Motivation
Why to collect RD data

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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

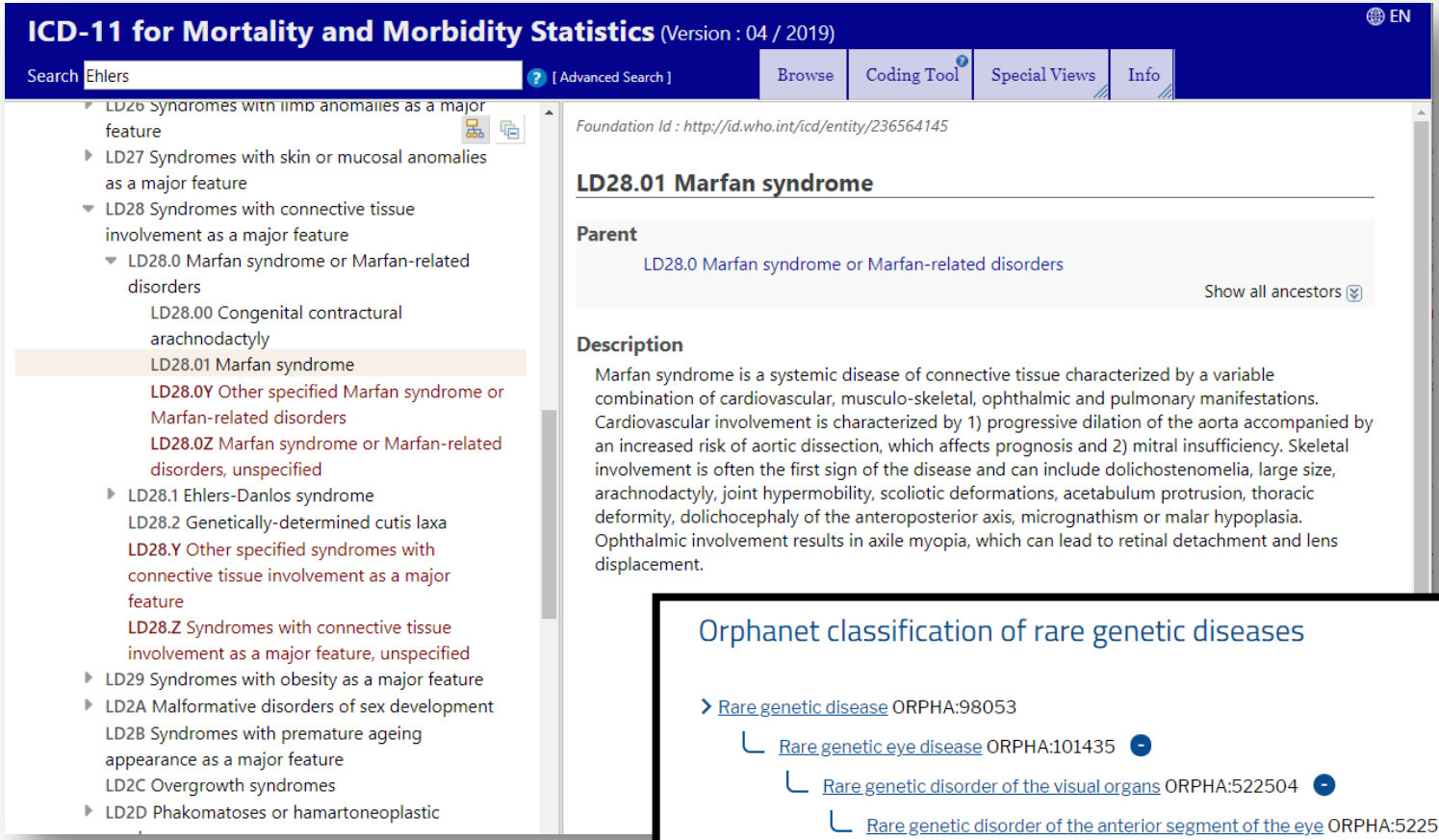
Not all-round solution!
It is important to mark/code RD cases in the documentation in the standard way (Orpha codes) and first of all **in EHR systems of health care providers.**

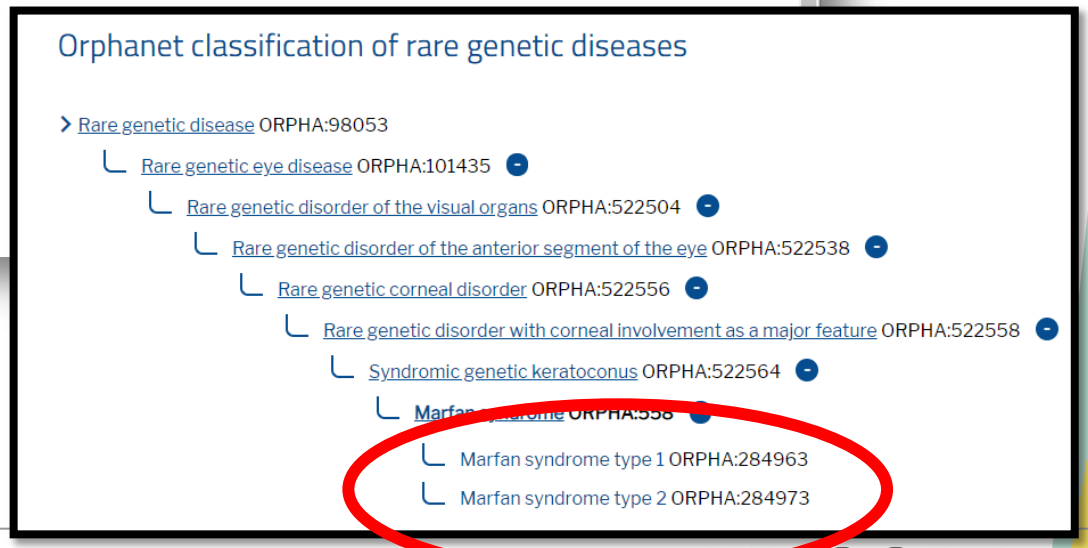
Next steps in the Czech Republic

- 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…





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THANK YOU FOR YOUR ATTENTION