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Czech Republic Outcome Report

DELIVERABLE

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Introduction

Although the major part of rare disease names is translated to the Czech language and ORPHAcodes are available, there is still a lack of objective rare diseases case data in the Czech Republic.

Hospital information systems are not yet suitable for entering rare disease cases. Currently, it is possible to enter the data of rare disease cases to the National Registry of Reproductive Health (Module of Congenital Malformations). Rare disease information can be coded through ORPHAcodes, OMIM, ICD-10, SSIEM. However, there is still a deficit of awareness about this possibility. Another problem remains, there isn't enough informed and trained coders. To this situation also the project goals for the Czech Republic correspond:

- **Start reporting ORPHAcodes-coded RD cases** in 2 major healthcare providers (hospitals) with at least 50 new cases per year in each to the National Registry of Reproductive Health (NRRH, web interface or b2b data interface)
- **Analyse technical readiness** of used Hospital Information System for producing such data
    - Motol University Hospital - information system UNIS; http://www.steiner.cz/produkt-univerzalni-nemocnicni-informacni-system
    - General University Hospital - information system Medea; http://www.stapro.eu/
- **Extend the reporting routines to other healthcare providers** (mainly those with ERN centers)
    - At least 7 hospitals/200 cases per year
- **Train coding clinical experts** in the use of ORPHAcodes focused on hospitals participating to ERNs
    - At least 3 training sessions (in Prague and Brno)

Recent progress and results

Reporting of rare disease cases

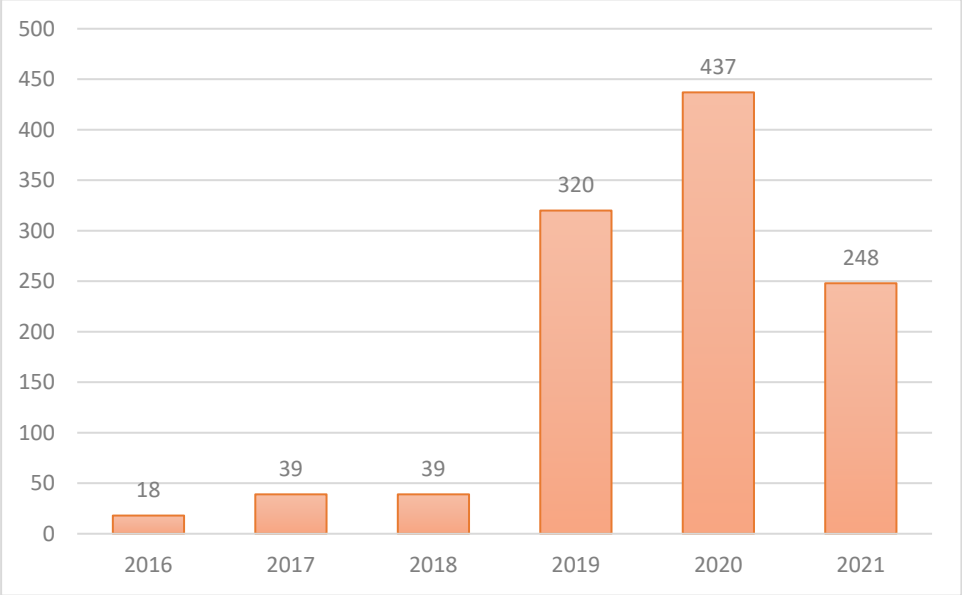
Motol University Hospital and General University Hospital started reporting rare disease cases already in 2016. During the RD-CODE project there has been enormous increase in the ORPHAcode coded cases reaching maximum numbers in 2020. Especially General University Hospital is reporting pediatric rheumatology and inborn errors of metabolism cases.

Since 2016 a total of 898 cases of rare diseases with ORPHAcode have been rereported to the National Registry of Reproductive Health (Module of Congenital Malformations) by the General University Hospital.

Table 1: The number of reported cases with ORPHAcodes in the National Registry of Reproductive Health, 2016–2021 (as of 15th November 2021).

	National Registry of Reproductive Health cases with ORPHAcode recorded		
Year	**General University Hospital**	**Other Healthcare Providers**	**Total**
2016	8	10	18
2017	18	21	39
2018	2	37	39
2019	273	47	320
2020	373	64	437
2021	224	24	248
Total	**870**	**135**	**1005**

Figure 1: The number of reported cases with ORPHAcodes in the National Registry of Reproductive Health, 2016–2021 (as of 15th November 2021).



