III.

3rd Czech National Action Plan for Rare Diseases

Report on the Fulfilment of Tasks from the National Action Plan for Rare Diseases for 2015–2017 and the National Action Plan for Rare Diseases for 2018–2020.

Introduction

Rare diseases (RD) are a clinically heterogeneous group of approximately 6000–8000 different diseases whose common feature is their very low prevalence in the general population (i.e. affecting fewer than 1:2000 inhabitants according to the EU definition). If diagnosed late or misdiagnosed, RD often result in irreversible harm to the patient's health, or even death, thereby inadvertently placing a burden on health and social budgets.

Approved through Government Resolution No. 466 of 14 June 2010, the "National Strategy for Rare Diseases for 2010–2020" outlines the overall issue of rare diseases from the perspective of both the EU and the Czech Republic and proposes key objectives and measures to improve the situation in this area in the Czech Republic. The National Strategy for Rare Diseases for 2010–2020 (National Strategy) aims to ensure the timely diagnosis of and availability of adequate treatment for RD, coordinate and centralise effective RD patient care, improve RD education and awareness among both the medical community and the general public, promote cooperation at the national and international level, improve RD identification within the International Classification of Diseases (ICD-10), as well as to develop European cooperation. The National Strategy is the basis for individual national action plans within the purview of the Ministry of Health.

The second "National Action Plan for Rare Diseases for 2015–2017", which was approved through Government Resolution No. 76 of 4 February 2015 and ended on 31 December 2017, specified priority tasks and activities to allow for the gradual implementation of the National Strategy's objectives and measures. Secondary tasks, tools, responsibilities, deadlines, financial resources and indicators of the fulfilment of the various tasks under the 2nd National Action Plan have been set in a way that makes them feasible within the given period.

In 2015–2017, tasks and activities within the following key areas have been specified under the 2nd National Action Plan:

1. Improving RD awareness
2. RD education, including the incorporation of specific RD-related issues into undergraduate and postgraduate education for health care professionals
3. RD prevention, with a focus on the development of preconception and prenatal diagnosis
4. Increasing the number of RD diagnosed as part of neonatal screening

5. Improving the availability and quality of care for RD patients, improving diagnosis and treatment effectiveness and ensuring that all RD patients have equal access to indicated and high-quality health care
6. Improving the quality of life and social integration of people with RD
7. Supporting RD science and research, developing basic and applied research
8. Harmonising and developing data collection and biological sampling within RD at both the national and regional levels
9. Supporting and strengthening the role of RD patient organisations
10. Inter-ministerial, interdisciplinary and international RD cooperation
11. International RD cooperation

In the 2nd National Action Plan for 2015–2017, emphasis is placed mainly on support for timely identification and diagnosis, centralisation of patient care, harmonisation and development of data collection, development of standards of care, improved awareness and education, continuity of on-going international cooperation and establishing new contacts, as well as Czech centres' involvement in common European databases and registers that collect clinical data. In addition, support for the participation of our leading clinical institutes in the European Reference Network for RD has also been highlighted (www.ec.europa.eu/health/ern_en).

In order to improve the quality of life and social inclusion of RD patients, it will be important to establish social care beds, respite centres and rehabilitation facilities and expand long-term social care beds with trained personnel and special equipment. Attention needs to be given to effective and timely RD pharmacotherapy following up on internationally recognised standards and best practices, as well as increased education and awareness on RD issues among both the professional community and the general public. Finally, RD objectives include standardising and harmonising RD health procedures in accordance with good practices in the European Union (see e.g. EUCERD recommendations – www.eucerd.eu/?page_id=13).

Report on the Fulfilment of Tasks from the National Action Plan for Rare Diseases for 2015–2017

1. Improving RD awareness

1.1. Operating and updating web portals for RD and setting up a helpline

In order to improve RD awareness, the new www.vzacni.cz portal has been set up which uses impressive patient stories to present specific diagnoses and, for each RD diagnosis, it provides comprehensive case reports, contact information and information about the given patient organisation and relevant professional resources. The portal has become an important source of verified information for the media, patients and the professional community. A publication containing stories has been released and it won the main award of the Government Committee for Disabled Citizens in the print category in 2016, and this portal continues to be developed. Its aim is to raise RD awareness and present specific and expert-verified information on this issue. An important source of information on developments in the rare disease area is the website and Facebook (https://cs-cz.facebook.com/CAVO.vzacna.onemocneni/) portal of the Rare Diseases Czech Republic (www.vzacna-onemocneni.cz) and information pages of the National Coordination Centre that was established by the Ministry of Health at Motol University Hospital in 2015 (www.nkcvo.cz). At the European level, the most up-to-date source of professional information on individual RO for both the professional community and the general public is the www.orpha.net portal, and the NCCRD centre guarantees its international and Czech version www.orphanet.cz. This portal is recommended by the Czech Society of Medical Genetics and Genomics at Jan Evangelista Purkyně Czech Medical Association as a key source of comprehensive information on RD. Also, the NCCRD maintains the expert website https://vzacnenemoci.nkcvo.cz/ which summarises activities in the area of RD legislation and expert advice from domestic and international sources.

Rare Diseases Czech Republic (RDCZRDCZ, RD patient organisations associated under RDCZ and individual patients also participate in international projects that contribute to sharing information on individual diagnoses, such as RareConnect (www.rareconnect.org), or take part in surveys whose results are an additional source of information for RD communication in the Czech Republic (Rare Barometer Voices; www.eurordis.org/voices) at the European association of RD patient organisations – Eurordis (www.eurordis.org).

In the given period, the services of an e-mail information helpline for both the professional community and the general public have also been developed – help@vzacna-onemocneni.cz responding within 2 business days of receipt of an inquiry. This helpline is operated in cooperation with NCCRD and RDCZ. The aim is to provide RD expert advice to doctors, especially at the stage of disease diagnosis, and to direct patients and their families with undiagnosed diseases towards specialised centres. The e-mail address is also used by actual patients who are searching for the right specialists for their specific RD, especially in cases of unclear RD diagnoses. In order to implement the e-mail helpline, a direct information campaign was carried out among the professional community, with a focus on general practitioners for children (the Association of General Practitioners for Children and Youth of the Czech Republic; www.detskylekar.cz and the Association of General Practitioners /for adult patients/ – https://splcr.cz/). Also, a growing number of public inquiries have been received at the contact e-mail address cavo@vzacna-onemocneni.cz., where inquirers can get a response via e-mail within 48 hours. At this e-mail, 32 inquiries were answered in 2015, and the number increased to 54 in 2016 and as many as 84 inquiries in 2017. The planned telephone helpline has not been set up due to the associated administrative, logistical and financial demands.

Individual patient organisations greatly improve the information quality of their websites and, rather than serving only the organisation members, they usually take the role of a source of information and advice for all people with the given diagnosis (www.vzacna-onemocneni.cz/o-nas/clenstvi/prehled-clenu-asociace.html). Currently, their role is irreplaceable and is of great benefit to patients with the specific RD. Many patient organisations also operate counselling centres staffed by a leading expert in the given field. They communicate significantly via the RDCZ Facebook portal.

