National best practices
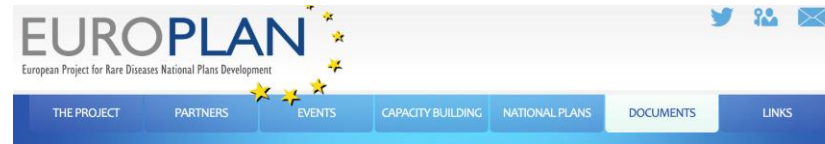# for ERN integration:
Czech Republic

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Historical overview

Czech Republic
- Czech National Strategy for Rare Diseases: 2010-2020 Press release - czech (cz)
- Czech National Strategy for Rare Diseases: 2010-2020 Press release - english (en)
- Czech National Action Plan for Rare Diseases 2012-2014 - czech (cz)
- Czech National Action Plan for Rare Diseases 2012-2014 - english (en)
- Second Czech National Action Plan for Rare Diseases 2015-2017 - czech (cz)
- Second Czech National Action Plan for rare diseases 2015-2017 - unofficial english version (en)
- Second Czech National Action Plan for rare diseases 2015-2017 - Annex 1 - unofficial english version (en)

- National strategy for rare diseases 2010-2020
- Interdisciplinary Council for Rare Diseases at the MoH
  - „Advisory board" meeting annually, no competencies, no funding
- National Coordination Center for Rare Diseases at Motol University Hospital
  - No funding, no competencies, no defined link to policy makers
- Patient environment evolution

Historical overview

2017

- 17 ERNs - 28 full members

2022

- 22 ERNs - 48 full member centres of 13 HCPs
  - No ERN coordinator
- Formal status od „highly specialised care centres for RD"
  - Competencies and integration into existing structures not defined
  - Capacity limited, economically largely unprofitable

Availability of RD data in the CZ – up to 2022/3

- **All ERN centers hosted by HCs directly managed by the MoH**
  - Their EHR vary significantly
  - RD patients identified via their birth code + ICD10
  - Only proportion of RD patients can be traced within the National Healthcare Information System (NZIS)
- **Information largely unavailable on**
  - Numbers of patients with defined RD
  - Their geographical distribution
  - Care characteristics: quality and costs

Czech Republic and ERN integration: „Pilot project"

- To map RD care in 4 ERN centers / 3 HCPs via patient tracking in the National Healthcare Information System
- To serve as a basis for **negotiation with payers** Re optimisation of reimbursement
- **Patient identification**
  - Specific „signal" code in EHR (later +Orphacode)
  - Data analysis of patient characteristics and clinical intervention codes and their reimbursement against personal costs of clinical teams
- **Preliminary results**
  - Disproportion between staff costs and reimbursement of their work
  - Difficult data interpretation due to lack of identification of centers within HCPs

VŠEOBECNÁ FAKULTNÍ
NEMOCNICE V PRAZE

European
Reference
Network
for rare or low prevalence
complex diseases

Network
Immunodeficiency,
Autoinflammatory and
Autoimmune Diseases
(ERN RITA)

Member
Všeobecná fakultní
nemocnice v Praze —
Česká republika

RD patient identification in EHR from 2024

- **Recognition of the necessity of identifying RD patients in EHR**
  - A pre-requisite of any potential re-organisation/funding optimisation
- **RD patient identification rule**
  - Specific „signal" code to accompany an Orphacode for RD healthcare billing
  - Exclusively contracted for ERN centers only
  - Orphacode entry = confirmation of RD diagnosis provided by an ERN centre expert
- **An interim funding solution**
  - Extra bulk payment to each ERN center („bonification") by payers (insurance companies)
    - **recognising extra work and effort associated with RD care**

RD data systematic collection

- **Project „RARE"**
  - A national RD registry forrunner
  - Preparation of the generic RD dataset in collaboration with the DIGOVO project
- **National RD registry**
  - Proposed as an adendum to the recent amendment of the Health Services Act No. 372/2011 Coll.
  - Legally operational probably from 01/2025
  - Interoperability with
    - **Hospital DHR and standard RD dataset for direct data export (single data entry)**
    - **Other existing RD registries**

Source data - EHR

- **Project „DIGOVO": DIGitalisation and Optimisation of RD (=VO) care**
  - Functional and implementation specification of the data standard for RDs and its pilot implementation
  - Resouces:
    - **European Common Data Elements (CDE)**
    - **French SDE, CDE and EUCERD JA**
- **Data directly entered in EHR**
  - Prevents mistakes of multiple data re-entry
  - Reduces time and effort
  - Allows for data sharing

EUROPEAN PLATFORM ON RARE DISEASE 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	• String	https://eu-rd-platform.jrc.ec.europa.eu/spider
2. Personal Information	2.1.	Date of birth	Patient's date of birth	• Date (dd/mm/yyyy)	
	2.2.	Sex	Patient's sex at birth	• Female • Male • Undetermined • Foetus (Unknown)	
3. Patient Status	3.1.	Patient's status	Patient alive or dead	• Alive • Dead • Lost in follow-up • Opted-out	If dead then answer question 3.2
	3.2.	Date of death	Patient's date of death	• Date (dd/mm/yyyy)	
4. Care pathway	4.1.	First contact with specialised centre	Date of first contact with specialised centre	• Date (dd/mm/yyyy)	
5. Disease history	5.1.	Age at onset	Age at which symptoms/signs first appeared	• Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined	
	5.2.	Age at diagnosis	Age at which diagnosis was made	• Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined	
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	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)	
7. Research	7.1.	Agreement to be contacted for research purposes	Patient's permission exists for being contacted for research purposes	• YES • NO	
	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	
	7.3.	Biological sample	Patient's biological sample available for research	• YES • NO	If YES answer question 7.4
	7.4.	Link to a biobank	Biological sample stored in a biobank	• YES (if appropriate use link) • NO	https://directory.bbmri-eric.eu
8.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/

SYPOVO team

Thank you

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