



# Motivation

## Why to collect RD data

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★ <http://rd-code.eu>

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# Motivation for RD data collection

- > Any disease affecting fewer than 5 people in 10,000 in the EU is considered as rare. (by prevalence)
- > Do we know the prevalence of Rare Diseases?
- > No health data collection by prevalence in the Czech Republic (only by incidence, by date of health service provided, by date of death, by other mechanism)
- > Individual disease cases identified (coded) by ICD-10 (WHO version) in the Czech Republic. Is it sufficient for RD?
- > Not sufficient! **Only 240 ICD-10 codes specific for RD entities**. But there are about 7,000 RD entities at all...

# ...but there are also clinical registries!

- > only for some RD in the Czech Republic (e.g. cystic fibrosis, diabetes in children, muscular dystrophies) or variants/groups specific data collections exist = **clinical registries (CR)**
- > (or orphan registries, registries of patients treated with orphan drugs, disease specific registries, patient registries,...)
- > CR collect detailed information, but they are limited:
  - ★ no obligatory reporting (subject to consent for patients, voluntary for clinicians), no full population coverage, no central support
  - ★ often only research project for a limited time
  - ★ only one workplace or limited selection of workplaces/centres
  - ★ different data models = made for limited purposes, incompatibility... but also the benefits, for example a detailed data, including the results of the examination
- > **CR are not suitable for the overview about RD** and for the prevalence data collection.

# ...but there are administrative data for health insurance funds!

## > Important limitations of administrative data:

- ★ Not containing info about health care outside obligatory public health insurance
- ★ Distorted by administrative coding rules and methodology (e.g. limitations of some diagnostic test only for specific diagnostic codes reported!), differences between in-patient and out-patient care
- ★ Focused on episodes of care (fragmentation), not individual patients
- ★ Diagnoses/problems coded by ICD-10\*

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\*) The goal is to identify RD cases by Orpha codes in administrative data – first attempt is to include Orpha codes in the specific reports for health insurance funds (see in the following presentation)

# ...there is also the National registry of congenital malformations (NRVV)

- > For majority of RD the right solutions for the reporting on the national level in the Czech Republic
- > Beneficial for Orpha codes initial implementation (integration into health information systems/EHR)
- > ... in addition Orpha codes coding by OMIM, SSIEM allowed
- > Methodology suitable for prevalence collection
- > RD cases identification with connection of other health care data from National Health Information System (NHIS)= analyses, reasearch, passage through health care system

Not all-round solution!

It is important to mark/code RD cases in the documentation in the standard way (Orpha codes) and first of all in EHR systems of health care providers.

# Next steps in the Czech Republic

- > Launch of orpha.net portal Czech version
- > Updates of the Czech translation of the Orphanet RD terminology
- > Updates of Orphanet Classifications (on the international level)
- > Implementation of Orpha codes to systems of health data exchange (eHealth, ERNs, crossborder care)

# Future of RD coding?

## ICD-11 will not solve all problems with granularity...

ICD-11 for Mortality and Morbidity Statistics (Version : 04 / 2019)

Search Ehlers [Advanced Search] Browse Coding Tool Special Views Info

Foundation Id : <http://id.who.int/icd/entity/236564145>

### LD28.01 Marfan syndrome

**Parent**  
LD28.0 Marfan syndrome or Marfan-related disorders [Show all ancestors](#)

**Description**  
Marfan syndrome is a systemic disease of connective tissue characterized by a variable combination of cardiovascular, musculo-skeletal, ophthalmic and pulmonary manifestations. Cardiovascular involvement is characterized by 1) progressive dilation of the aorta accompanied by an increased risk of aortic dissection, which affects prognosis and 2) mitral insufficiency. Skeletal involvement is often the first sign of the disease and can include dolichostenomelia, large size, arachnodactyly, joint hypermobility, scoliotic deformations, acetabulum protrusion, thoracic deformity, dolichocephaly of the anteroposterior axis, micrognathism or malar hypoplasia. Ophthalmic involvement results in axile myopia, which can lead to retinal detachment and lens displacement.

### Orphanet classification of rare genetic diseases

- > [Rare genetic disease](#) ORPHA:98053
  - [Rare genetic eye disease](#) ORPHA:101435
    - [Rare genetic disorder of the visual organs](#) ORPHA:522504
      - [Rare genetic disorder of the anterior segment of the eye](#) ORPHA:522538
        - [Rare genetic corneal disorder](#) ORPHA:522556
          - [Rare genetic disorder with corneal involvement as a major feature](#) ORPHA:522558
            - [Syndromic genetic keratoconus](#) ORPHA:522564
              - [Marfan syndrome](#) ORPHA:558
                - [Marfan syndrome type 1](#) ORPHA:284963
                - [Marfan syndrome type 2](#) ORPHA:284973

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**THANK YOU FOR YOUR ATTENTION**