The portal www.novorozenecky-screening.cz, which is maintained by the National Coordination Centre for Neonatal Screening at the General University Hospital, is intended for both the general public and the professional community and it provides detailed information on the nationwide neonatal screening programme in Czech Republic, which is aimed at early identification of serious RD in newborns, in accordance with international recommendations (www.isns-neoscreening.org/). The portal is divided into two parts, one for the general public, especially parents as they are directly interested in neonatal screening, and the other for the professional community, i.e. health care professionals. The expansion of neonatal screening to include additional RD in line with the Methodological instruction outlined in Ministry of Health Bulletin no. 6/2016 (www.novorozeneckyscreening.cz/file/71/zdravotnictvi-06-16.pdf) was a great success. Neonatal screening is carried out in accordance with applicable legislation, i.e. Act No. 372/2011 Sb., on health

care services and the conditions under which they are provided, as amended, and Act No. 373/2011 Sb., on specific health care services, as amended.

Over the past period, the register of rare examinations www.registr-raritnich-vysetreni.cz was also developed in collaboration with NCCRD and the Czech Society of Clinical Biochemistry at Jan Evangelista Purkyně Czech Medical Association (www.cskb.cz). Similarly, the register and information portal of an RD subgroup – congenital developmental defects was also further developed within the portal http://www.vrozene-vady.cz/. The latter working group also worked with international associations for this issue – EUROCAT (www.eurocat-network.eu/).

The project establishing the National Information and Education Portal on Childhood Oncological Diseases was implemented. Designed as a collaboration between Motol University Hospital and the Institute of Bioinformatics at Masaryk University in Brno, the portal aims to objectively inform the general public about children's cancers, their treatment and prognosis – http://detskaonkologie.registry.cz/.

1.2. RD awareness among the professional community and the general public

In 2015–2017, RD awareness improved through media presentations in radio, television, the press and especially on-line (www.vzacna-onemocneni.cz/media-o-vzacnych-onemocneni/clanky/clanky-v-tisku-2017.html). Social networks and on-line communication have had a significant impact and – due to their economic efficiency and their ability to reach the target groups – they have become the main communication channel of professional societies, patient organisations and individual projects (see the RDCZ Facebook portal). Overall, it is safe to say that the influence of the media in this area has increased markedly, as with other health issues. Also, editors in the various media have also become more knowledgeable. On the other hand, this positive trend increases the complexity of communication strategies for professional societies and patient organisations. Very important in this respect is professional cooperation between RDCZ and its expert advisers.

One of the most important opportunities to present rare diseases both to the professional community and the general public is the Rare Disease Day (www.rarediseaseday.org), which is held regularly on the last day of February. In the Czech Republic, RDCZ is the main coordinator of this event and has been authorised by Eurordis to use the symbols and supporting materials for this day. In 2015, 2016 and 2017, communication campaigns were implemented for this day using TV, radio, video screens, print, on-line and public relations campaigns. These activities were mostly targeted at

the general public. Patient organisations and NCCRD staff contribute significantly to these campaigns. The professional community was informed in professional journals, at lectures, seminars, etc. In 2015, NCCRD provided a series of 20 information lectures for paediatricians from all regions of the Czech Republic, as part of 8 regional meetings of the Association of General Practitioners for Children and Youth (www.detskylekar.cz/).

In 2015–2017, expert conferences (domestic and international within the purview of NCCRD), seminars and round tables were held and 4 meetings within the Health Committee of the Chamber of Deputies of the Czech Republic were organised in order to improve awareness among legislators. RDCZ also publishes an information newsletter on rare diseases (www.vzacna-onemocneni.cz/o-nas/zpravodaj-cavo.html), a number of professional societies and patient organisations publish their own newsletters focusing on specific RD (e.g. www.slg.cz, www.hematology.cz, www.linkos.cz. www.pediatrics.cz).

Awareness and understanding among public administration, the professional community and the general public have also been improved through activities of the Ministry of Health, namely through creating a system to communicate with patient organisations, establishing a patient rights support department within the organisational structure of the Ministry of Health, organising meetings of patient organisations at the Ministry of Health, and establishing the Patient Council to the Minister of Health – www.mzcr.cz/dokumenty/ministr-zdravotnictvi-jmenoval-predsednictvo-pacientske-rady_14570_1.html. Similarly, Patient Councils with the Czech VZP health insurance company (www.vzp.cz) or the Association of Health Insurance Companies of the Czech Republic (www.szpcr.cz), including seminars organised for the State Institute for Drug Control (www.sukl.cz), help improve awareness. University Hospital Brno staff who cooperate with NCCRD organised regular educational seminars.

2. RD education

RD issues have become a part of undergraduate and postgraduate education for doctors and non-medical health care professionals, and educational courses and seminars are held by individual specialised centres. In this area, NCCRD and RDCZ cooperate with the Institute for Postgraduate Medical Education (www.ipvz.cz) for non-doctors (e.g. bioanalysts in medical genetics) and the Coordination subject board for specialised education for doctors at Charles University (www.lf2.cuni.cz/specializacni-vzdelavani/organizacni-struktura/koordinacni-oborova-rada-kor-pro-

specializacni). While there has been an improvement compared to the previous period (1st National Action Plan for 2012–2015), RD education is not yet systemic and this specific issue needs to be expanded in the next period, especially in terms of education programmes for paediatrics, internal medicine and general practitioners.

As part of implementing the 2nd National Action Plan, NCCRD organised seminars and workshops for the professional community, and professional blocks relating to RD were included at congresses of the various medical disciplines (e.g. dermatology, nephrology, paediatrics, oncology). A number of research articles and reviews on RD have been published by NCCRD in the Czech and foreign press. Education in the area of rare diseases is a joint effort by professional societies, accredited departments, faculties of medicine and the Institute for Postgraduate Medical Education. A great achievement was the publication of a well-structured monograph entitled "Vzácné nádory v onkologii" (Rare tumours in oncology) in 2015 (www.martinus.cz/?uItem=206322).

In collaboration with colleagues from NCCRD and the 2nd Faculty of Medicine of Charles University at Motol University Hospital, RDCZ launched a project to educate fourth-year medical students on RD within medical genetics lectures. Patients or parents with patients participate directly in practical exercises for medical students, where they talk about their diseases, symptoms and experience with treatment, including social issues. In regular anonymous surveys, this project was praised by students as one of the most valuable ones. For the first time, medical students thus have an opportunity to encounter a specific RD diagnosis, which may help them significantly to improve early RD diagnosis in their future practice. In general, this improves awareness of the existence of RD among the professional community. In 2017, the project was also expanded in a pilot scheme to some other faculties of medicine in the Czech Republic (Palacký University in Olomouc or Masaryk University in Brno).

RD experts from NCCRD, patients or individual patient organisations (and RDCZ) gave presentations at professional conferences and seminars, mainly in collaboration with the Association of General Practitioners of the Czech Republic, the Professional Society of General Practitioners Association of Paediatric General Practitioners at Jan Evangelista Purkyně Czech Medical Association, the Association of General Practitioners for Children and Youth of the Czech Republic. These educational events were supported by activities within predefined projects of 3 Norwegian Grants at NCCRD.

