2nd Czech National Action Plan for Rare Diseases

Report on the Fulfilment of Tasks from the National Action Plan for Rare Diseases for 2012–2014 and the National Action Plan for Rare Diseases for 2015–2017.

Introduction:

Rare diseases (hereinafter "RD") are a clinically heterogeneous group of approximately 6000–8000 various diseases whose common feature is the very low prevalence of each of these diseases in the general population (i.e. affecting fewer than 1 in 2000 inhabitants). In case of incorrect or late diagnoses of these diseases, irreversible damage to health or death of patients often occurs. RD encompass all groups of diseases and account for up to 5 % of morbidity and mortality in early childhood.

On 14th June 2010, by Government Resolution No. 466, the Government of the Czech Republic approved the "National Strategy for Rare Diseases for 2010–2020" summarizing the issue of rare diseases from the perspective of both the EU and the Czech Republic and proposed key objectives and measures to improve the situation in this area in the Czech Republic. The National Strategy for Rare Diseases for 2010–2020 aims to ensure the timely diagnosis of and availability of adequate treatment for RD, to coordinate and centralise effective RD patient care, improve education and awareness among both the medical community and the general public in this area, cooperation at the national and international levels as well as the improvement of RD identification within the International Classification of Diseases system, as well as the development of European cooperation. The "National Action Plan for Rare Diseases for 2012–2014" specifies priority tasks and activities that allow for the gradual implementation of the objectives and measures of the National Strategy. The sub-tasks, tools, accountability, deadlines, funds and performance indicators for specific tasks of the National Action Plan were set so as to be practicable in the period in question and so as to have a minimal financial impact on the state budget and public health insurance.

In 2012–2014, the fulfilment of RD tasks and activities concerning the following was imposed:

- the improvement of RD awareness among the professional community and the general public;
- education in the field of RD, including the incorporation of specific RD-related areas of problems into undergraduate and postgraduate education for healthcare professionals;
- RD prevention with a focus on the development of preconception and prenatal diagnoses;
- increase the number of RD diagnosed as part of neonatal screening in the laboratory;
- improvement of the availability and quality of care for RD patients, making diagnosis and treatment more efficient and ensuring that all RD patients have equal access to indicated and high-quality healthcare;

- improvement of the quality of life and social integration of individuals suffering from RD;
- support of science and research in the field of RD, development of basic and applied research;
- harmonisation and development of data collection and biological sampling within RD at both the national and regional levels;
- support and enhancement of the role of RD patient organisations
- Inter-ministerial, interdisciplinary and international cooperation in the field of RD.

The National Action Plan for Rare Diseases for 2015–2017 specifies priority tasks and activities to allow for further implementation of the objectives and measures of the National Strategy in this period. The tasks and activities continually build on the priority tasks fulfilled in 2012–2014 and were set so as to be practicable in the given period and so as to have a minimal financial impact on the state budget and public health insurance.

Report on the Fulfilment of Tasks ensuing from the National Action Plan for Rare Diseases for 2012–2014

1. Improving RD awareness

1.1. Operating and updating web portals for RD

As part of improving awareness in the field of RD, web portals were operated and created. Websites of individual specialized centres for RD treatment were created, and a comprehensive web portal for RD (www.vzacnenemoci.cz) was operated, which aims to raise general awareness of rare diseases among the general public as well as among professionals providing healthcare to patients with these diseases.

www.novorozeneckyscreening.cz is a web portal intended for the general public and the professional community that provides detailed information on the neonatal screening programme aimed at identifying severe RD in newborns. The portal is divided into two parts, one for the general public, especially for parents who are directly concerned with neonatal screening, and the other part is for the professional community.

The www.ebcentrum.cz and www.debra-cr.cz portals for RD in the field of dermatology are intended for the general public as well as the professional community. They list basic information on the EB Centre (EB Centrum ČR is a specialised unit created in 2001 at the Children´s Dermatology Department of the University Hospital Brno in the Children´s Hospital in Černá Pole) and list the contact details of individual specialists. The EB Centre portal is linked to the DEBRA ČR website, which is intended for patients, their families and the general public.

