style-type: none"> <li>Date (dd/mm/yyyy)</li> </ul>	
4. Care pathway	4.1.	First contact with specialised centre	Date of first contact with specialised centre	<ul style="list-style-type: none"> <li>Date (dd/mm/yyyy)</li> </ul>	

5. Disease history	5.1.	Age at onset	Age at which symptoms/signs first appeared	<ul style="list-style-type: none"> <li>• Antenatal</li> <li>• At birth</li> <li>• Date (dd/mm/yyyy)</li> <li>• Undetermined</li> </ul>	
	5.2.	Age at diagnosis	Age at which diagnosis was made	<ul style="list-style-type: none"> <li>• Antenatal</li> <li>• At birth</li> <li>• Date (dd/mm/yyyy)</li> <li>• Undetermined</li> </ul>	
6 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	<a href="http://www.orphadata.org/cgi-bin/inc/product1.inc.php">http://www.orphadata.org/cgi-bin/inc/product1.inc.php</a>
	6.2.	Genetic diagnosis	Genetic diagnosis retained by the specialised centre	International classification of mutations (HGVS) (strongly recommended – see link) / HGNC / OMIM code	<a href="http://www.hgvs.org">http://www.hgvs.org</a>
	6.3.	Undiagnosed case	How the undiagnosed case is defined	<ul style="list-style-type: none"> <li>• Phenotype (HPO)</li> <li>• Genotype (HGVS)</li> </ul>	
7. Research	7.1.	Agreement to be contacted for research purposes	Patient's permission exists for being contacted for research purposes	<ul style="list-style-type: none"> <li>• YES</li> <li>• NO</li> </ul>	
	7.2.	Consent to the reuse of data	Patient's consent exists for his/her data to be reused for other research purposes	<ul style="list-style-type: none"> <li>• YES</li> <li>• NO</li> </ul>	
	7.3.	Biological sample	Patient's biological sample available for research	<ul style="list-style-type: none"> <li>• YES</li> <li>• NO</li> </ul>	If YES answer question 7.4
	7.4.	Link to a biobank	Biological sample stored in a biobank	<ul style="list-style-type: none"> <li>• YES (if appropriate use link)</li> <li>• NO</li> </ul>	<a href="https://directory.bbmri-eric.eu">https://directory.bbmri-eric.eu</a>
8.Disability	8.1.	Classification of functioning/disability	Patient's disability profile according to International Classification of Functioning and Disability (ICF)	<ul style="list-style-type: none"> <li>• Disability profile / Score</li> </ul>	<a href="http://www.who.int/classifications/icf/whodasii/en/">http://www.who.int/classifications/icf/whodasii/en/</a>