3. RD prevention – preconception and prenatal screening

With respect to systemically implemented preconception screening, no activities were done to that end due to unresolved ethical, health and economic issues that are associated with this area. In the field of prenatal screening, recommendations for implementing the screening were unified by the Society of Medical Genetics and Genomics and the Czech Society of Clinical Biochemistry at Jan Evangelista Purkyně Czech Medical Association (www.slg.cz/stitky/doporuceni). Prenatal RD diagnosis has become an established part of care for pregnant women and is fully covered by public health insurance (www.slg.cz).

4. Improving RD screening and diagnosis

4.1 Expanding general laboratory neonatal screening

The screening programme was expanded in 2016 and the examination now includes 18 RD pursuant to the Methodological Instruction published in Ministry of Health Bulletin no. 6/2016, which had been prepared – among others – by colleagues from the National Coordination Centre for Neonatal Screening at General University Hospital in Prague (www.novorozenecky-screening.cz). In 2014, a multi-year prospective study of parental stress due to false positives in neonatal laboratory screening was also launched, the results will be evaluated in 2018. Currently, neonatal screening has a detection rate of approximately 1:1100 newborns according to latest KCNS data, which ranks the Czech Republic among the top European countries in this area (www.novorozeneckyscreening.cz/vysledky-ns-2012-19-10-2015-131436).

4.2 Development of foetal cardiology centres and 4.3. Association of expert genetic departments for RD diagnosis in cardiology

As part of birth defect screening, standard foetal echocardiography is carried out in the 20th week of pregnancy by the gynaecologist, a suspected birth defect or heart rhythm disorder is further examined at a specialised department by a paediatric cardiologist who, if necessary, carries out further monitoring and suggests treatment options. The examination is performed in a pregnant woman without prior preparation, between the 18th and the 22nd week of pregnancy. The examination is non-invasive and it is done transabdominally. Within the European Union, the Czech Republic is a leading country in terms of the detection rate of rare congenital heart defects in the

prenatal period. Foetal cardiology centres operate at Motol University Hospital, Brno University Hospital and Olomouc University Hospital and they require interdisciplinary collaboration between cardiologists, gynaecologists and medical geneticists (www.lubusky.com/clanky/98.pdf). At the Institute for Clinical and Experimental Medicine (IKEM) Cardiology Clinic, an expert department has been created for prenatal and postnatal RD, which operates nationwide. NCCRD continues to work with the Paediatric Cardiology working group led by colleagues from the Paediatric Cardiocentre of Motol University Hospital and CU 2nd Faculty of Medicine on the development of the foetal cardiology centres.

4.3. Quality and availability of RD diagnostic laboratories

In terms of the analytical quality of diagnostic laboratories, their accreditation according to ISO 15189.2013 has been further developed in accordance with Act No. 373/2011 Sb. and in cooperation with the Czech Institute for Accreditation (www.cia.cz). NCCRD worked with the Ministry of Health to amend Sections 28 and 29 of Act No. 373/2011 Sb. In connection with the development of RD genome diagnosis, including prenatal and preimplantation RD diagnosis. The Committee of the Society of Medical Genetics and Genomics at Jan Evangelista Purkyně Czech Medical Association has refined recommendations relating to genetic examination indications, and updated recommendations for the „Unbreakable limits for laboratories“ that carry out genetic examinations (www.slg.cz/system/files/nepodkrocitelne-meze-2013.pdf).

Professional recommendations have been issued with respect to informed consent before genetic testing, and the implementation of cytogenetic examinations for rare congenital developmental defects, and recommendations have been updated for good laboratory practices for selected rare diseases. As part of an amendment to Decree No. 134/1998 Sb., which publishes the list of health care services with point values, as amended, molecular genetic codes for reporting a reimbursement of genetic examinations for rare diseases were updated from 1 January 2018, in terms of stratifying the most frequent genetic examinations. In terms of accessibility, the network of genetic departments is adequate for the Czech Republic, with minimal regional differences. The Society of Medical Genetics and Genomics at Jan Evangelista Purkyně Czech Medical Association maintains an up-to-date database of departments which is available at https://new.slg.cz/pracoviste/.
In 2017, NCCRD helped develop the stratification of the reporting of reimbursement of genetic laboratory testing (spec. 816) from public health insurance for the most frequent genetic RD in cooperation with the VZP health insurance company and the Association of Health Insurance Companies.

5. Improving the availability and quality of care for RD patients

5.1. Centralisation, coordination and integration of care for RD patients

Unfortunately, this item was not accomplished in the period 2015–2017, despite resolutions and recommendations of the Interministerial and interdisciplinary working group on rare diseases at the Ministry of Health, as well as recommendations and calls of NCCRD, RDCZ and professional societies. Within the period under review, in 2015 a Decision was issued to the National Coordination Centre for Rare Diseases that – while not a provider of health services – plays a coordinating role for issues relating to rare diseases. As for other existing centres (centre for patients with cystic fibrosis, hereditary metabolic disorders, epidermolysis bullosa congenita), centres of highly specialised care ceased to exist due to the expiry of the period for which the status had been granted. Subsequently, the Ministry of Health did not announce a new call for centres of highly specialised care for RD pursuant to Section 112 of Act No. 372/2011 Sb.

The only centres of highly specialised care for RD patients that have been established under a call for applications for the status of a centre of highly specialised health care under Section 112 of Act No. 372/2011 Sb., are the centres that are part of the national oncology network. An up-to-date list of centres is available at https://www.linkos.cz/lekar-a-multidisciplinarni-tym/diagnostika-a-lecba/narodni-onkologicka-sit/. Complex oncology centres are involved in the diagnosis and treatment of rare oncological diseases pursuant to the provisions of the Ministry of Health Bulletin no. 5/2014 and no. 4/2015 (http://www.mzcr.cz/Legislativa/obsah/vestnik-mz_1768_11.html).

Nevertheless, the involvement of Czech specialist departments in the European Reference Networks for Rare Diseases is crucial and distinctly positive for the future (www.ec.europa.eu/health/ern_en). A total of 8 health service providers (organisations controlled directly by the Ministry of Health, mostly university hospitals) participated in ERN, including 26 professional departments. They were successfully involved in 17 professional ERN networks in the various RD areas. In this respect, the Czech Republic thus became the eighth most successful country in Europe and by far the most successful country in Central and Eastern Europe (i.e. the new EU member states known as EU13). Since the initial analysis and evaluation of the registered workplaces was done according to the strict criteria of the European Commission, it is a confirmation of the high quality of our departments focusing on RD diagnosis, treatment and research. This should allow for and simplify the process of announcing a call for centres of highly specialised care in the Czech Republic in 2018.