The Czech Republic participated in the European project Orphanet (www.orphanet.cz), and its Czech version (www.orphanet.cz) was expanded. News, documents and information on events of national interest related to RD, medicinal products for RD and an overview of services provided in the Czech Republic are available at the Orphanet website for the Czech Republic.

The national registry of rare disease medical tests (www.registr-raritnich-vysetreni.cz) has been developed, which includes a subgroup of rare congenital developmental defects at the www.vrozene-vady.cz portal. The www.vzacna-onemocneni.cz portal was operated as a website of Česká asociace pro vzácná onemocnění (Rare Diseases Czech Republic - RDCZ), associating RD patient associations and individual patients. The portal contains an overview of their interests and promotes awareness of problems related to RD among healthcare professionals, officials at the national and international levels and the public.

The project of establishing the Czech National Informational and Educational Portal on Childhood Cancers was implemented, created in cooperation with Motol University Hospital and the Institute of Bioinformatics at Masaryk University in Brno, with the aim to provide objective information to the general public on childhood cancers, their treatment and prognoses.

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

The period from 2012 to 2014 saw an increase in the awareness about RD via the media (the radio, television and the press). Promotional video documentaries concerning RD were, such as the documentary film entitled “Vzácná onemocnění” (Rare diseases), a series of lectures and seminars for the professional community and the general public, as well as expert interviews on Česká televize and Český rozhlas. Articles on RD, quality of life and the social inclusion of rare disease patients were published in daily newspapers and medical media. It would be beneficial to engage in activities in this area in the next period as well.

In cooperation with patient organisations and Eurordis.org, the European alliance of patient organisations, the World and European Rare Disease Days were publicised. The Czech umbrella patient organization Rare Diseases Czech Republic (RDCZ) published Bulletin and organised member reunions. Seminars on RD were organised in the Chamber of Deputies of the Czech Republic

2. Education in the field of RD

RD issues are a part of undergraduate and postgraduate educational programs for doctors and non-medical healthcare professionals, and specific educational courses and seminars are held at individual specialised centres. However, the education has not been systematic so far and it is therefore necessary to extend the specific area of problems connected with RD in educational programmes for general medical practice and internal medicine in the upcoming period. As part of implementing the National Action Plan, seminars and workshops were organised for the professional community, and professional back-to-back seminars on RD were scheduled at congresses in the various medical disciplines. A number of research articles and reviews on RD were published in the Czech and foreign press. Education in the area of RD is a joint effort of professional societies, accredited departments, faculties of medicine and the Institute for Postgraduate Medical Education. Two printed publications on RD were

published - “Vzácná onemocnění v kostce” (Rare diseases in a nutshell) and a special edition of “Postgraduální medicína” (Postgraduate medicine).

RDCZ implemented the project entitled “Early diagnosis of rare diseases”, intended for medical practitioners for children and youth and to general medical practitioners. The vast majority of Czech medical practitioners for children were approached, and lectures on RD took place in cooperation with the Professional Association of Medical Practitioners for Children. The poster entitled “Včasná diagnostika” (Early diagnosis) was distributed into their practices.

The EB Centre and DEBRA ČR held seminars for paediatricians and general practitioners in 2013 and 2014. The conference on children´s dermatology by the Czech dermatological society of the Czech medical society of Jan Evangelista Purkyně focused on rare diseases in dermatology.

The following help-mail was launched: help@vzacna-onemocneni.cz. For specific information on RD, it is possible to contact this email intended for doctors and the public. As far as expertise is concerned, the operation of the informational e-mail is provided by the National Coordination Centre for Rare Diseases in cooperation with other centres and respective experts from the entire Czech Republic.

3. RD prevention – preconception and prenatal screening

As far as systemically implemented preconception screening is concerned, no desirable activities were carried out due to unresolved ethical issues associated with this area. As far as pre-natal screening is concerned, unified recommendations on its implementation were made by the professional Society of Medical Genetics of CzMA of Jan Evangelista Purkyně. Furthermore, the DNA diagnostics of RD in the Czech Republic and its connection to international initiatives is being developed. Pre-natal diagnostics, or, as the case may be, the treatment of congenital cardiac defects, has become an established aspect of care for pregnant women. The EB Centre implements

pre-natal and preimplantation genetic diagnoses in cooperation with gynaecological clinics and centres of assisted reproduction in the Czech Republic. As part of comprehensive genetic counselling, the EB Centre carries out genetic screening of relatives of patients with EB (Epidermolysis Bullosa). In 2012–2014, a network of echocardiology laboratories was created to cooperate in the early diagnosis of pulmonary hypertension syndrome.