Module of Congenital Malformations of the National Registry of Reproductive Health allows to enter data in two different dataset formats: congenital malformation or genetic disease. The genetic disease data format is customized for entering ORPHAcode coded rare diseases with limited additional information available during the outpatient visit and ongoing diagnostic process. During RD-CODE project, collaborating workplaces accommodated to fill in the simpler dataset for genetic diseases.

Table 2: Number of cases with ORPHAcodes in the National Registry of Reproductive Health by type of diagnosis, 2019–2021 (as of 15th November 2021).

Year	Congenital malformations	Genetically determined diseases
2019	155	165
2020	57	380
2021	37	211

Figure 2: Proportion of ORPHAcodes in the National Registry of Reproductive Health by type of diagnosis, 2019–2021 (as of 15th November 2021).

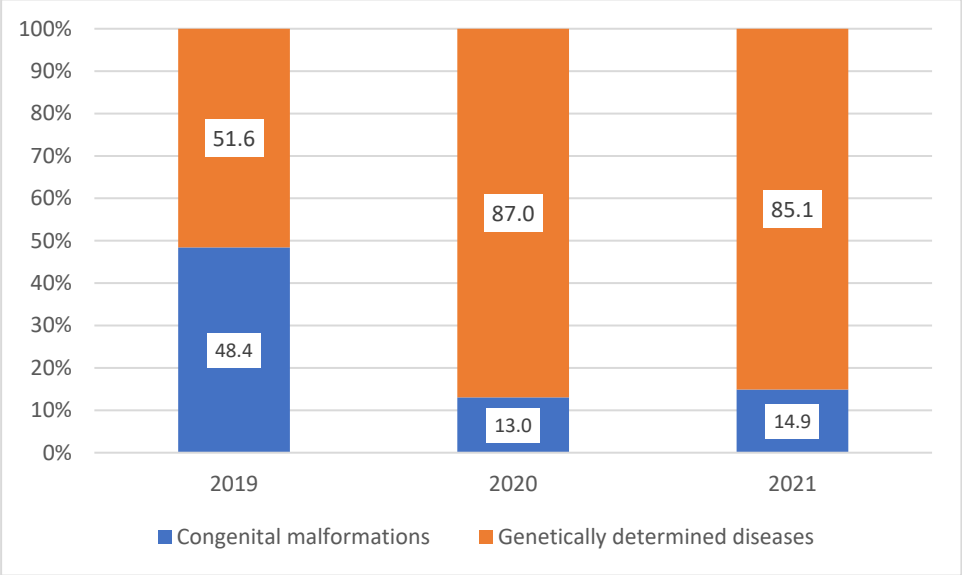


Table 3: The most frequent diseases coded in the National Registry of Reproductive Health with ORPHAcode, 2019–2021 (ORPHAcodes with 5 or more records are listed).

ORPHAcode	ORPHAcode entity title	Frequency
42642	PFAPA syndrome	162
247846 (currently 85410)	OBSOLETE: Oligoarticular juvenile idiopathic arthritis without anti-nuclear antibodies	65
404580	Polyarticular juvenile idiopathic arthritis	52
85410	Oligoarticular juvenile idiopathic arthritis	36
79241	Biotinidase deficiency	28
247854 (currently 85408)	OBSOLETE: Rheumatoid factor-negative juvenile idiopathic arthritis with anti-nuclear antibodies	25
870	Down syndrome	22
85414	Systemic-onset juvenile idiopathic arthritis	21
85438	Enthesitis-related juvenile idiopathic arthritis	20
280914	Idiopathic anterior uveitis	13
42	Medium chain acyl-CoA dehydrogenase deficiency	13
3380	Trisomy 18	12
85408	Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis	11
2185	Congenital hydrocephalus	10
716	Phenylketonuria	9
1041	Hydrops fetalis	8

324964	Chronic nonbacterial osteomyelitis/Chronic recurrent multifocal osteomyelitis	8
93552	Pediatric systemic lupus erythematosus	8
85435	Rheumatoid factor-positive polyarticular juvenile idiopathic arthritis	8
43	X-linked adrenoleukodystrophy	8
90636	Autosomal recessive non-syndromic sensorineural deafness type DFNB	7
79254	Classic phenylketonuria	7
342	Familial Mediterranean fever	6
352	Galactosemia	6
90289	Localized scleroderma	6
660	Omphalocele	6
324939	Periodic fever syndrome of childhood	6
2014	Cleft palate	5
2368	Gastroschisis	5
264580	Glycogen storage disease due to liver phosphorylase kinase deficiency	5
511	Maple syrup urine disease	5
247861 (currently 85408)	OBSOLETE: Rheumatoid factor-negative juvenile idiopathic arthritis without anti-nuclear antibodies	5
85436	Psoriasis-related juvenile idiopathic arthritis	5
3378	Trisomy 13	5

When mapping to ICD-10 codes, most of the cases were classified to the chapters:

- Endocrine, nutritional and metabolic diseases (E00-E90) = 354 cases,
- Diseases of the musculoskeletal system and connective tissue (M00-M99) = 242 cases,
- Congenital malformations, deformations and chromosomal abnormalities (Q00-Q99) = 239 cases.