5.2. Best practices in the diagnosis and treatment of selected RD

Within the 2nd National Action Plan, the best practices for RD diagnosis were developed by NCCRD in collaboration with the Society of Medical Genetics and Genomics at the Jan Evangelista Purkyně Czech Medical Association and they are clearly presented on its website (www.slg.cz/stitky/doporuceni). The application of the latest findings in RD diagnosis and treatment into practice was supported through grants (www.gacr.cz, www.tacr.cz and www.azvcr.cz), and support was also provided for expanding the range of existing patient registers and establishing new ones, improving screening and diagnosis, and efforts to introduce standards of care for individual rare diseases. Finally, many projects were supported by the Ministry of Health through the subsidy policy chapter of National Action Plans and Concepts in 2015 and Health Care Development Projects in 2016 and 2017 (see the published results of subsidy programmes on the Ministry of Health website).

5.3. Effective and timely RD pharmacotherapy (orphan drugs) following up on internationally recognised standards and best practices

With effect from 1 September 2015, the terms „rare diseases“ and „orphan medicinal products“ (Section 11(1)(f) – www.zakonyprolidi.cz/cs/1997-48) were introduced into the Czech legal system (Act No. 48/1997 Sb., on public health insurance and amending and supplementing some related acts, as amended), thereby implementing Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products. In the period under review, efforts to reduce the duration of the administrative procedures for the approval of orphan medicinal product reimbursement failed, and this duration is still one of the longest in the EU (more than 2 years), despite the fact that an Eurordis recommendation sets the optimal duration for this procedure at up to 6 months. The State Institute for Drug Control (SÚKL) participated as an observer in the MoCA working group (Mechanism of Coordinated Access to orphan medicinal products; www.eurordis.org/content/Moca), which aims to promote earlier access to treatment. This group operates under the MEDEV European informal consultation platform (www.medev-com.eu). The latest available list of registered orphan medicinal product in the Czech Republic is available at – www.vzacna-onemocneni.cz/vzacna-onemocneni/2013-07-03-12-52-28/lecive-pripravky-pro-vzacna-onemocneni.html.

6. Improving the quality of life and social integration of people with RD

Patient organisations play an increasingly important role in the areas of availability of information, provision and arrangement of social services, early diagnosis, as well as record keeping and communication with patients. These activities are also of great help to doctors, other health care professionals and professional societies in raising awareness about prevention and screening possibilities, thus contributing to the early detection of some RD, and they also contribute significantly to establishing standards of care among patients. In the period under review, there was also significant improvement in terms of support for patient organisations, understanding their needs, and efforts to incorporate the views of patients/patient organisations into professional communication. In terms of communication between patients and the Ministry of Health, the situation clearly improved.

Regular patient meetings take place at the Ministry of Health and the Patient Council to the Minister of Health was established in late 2017 (www.mzcr.cz/dokumenty/pacientska-rada-jmenovani-clenu_14328_910_1.html). These activities have already translated into specific solutions and measures to help patients. The Unit of patients' rights support was set up within the organisational structure of the Ministry of Health (www.mzcr.cz/dokumenty/ministerstvo-pacientuzrizuje-pacientskou-radu-a-samostatne-oddeleni-podpory-pr_14310_3692_1.html) and the communication e-mail was set up: propacienty@mzcr.cz.

6.1. Introduction of the ICF system (ICF – International Classification of Functioning, Disability and Health)

Electronic supporting documents for ICF (http://www.uzis.cz/katalog/klasifikace/mkf-mezinarodni-klasifikace-funkcnich-schopnosti-disability-zdravi), the ICF Checklist electronic form and additional questionnaires aiming to objectivise functional status and self-sufficiency were published in the period of the 2nd National Action Plan (the WHODAS 2.0 questionnaire was published, and the translated Barthel test and SF-36 questionnaire are being prepared for publication). On 16 March 2017, the first of a series of planned meetings to support the implementation of ICF in the Czech Republic took place and it was attended by the Ministry of Health, the Ministry of Labour and Social Affairs, the Czech Statistical Office, and the Institute of Health Information and Statistics. Steps are being taken to introduce ICF as the official classification system in the Czech Republic through a

Czech Statistical Office Communication. A pilot study of the classification of selected patient groups according to their functional status using objectivising methods (including ICF) is being implemented by the Clinic of Rehabilitation Medicine of the 1st Faculty of Medicine at Charles University and General University Hospital in cooperation with other providers and the VZP insurance company (for details see www.mzcr.cz/obsah/mezinarodni-klasifikace-funkcnich-schopnostidisability-a-zdravimkf-_1982_3.html).

6.2. Educating the professional public about the ICF system

Every year, the Clinic of rehabilitation medicine of the 1st Faculty of Medicine at Charles University and General University Hospital organise courses on ICF use (www.is.cuni.cz/webapps/zzp/download/130172893). The Institute of Health Information and Statistics is going to publish supporting documents for the development of specific ICF clinical forms for specific diagnoses or situations (based on the ICF Core Sets international project; www.uzis.cz/system/files/u44/2017-11-07-13_Svestkova.pdf a http://www.icf-core-sets.org/).

6.3. Health and social services

With respect to the area of health and social services, it has to be noted that tasks in this area failed to be addressed adequately due to the complexity of the issues and the need for cooperation between several ministries, health insurance companies and patient organisations. In order to make cooperation between the Ministry of Health, the Ministry of Labour and Social Affairs and patients more effective, representatives of the Ministry of Labour and Social Affairs were invited to patient meetings at the Ministry of Health. The Ministry of Labour and Social Affairs works with the Ministry of Health and organisations of persons with disabilities to address the coordination of rehabilitation as a tool to improve the quality of life and achieve full social inclusion. Also, there has been an increase in care allowance which, in turn, improved the ability of dependent persons to secure an adequate range of services, i.e. this measure also positively affected a number of people with RD. In this context, an interministerial working group for addressing the issue of cross-sectional social and health services was set up – the aims is to continue work to further address the issue of long-term health and social care and to create conditions for the application of the principles of equity in health and social services.

7. Support for basic and applied research in the field of RD

7.1. Targeted support for RD science and research at the national and international level

In the period of the 2nd National Action Plan, projects were implemented within the Czech Health Research Council (www.azvcr.cz), which had taken over the role of the Internal Grant Agency (IGA) of the Ministry of Health. No calls relating specifically to RD have been announced to date. For a clear overview of the projects being implemented and the calls please visit www.azvcr.cz/podpora-vyzkumu. Similarly, basic research projects are implemented within the activities of the Grant Agency of the Czech Republic – www.gacr.cz and the Technology Agency of the Czech Republic – www.tacr.cz. At the end of 2017, successful collaboration with the Ministry of Education, Youth and Sports led to participation in the E-rare.eu international European projects – these are an initiative of the European Commission for international research in the field of RD, where national teams are funded from national resources within international consortia.

International EU grants are one of the areas in which participation in research consortia took place (the projects RD-connect.eu, RD-neuromics.eu and eurenomics.eu, the 7th Framework Programme of the EU, Horizon 2020 – VisionNMD, Solve-RD) in the area of RD. The implementation of the Norway Grants project entitled „National Coordination Centre for Rare Diseases at Motol University Hospital“ within the purview of NCCRD was successfully completed. The project's activities raised awareness and knowledge of RD among both the professional community and the general public, and helped implement the introduction of new methods of molecular-cytogenetic and genetic diagnostics for the purposes of prenatal and postnatal RD diagnosis, improve the pharmacoeconomic estimation of diagnostic and therapeutic costs for selected RD, improve complex patient care using experience from Norwegian health care services, and develop undergraduate and postgraduate teaching courses (https://nkcvo.cz/). This project was evaluated by the Norwegian Embassy in Prague as the most successful project in the Czech Republic for the past programming period of Norway Grants for the Czech Republic.

