



Motivation

Why to collect RD data

Miroslav Zvolský

Department of Clinical Classifications,
IHIS CR

Version 22nd April 2020

★ <http://rd-code.eu>

This presentation is part of the project 826607/ 'RD-CODE' which has received funding from the European Union's Health Programme (2014-2020).

The content of presentation represents the views of the author only and is his/her sole responsibility; it can not be considered to reflect the views of the European Commission and/or the Consumers, Health, Agriculture and Food Executive Agency or any other body of the European Union. The European Commission and the Agency do not accept any responsibility for use that may be made of the information it contains.



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*
-

*) 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, research, 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 EN

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 axial 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

miroslav.zvolsky@uzis.cz

rd-code@uzis.cz

THANK YOU FOR YOUR ATTENTION