Survey for getting more detailed information about the ways of entering data to the register and the conditions of hospital information systems

Survey interviews were conducted in the cooperating centers with our coders in Motol University Hospital and General University Hospital. All interviews were semi-structured with the space for discussion and comments. Questions formulated for the purposes of interviews were noted in the previous report. Part of the interview was also devoted to introducing National Register of Congenital Malformations and to entering a rare disease case data trial. All coders had the opportunity to comment about changes for the web interface to make coding as simple as possible for them.

RD-CODE Training Courses

The first training course was organized on February 19th 2020 in Motol University Hospital with 17 participants. Other lessons had to be postponed due to the COVID-19 outbreak. The training was intended for persons reporting health care and for persons responsible for reporting to the IHIS CR registers (either from individual clinics or ERN participating workplaces).

The aim of this training is to show why it is important to report rare disease cases and how and where to report them in the Czech Republic.

The area of reporting rare diseases is now in the center of attention of both insurance companies and workplaces involved in ERNs and it is also related to the pilot project of centers for rare diseases in the Czech Republic.

The initial training had been designed as 2 hours seminar and practical exercise how to work with National registry of congenital malformations. All materials in Czech are available at http://rd-code.ublg.cz/.

There were 2 following courses during 2021, in the General University Hospital in Prague (June) and University Hospital in Brno with 14 and 13 participants.

For interested experts from other healthcare providers and other institutions, training material were redesigned to the Moodle e-learning course which is available since summer 2021 at https://skoleni.uzis.cz/.

Updates of the Czech translation of Orphanet rare diseases nomenclature

During the 2021 Czech translation of Orphanet rare diseases nomenclature has been updated and now we have the process of continual adoption of Orphanet updates for the future.

The Czech version of the Orphanet rare diseases nomenclature content is available at:

- Browser at the Orphanet website,
- Downloadable files via Orphadata, other Orphanet tools like Data Visualisation Tools and API,
- Czech website https://slg.cz/vzacna-onemocneni/.

The Czech ICD-10 portal was updated with all Orphanet terminology https://mkn10.uzis.cz/.

Closer cooperation has been launched with Czech Health Insurance Funds, when they included the ORPHAcodes value in their datasets for reimbursement purposes starting in January 2021 (partially in some data messages during 2020). The cooperation continues and the methodology for updating the valuesets of ORPHAcodes has been introduced.

Conclusion

During the RD-CODE project we have enforced the reporting volume of ORPHAcodes coded cases to the National Registry of Reproductive Health reaching preset indicators. We occurred problems in redesigning internal information systems in collaborating hospitals. The process of changing these software products needs strong competencies and standards to be developed on the national level. As a secondary achievement, some steps to design specifications of those changes started in the collaborating hospitals.

There are other positive side effects of RD-CODE project in the Czech Republic:

- In the parallel project of National Centre for Medical Nomenclatures and Classifications the Czech translation of the Orphanet website has been done and translation of the Human Phenotype Ontology has started,
- Expertise of the Czech RD-CODE team including analysts and coders raised, outcomes of the RD-CODE project were transferred and used by clinical coders in the Czech Republic,
- Raising of awareness about ORPHAcodes, media campaign, rare disease coding was main topic of the conference KlasifiKon 2020 (https://www.ceskatelevize.cz/porady/11412378947-90-ct24/221411058130811-jak-efektivneji-pecovat-o-pacienty-se-vzacnymi-onemocnenimi ; https://terapie.digital/zpravodajstvi/z-domova/vzacna-onemocneni-mluvi-cesky.html)
- Workplaces included in European Reference Networks debating collection of data about rare diseases in the Czech Republic with piloting project starting in 2021,
- Other healthcare providers are informed about ORPHAcodes and started to implement them in their SW (Hospital Information Systems) e.g. Hospital České Budějovice.

The biggest challenges for the future include:

- Continuous updates of the Czech translation of Orphanet Rare Disease Nomenclature and value sets based on ORPHAcodes,
- Developing standard for rare diseases collection in hospitals connected to newly developed eHealth solutions,
- Long term communication with ERN workplaces and hospitals in the Czech Republic forcing the use of ORPHAcodes in the healthcare data.