7.2 Developing cooperation between faculty and non-faculty research departments, at medical and non-medical faculties

In the long-term, NCCRD has been cooperating on joint projects with the Czech Academy of Sciences – Biocev and the Institute of Biotechnology in implementing RD projects in the field of molecular biology (www.ibt.cas.cz/vyzkum/laboratore/laborator-molekularni-patogenetiky/) and

with the Department of Genetic Toxicology and Nanotoxicology – Institute of Experimental Medicine of CAS, v.v.i., Prague (www.iem.cas.cz/cs/vyzkum/oddeleni/geneticke-toxikologie/).

7.3. Institutional support for the Czech Republic's international cooperation in RD research and development

Through the Ministry of Education, Youth and Sports, the Czech Republic actively participated in a rare diseases initiative under ERA-Net for Research Programmes on Rare Diseases in 2017, which allowed Czech research organisations to participate in its current call whose financing will start in 2018 (http://www.msmt.cz/vyzkum-a-vyvoj-2/iniciativa-v-oblasti-vzacnych-onemocneni-v-ramci-era-net). The E-Rare initiative was launched within the EU in 2006 to coordinate and deepen Member States' research activities relating to rare diseases, thereby complementing other European Commission initiatives in this area. Currently, the 3rd run of the initiative entitled "E-Rare-3" (2014–2019) is being implemented. From 2009, the "E-Rare 3" initiative will become part of the "European Joint Programme Cofund on Rare Diseases" that is being prepared by the European Commission. Preparations of the EJP RD programme were launched by the Commission in 2017, and the Czech Republic represented by the Ministry of Education, Youth and Sports will participate in the programme. This initiative is part of the EU Framework Program for Research and Innovation, Horizon 2020 (2014–2020), and it aims to support the coordination of national R&D activities related to rare diseases. The first call for proposals for international projects focusing on rare diseases is supposed to be announced in 2019. The EJP RD programme will be a key programme not only to support research and development projects, but also for other international cooperation activities in the area of rare diseases. This program will thus bring together all relevant stakeholders from among research organisations, research infrastructures, patient organisations, regulatory bodies and the corporate sector. The financing of EJP RD projects will be based on a combination of public funds provided by the European Commission and those EU Member States that chose to participate in EJP RD activities.

8. Harmonisation and development of data collection and biological sampling

8.1 National RD data collection, improvements in the data collection methodology and support for participation in international RD data collection projects

In the National Register of Congenital Malformations (NRVV; http://www.uzis.cz/registry-nzis/nrvv) there is an option to enter RD using OrphaCodes. The terminology relating to diseases was translated

into Czech in collaboration with NCCRD and Orpha.net and is available for download from the address: http://www.orphadata.org/cgi-bin/inc/product1.inc.php. Due to barriers when entering data (complicated handler programme, complicated questionnaire), the majority of the diseases are currently entered using the ICD-10 code. This classification was also provided to health insurance companies for the stratification of services for molecular genetic diagnosis (www.vzp.cz/o-nas/aktuality/informace-pro-poskytovatele-hrazenych-sluzeb-laborator-lekarske-genetiky-a-sdilene-odbornosti-pro-rok-2018).

Collection of data on rare diseases was mainly limited by the insufficient detail of the International Classification of Diseases (ICD-10), which is commonly used as a classification and coding tool, and by the absence of a minimum data model for RD data collection. The Institute of Health Information and Statistics (IHIS) worked hard to address both problems through introducing more detailed coding and classification systems (OMIM, Orphacodes, SSIEM) into data collection of the National Register of Reproductive Health (NRRZ) and into the Data Standard of the Ministry of Health of the Czech Republic. The plan included partial translation, support and documentation for these tools and the intensification of international cooperation in their use. A representative of IHIS took part in foreign seminars on the topic. The use of the Orphacodes coding system, which will be included as a whole in the upcoming 11th revision of the ICD, should help with the transition to ICD-11. Using foreign models and methodologies, the IHIS has created a minimal data model for RD data collection (MDS RD – minimal data set). The MDS RD should serve as a model for creating any new (and modifying existing) registers, databases and data models for electronic health records that can be potentially used in identifying cases of rare diseases.

Within the various medical specialities, RD registers were installed under the auspices of individual professional societies (e.g. the cardiology registers KARDIO-ICD, KATAB, REPACE, REPLY, TAVI, register of rare diseases of erythron, and register of the Hereditary Ataxia Centre) in 2012–2014. A joint Czech and Slovak registry of birth defects with pulmonary hypertension was created.

8.2. Legal framework for data collection and biological sampling

The legal aspects of sampling for RD diagnosis and research are arranged in accordance with Sections 28 and 29 of Act No. 373/2011 Sb. and Act No. 101/2000 Sb., on the protection of personal data and on amendments to some acts, as amended.

9. Support and strengthening of the role of RD patient organisations

9.1. Collaboration with patient organisations in the Czech Republic, the development of cooperation with the EURORDIS.org European patient organisation

The Rare Diseases Czech Republic (RDCZ) is the umbrella organisation for RD patient organisations. RDCZ currently brings together 30 patient organisations and patients representing more than 50 RD diagnoses. RDCZ is a member of EURORDIS.org, a European organisation representing RD patients, and it represents the Czech Republic at regular CNA (Council of National Alliances for RD) meetings. RDCZ participates in EURORDIS.org meetings and it then applies the acquired knowledge and experience in its activities in the Czech Republic. In the Czech Republic, RDCZ and NCCRD are members of the Interministerial and Interdisciplinary Working Group on Rare Diseases at the Ministry of Health, the Chairperson of RDCZ was appointed a member of the Patient Council to the Minister of Health. The members of this council also include representatives of 5 other RD patient organisations (www.vzacna-onemocneni.cz).

9.2. Support for the activities of patient organisations

Several activities were created or developed to support the activities of patient organisations. The most useful ones that are also acknowledged by patient organisations include the Academy of Patient Organisations (APO, www.pacientskaakademie.cz/). This educational project has had a significant effect on the growing professionalization of patient organisations. However, educational projects for patients and patient organisations are also being prepared by other institutions or societies, often in the pharmaceutical field.

RDCZ and several patient organisations have successfully passed the “Seal of Reliability” audit organised by the Association of Public Benefit Organisations (AVPO, www.avpo.cz/wp-content/uploads/2016/05/VZ-AVPO-%C4%8CR-2015.pdf). This system not only helps identify non-profit organisations that apply all the proper processes of good organisation management but it also helps organisations as feedback. Patient organisations have an opportunity to participate in grants that support their activities, which are announced by the Ministry of Health, the Government of the Czech Republic, and in the area of social care also by the Ministry of Labour and Social Affairs. However, the number of high-quality and professionally managed private grants of individual successful societies keeps growing.

10. Interministerial and interdisciplinary collaboration

10.1. Coordination of the implementation of tasks arising from the National Strategy for the Prevention of Rare Diseases for 2010–2020 and the respective action plans

In 2010, the Interministerial and interdisciplinary working group on rare diseases was set up at the Ministry of Health, comprising representatives of the Ministry of Health, the Ministry of Labour and Social Affairs, the VZP health insurance company, the Association of Health Insurance Companies of the Czech Republic, the professional societies of the Jan Evangelista Purkyně Czech Medical Association, the WHO Office in the Czech Republic, IHIS, RDCZ patient organisations and other experts. Between 2015 and 2017, this working group continued to coordinate the implementation of tasks and activities under the National Strategy for Rare Diseases for 2010–2020 and the National Action Plan for 2015–2017. Within the implementation of the 2nd National Action Plan, the main accomplishments include the development of the procedure and the successful participation of Czech institutes and departments in ERN networks (see above).

11. International RD cooperation

11.1. International RD cooperation, exchange of experience, data and information

In 2017, NCCRD organised several international workshops for EU projects – 3Gb-test.eu, Human Genome Variation Society in Prague (www.vep.variome.org) on the topic of RD genetic diagnosis. There was continued cooperation with EAHC DG Sanco and the grant consortia RD-Connect.eu a RD-Action.eu). Also, there was continued cooperation with the International Rare Disease Research Consortium, where the Czech Republic has a representative on the Diagnostic Committee (www.irdirc.org) and with the Committee for Orphan Medicinal Products of the European Medicines Agency (COMP EMA – www.ema.europa.eu/ema/index.jsp?curl=pages/about_us/general/general_content_000263.jsp).

11.2. Cooperation within the European Commission's EuroPlan/Eurordis project

RDCZ cooperated with the European RD patient organisation – Eurordis (see item 9.1.), and together with NCCRD it cooperated with the Frambu.no Norwegian centre for rare diseases. Each year, several patient organisations, patients and experts participated in educational patient seminars and meetings in Frambu, all thanks to a grant under the Norway Grants. As part of the Europlan project, a lecture on the topic "Access to Specialised Care for European Citizens living with a Rare Disease" was

given by Eurordis representative in the autumn of 2017, which highlighted the importance of the emerging European Reference Networks for rare diseases. This lecture took place as part of a meeting of the Interministerial and interdisciplinary working group and was attended by Czech representatives of providers who participate in European reference networks for rare diseases.

11.3. Standard classification of rare diseases for the planned ICD-11 revision and in cooperation with the Orphanet consortium

In the Czech Republic, the 10th revision of the International Classification of Diseases is currently used for the collection of data in the health sector in terms of diagnoses. ICD-10 does not allow accurate and effective RD encoding, i.e. the identification of individual nosological units of rare diseases in reported data, because the information is only recorded at the level of disease groups that serve as umbrellas for individual (quite heterogeneous) diseases. At the international level, a leading system for RD coding is the OrphaCode system that was created based on the terminology system of the Orphanet consortium. Orphanet cooperates with the International Health Organisation and units of the Orpha Codes system will probably be integrated into the 11th revision of the International Classification of Diseases.

Since 1997, IHIS allows reporting OrphaCodes (along with other classification systems – OMIM and SSIEM) in the National Register of Congenital Malformations. At the same time, the entire RD terminology of Orphanet was translated into Czech in 2016 and 2017. IHIS is considering ways to further enable accurate identification using OrphaCodes in other data collections, and to integrate at least some RD terms into ICD-10 documents (e.g. into the alphabetical list modelled on the German Alpha-id system of enhanced terminology).

12. National Action Plan for Rare Diseases for 2018–2020

In recent years, it has been clear that in terms of rare diseases, the National Strategy for Rare Diseases for 2010–2020 failed to be implemented systemically. A key element of a systemic RD approach that failed to be accomplished on the part of the health care system is the establishment of centres of highly specialised RD care pursuant to Section 112 of Act No. 372/2011 Sb. Therefore, the establishment of centres of highly specialised RD care is one of the priority tasks of the new National Action Plan for 2018–2020.

In the Czech Republic, the overall approach to addressing RD issues is starting to lag behind EU-level developments and trends in this area. One of the reasons is the failure to acknowledge the

importance of the entire RD area within the health care system, public administration, benefits to society, and of course, benefits to RD patients themselves, who account for up to 5 % of our total population. Other reasons include the fact that the Czech Republic has one of the slowest approaches to the reimbursement of new medicines (e.g. highly innovative medicinal products/orphan medicinal products) in the EU, there are no centres of highly specialised RD care, patients' access to appropriate standards of RD care is deteriorating, patients often lose the system of high-quality complex care as they transition to adulthood where this issue is not addressed virtually at all. The complex RD issue in the Roma population also remains unresolved.

On the other hand, in recent years there have been several very positive activities in the field of RD that need to be highlighted. These mainly include the participation of Czech professional RD departments in the European Reference Networks for RD, and the active approach and role taken by public administration in this area. There has been considerable success in developing and promoting cooperation between public administration, professional societies and patient organisations. These positive activities can provide a very good basis for accomplishing the objectives of the 3rd National Action Plan for Rare Diseases.

12.1. Overall objectives for the upcoming period

The main objective of the National Action Plan for Rare Diseases for 2018–2020 is to implement the diagnosis and treatment of rare diseases in a way that provides all RD patients with access to indicated and high-quality health care, ensures an option for comprehensive care and treatment in accordance with the latest findings in the RD field, on the basis of equal access, treatment and solidarity.

Therefore, the National Action Plan for Rare Diseases for 2018–2020 places emphasis mainly on supporting timely RD identification and diagnosis, ensuring the development of comprehensive care and the centralisation of care for RD patients through establishing centres of highly-specialised RD care and engaging these centres in emerging ERNs, and also on ensuring transition from paediatric to adult care while providing the same standards of care and access to that care. In addition, emphasis is placed on improving awareness of RD issues, developing undergraduate and postgraduate education for doctors and non-medical health care personnel, and developing cooperation between professional societies and patient organisations. It will be necessary to focus on issuing standards of care for RD patients, in line with latest findings and in cooperation between the various disciplines.

For RD, it will also be important to participate in international research projects supported by the Ministry of Education, Youth and Sports (e.g. E-Rare, EJP RD, Horizon 2020), Czech research projects within the Czech Health Research Council, and the development of cross-border diagnostic and therapeutic care, in line with the provisions of the Directive on the application of patients' rights in cross-border healthcare (2011/24/EU), which was transposed into Czech legislation in 2014.

In order to improve the quality of life and social inclusion of RD patients, it will be important to reorganise and specify social beds, rehabilitation facilities and long-term care beds with trained personnel and special equipment. It will also be necessary to support the development of palliative care for RD patients. Attention needs to be given to effective and timely RD pharmacotherapy following up on internationally recognised standards and best practices, as well as increased education and awareness on RD issues among both the professional community and the general public. The objectives, tasks, deadlines, outputs, coordinators, cooperating entities, and assumptions for accomplishing the National Action Plan are outlined in the table. In cases were accomplishing a specific task would have an impact on public budgets, it will include an analysis of the financial impacts on these budgets, which will be refined during the preparation of the implementation of the various individual measures under this plan, in cooperation with the relevant department of the Ministry of Health and health insurance companies.

12.2. Priorities for the upcoming period

- Establishing centres of highly-specialised RD care, linking these centres to European RD care structures, supporting and developing their co-financing
- Providing conceptual support for Czech departments participating in ERN
- Providing the conceptual aspects required for timely access to care and treatment, timely and effective access to new treatment options in line with access in other developed EU countries
- Ensuring and securing comprehensive care for RD patients during transition from paediatric to adult care
- Supporting and developing RD education and training at all levels of doctors and non-medical health care professionals
- Potentially expanding neonatal RD screening, and preparing recommendations for preconception RD screening

- Engaging Czech structures in international RD research projects
- A system for adopting existing standards of care based on latest findings and implementing and applying them in practice in a targeted manner, strengthening international cooperation in line with ERN recommendations
- Developing cooperation between patients, patient organisations and the professional public, including support for early diagnosis
- Promoting the application of patients' rights to the provision of highly-professional care within the EU (Directive 2011/24/EU on the application of patients' rights in cross-border healthcare)
- Adopting the Orphanet international classification – Orphacodes into the health care system
- Developing and providing information support for RD centres and ERN (e.g. helplines)

National Action Plan for Rare Diseases for 2018–2020						
Objectives	**Tasks**	**Deadline**	**Output**	**Coordinator**	**Cooperation**	**Assumptions**
1. Improving RD awareness	*1.1 Operating, developing and updating RD web portals, on-line communication and consulting centres* *1.2 Developing and supporting outputs of latest RD findings from the activities of ERN and Centres of highly specialised RD care*	*on-going*	*Web portals, on-line communication, creation of RD materials, operation of helpline; coordination of joint activities of ERN and Centres of highly specialised RD care*	*RDCZ, Motol NCC*	*RD centres, RDCZ, ERN, KCNS, IWG, PS, PO, IHIS, HIC*	*Subsidy schemes*
	1.3. RD awareness among the professional community and the general public	*on-going*	*Seminars, conferences, communication and educational campaigns, cooperation with the media, research on RD awareness, opinion polls, a comprehensive nationwide campaign to promote RD awareness among both the professional community and the general public*	*RDCZ, Motol NCC*	*RD centres, RDCZ, ERN, KCNS, IWG, PS, PO, IHIS, HIC*	*Subsidy schemes*

2. RD education	*2.1 Professional community – (undergraduate and postgraduate education for doctors and non-medical health care professionals, lifelong learning in this field)* *2.2 Professional community – "Early diagnosis" project* *2.3 General public – RD education for PO representatives*	*on-going*	*Expansion of undergraduate and postgraduate RD courses, creation of a system for undergraduate and postgraduate RD education, dedication of professional publications, PO involvement in the RD education system, optimised procedures and support for early RD detection at primary care doctors*	*RDCZ, Motol NCC*	*RD centres, RDCZ, ERN, IPVZ, KCNS, FoM, IWG, MH, PS, PO, IHIS, HIC*	*Subsidy schemes*
3. Primary and secondary RD prevention	*3.1. Preconception and prenatal RD screening*	*on-going*	*Draft methodology for preconception and prenatal RD screening*	*ERN, Motol NCC, PS*	*RD centres, RDCZ, ERN, IPVZ, KCNS, FoM, IWG, MH, PS, PO, IHIS, HIC*	*Subsidy schemes Public health insurance*
4. Improving RD screening and diagnosis	*4.1. Possible expansion of nationwide neonatal screening (NS) to include early diagnosis of additional RD*	*on-going*	*Proposal for NS optimisation, preparation of NS methodology, pilot projects, evaluation of NS outputs to date*	*KCNS, PS*	*RD centres, RDCZ, ERN, IPVZ, KCNS, FoM, IWG, MH, PO, IHIS, HIC*	*AZV, subsidy schemes, public health insurance*
	4.2. Quality of RD	*on-going*	*Information for laboratories*	*Motol NCC, PS*	*RD centres,*	*Subsidy*

	diagnostic laboratories		*preparing for accreditation under ISO 15189 and Act No. 373/2012 Sb.* *Information on the availability of laboratory test methods for RD*		*RDCZ, ERN, IPVZ, KCNS, FoM, IWG, MH, PO, IHIS, HIC*	*schemes*
5. Improving the availability and quality of care for RD patients	*5.1. Establishment of centres of highly specialised RD care pursuant to Section 112 of Act No. 372/2011 Sb., Centralisation, coordination and integration of care for RD patients* *5.2. Supporting and developing active participation in international activities of ERN, involving patients in E-PAG*	*on-going*	*Granted status of highly specialised RD centres according to the groups of diagnoses (according to the ERN classification), support and development of cross-border care and systemic provision of care within ERN. Created facilities and administrative support for ERN operation.* *Support for patient involvement in E-PAG within individual.*	*ERN, IWG, MH Motol NCC, HIC*	*RDCZ, PS*	*Subsidy schemes, public health insurance*
	5.3. Best practices in the diagnosis and treatment of selected RD (including nursing practices)	*on-going*	*Best practices, methodologies, standards, pilot projects on selected RD groups, cooperation between PS and PO in developing RD standards, procedures and materials*	*RD centres, ERN, PS*	*RDCZ, KCNS, Motol NCC, PO, HIC*	*Subsidy schemes*
	5.4. Effective and timely pharmacotherapy using	*2019*	*Systemic setting of the provision of timely access to new treatment for*	*RD centres, ERN, MH, PS, SÚKL, HIC*	*RDCZ, Motol NCC*	*Subsidy schemes*

	orphan drugs, following up on internationally recognised standards and best practices, including ensuring the timely availability of comprehensive RD treatment at the level of EU standards		*orphan drugs, an analysis and design of effective RD pharmacotherapy (improving the accessibility and effectiveness of care, monitoring the effectiveness of treatment)*			
6. Improving the quality of life and social integration of people with RD	*6.1. Introducing the use of the ICF classification and other functional status assessment tools into wider clinical practice*	*on-going*	*Feasibility study for functional status data collection via ICF, ICF management and updates, creation of tools for recording functional status*	*RD centres, ERN, MH, PS, HIC, IHIS*	*MLSA*	*Subsidy schemes*
	6.2. Educating the professional public about the ICF system	*on-going*	*Seminars, conferences, educational programmes, methodologies*	*MLSA, MH, IHIS*	*MLSA*	*Subsidy schemes*
	6.3. Health and social services („health and social boundaries“)	*on-going*	*Proposal for a systemic solution for long-term RD health and social care, feasibility study*	*RDCZ, MH*	*RD centres, RDCZ, ERN, PS, PO, HIC*	*Health insurance, financing for social services*
	6.4. Developing and supporting palliative care for RD	*on-going*	*Provision and development of palliative care for RD patients*	*RD centres, ERN, FoM, FoS, Motol NCC, PS*	*RD centres, RDCZ, ERN, PS, PO, HIC*	*Subsidy schemes*
7. Support for basic	*7.1. Targeted support for*	*on-going*	*Research, projects, studies*	*RD centres, ERN,*	*CAS, AZV*	*Grant funds*

and applied research in the field of RD	*RD science and research: at the national level* *7.2 Developing cooperation between faculty and non-faculty research departments and medical and non-medical faculties*			*FoM, FoS, Motol NCC, PS*	*ČR, GAČR, MH, MEYS, TAČR*	
	7.3. Institutional support for the Czech Republic's international cooperation in RD research and development	*on-going*	*Research, projects, studies, involvement of the Czech Republic in the E-Rare project and the EJP RD programme)*	*RD centres, ERN, KCNS, FoM, Motol NCC, PS, IHIS*	*MEYS, MH*	*DG-Sante, E –rare, Horizon 2020*
8. Harmonisation and development of RD data collection and biological sampling	*8.1. National RD data collection, improving the methodology for RD data collection, supporting participation in international RD data collection projects*	*on-going*	*Data collection, statistics, analyses, IHIS methodology, international cooperation*	*MH, IHIS*	*MEYS, MH*	*Subsidy schemes*
	8.2. Legal framework for data collection and biological sampling	*2018*	*Regulation of RD data collection and biological sampling in accordance with GDPR*	*RDCZ* *Motol NCC, PO*	*RD centres, ERN, PS*	*Subsidy schemes*
9. Supporting and	*9.1. Collaboration with*	*on-going*	*Projects, seminars, conferences,*	*RDCZ, MH, Motol*	*RD centres,*	*Subsidy*

strengthening the role of RD patient organisations	*patient organisations in the Czech Republic, the development of cooperation with the Eurordis.org European association of patient organisations*		*cooperation with the media*	*NCC, PO*	*ERN, PS*	*schemes*
	9.2. Supporting the activities of patient organisations *9.3. Supporting the functioning and operation of patient organisations (websites, translations of professional materials, etc.)*	*on-going*	*Seminars, conferences, cooperation with the media* *Ensuring the operation and administrative activities of PO Operation of PO websites, support for PO functioning*	*MH*	*RD centres, ERN, PS*	*Subsidy schemes*
10. Interministerial and interdisciplinary collaboration	*10.1. Coordinating the implementation of tasks arising from the National Strategy for Rare Diseases for 2010–2020 and the respective action plans* *10.2. Providing the*	*on-going*	*Activities of the IWG RD, interim information and reporting on the implementation of NAP 2018–2020*	*RD centres, ERN, Motol NCC, IHIS*	*RDCZ, RD centres, ERN, Motol NCC, PS, PO*	*Subsidy schemes*

	conceptual aspects required for timely access to care and treatment, timely and effective access to new treatment options (Orphans) in line with access in other developed EU countries and the legislative regulation *10.3. Coordinating ERN activities and developing the information portal*					
11. International RD cooperation	*11.1. International RD cooperation, exchanging experience, data and information*	*on-going*	*International cooperation, projects*	*RD centres, ERN, MH, Motol NCC, HIC, IHIS*	*RDCZ, PS PO, IHIS*	*Subsidy schemes*
	11.2. Standard classification of rare diseases for the planned ICD revision, in cooperation with the Orphanet, introducing Orphacodes and SSIEM	*on-going*	*ICD-11, implementation of Orphacodes*	*RD centres, ERN, MH, Motol NCC, HIC, IHIS*	*RDCZ*	*Subsidy schemes*

13. Acronyms used

CAS – Czech Academy of Sciences (www.avcr.cz)
AZV ČR – Czech Health Research Council (www.azvcr.cz)
RDCZ – Rare Diseases Czech Republic (www.vzacna-onemocneni.cz)
RD centres – Centres of highly-specialised care established pursuant to Section 112 of Act No. 372/2011 Sb.
ČLS JEP – Jan Evangelista Purkyně Czech Medical Association (www.cls.cz)
CZSO – Czech Statistical Office
DG Sante – www. ec.europa.eu/info/departments/health-and-food-safety_en
EJP – EJP - Cofund - European Joint Programme
E-PAG – Eurordis Policy Action Groups - www.eurordis.org/content/policy-action-group-pag
E-rare – ERA-Net for Research Programmes on Rare Diseases of the European Commission (www.e-rare.eu)
ERN – Czech partners to the European Reference Networks for rare diseases - www.mzcr.cz/Odbornik/dokumenty/evropska-referencni-sit-pro-vzacna-onemocneni_13637_3715_3.html (April 2017)
EU – European Union
Eurordis – European association of patient associations for rare diseases (www.eurordis.org)
GAČR – Czech Science Foundation (www.gacr.cz)
Horizon 2020 – 8th Framework Programme of the European Commission (ec.europa.eu/programmes/horizon2020/)
KCNS VFN – Coordination Centre for Neonatal Screening at General University Hospital (www.novorozeneckyscreening.cz)
FoM – Faculties of medicine at Czech universities
LPVO – orphan drugs (as per Section 11(1f) of Act No. 48/1997 Sb., as amended)
ICF – International Classification of Functioning, Disability and Health (www.who.int/classifications/icf/en/)
ICD – International Classification of Diseases (http://www.uzis.cz/cz/mkn/index.html)
IWG – Interdepartmental and interdisciplinary working group for rare diseases at the Ministry of Health
MLSA – Ministry of Labour and Social Affairs (www.mpsv.cz)
MEYS – Ministry of Education, Youth and Sports (www.msmt.cz)
MH – Ministry of Health (www.mzcr.cz)
NG – Norway Grants (eeagrants.org/Who-we-are/Norway-Grants)
NCCRD – National Coordination Centre for Rare Diseases at Motol University Hospital (www.nkcvo.cz)
NSVO – National Strategy for Rare Diseases for 2010–2020
Orphanet – www.orpha.net
PS – Professional societies at Jan Evangelista Purkyně Czech Medical Association
FoS – Faculties of science at Czech Universities
PO – patient organisations associated under RDCZ
SLG ČLS JEP – Society of Medical Genetics at Jan Evangelista Purkyně Czech Medical Association
SSIEM- Society for the study of inborn errors of metabolism (www.ssiem.org)
SÚKL – State Institute for Drug Control (www.sukl.cz)
TAČR – Technology Agency of the Czech Republic (www.tacr.cz)
IHIS – Institute of Health Information and Statistics (www.uzis.cz)
RD – Rare diseases
WHO CZ – World Health Organization office in the Czech Republic (www.mzv.cz/mission.geneva/cz/odborne_organizace/ostatni_agendy/index.html)

Act No. 372/2011 Sb., on health care services and the conditions under which they are provided (Health Services Act), as amended – Act No. 372/2011 Sb.
Act No. 373/2011 Sb., on specific health care services, as amended – Act No. 373/2011 Sb.
HIC – health insurance companies (www.vzp.cz, www.szpcr.cz)