4. Improvement of RD screening and diagnoses

4.1 The extension of the population’s neonatal screening by early diagnoses of RD

In 2014, a pilot study entitled “Optimization of neonatal screening for inherited metabolic disorders in 2011–2013” conducted by Ústav dědičných metabolických poruch (the Institute for Inherited Metabolic Disorders) was completed. The study, conducted as part of a grant

project in the Internal Grant Agency of the Ministry of Health, focused on extending the population's neonatal screening by other inherited metabolic disorders and developing second-stage methods for neonatal lab screening focused on reducing false positive results. The extension of the screening programme to a total of 15 inherited metabolic disorders as of 1st January 2015 was approved by the Inter-ministerial and interdisciplinary working group for rare diseases of the Ministry of Health of the Czech Republic and by the health insurance companies officials, two new medical services were included in decree No. 134/1998, which governs the list of medical services with their scores. The extension of the list of screening examinations shall be given in the "Methodological Guideline on the Provision of neonatal lab screening and after-care" to be published in the Journal of the Ministry of Health. The following diseases were recommended for extension in existing neonatal lab screening: type one citrullinemia, argininemia, pyridoxine non-responsive form of cystathionine-b-synthesis deficiency, methyltetrahydrofolatereductase deficiency and biotinidase deficiency.

Public awareness is a significant part of the screening programme. In 2014, a questionnaire was conducted on the awareness by mothers of newborns on neonatal lab screening (NLS), which revealed certain deficiencies in the awareness of mothers on neonatal lab screening. In 2014, a pluriannual prospective study on parental stress caused by false positivity of NLS was also launched.

"Methodological guidelines on the implementation of screening of hearing in newborns" published in the Journal of the Ministry of Health in 7/2012 was drafted for the purposes of implementing neonatal screening of hearing. Selective screening (i.e. the examination of clinically ill persons or persons at risk of a genetic disease) took place as part of the standard curative and preventative care at respective facilities in the Czech Republic. In 2014, the EB Centre launched the diagnostics of all types of EB and thirteen other genetic dermatoses by sequencing methods of the new generation.

4.2 The amendment to Decree No. 98/2012 Sb. on medical records

The amendment to Decree No. 98/2012 Sb. on medical records was a response to certain ambiguities concerning the period permissible to file cards for neonatal lab screening in individual medical facilities. The retention periods varied, lasting 43 or more years from the last examination of the patient. An incoherent interpretation of the retention period caused problems for healthcare providers, with the providers having problems with creating a hierarchy of the retention period of cards under Annex No. 3 to Decree No. 98/2012 Sb. on medical records. Supplementing Annex Nos. 1 and 3 to Decree No. 98/2012 Sb. on medical records to include a card on the implementation of neonatal laboratory screening unified the retention period for this section of medical records in medical facilities. The draft of the amendment shall be submitted for discussion by the government of the Czech Republic.

4.3 Quality and availability of diagnostic laboratories for RD

As far as the analytical quality of diagnostic laboratories is concerned, their accreditation pursuant to ISO 15189 was further developed pursuant to Act No. 373/2011 Sb. and in cooperation with Český institut pro akreditaci (the Czech Institute for Accreditation). Activities related to amending the act on specific medical services have commenced, as practical knowledge and fast development of genomic technologies resulted in the necessity to update certain provisions. Recommendations concerning the indication of genetic examinations were made more specific and the minimum limits for laboratories conducting genetic screenings were updated. The Society of Medical Genetics of CzMA of Jan Evangelista Purkyně and the National Coordination Centre for Rare Diseases under Motol University Hospital cooperated with Všeobecná zdravotní pojišťovna (General Health Insurance Company) and the Association of Health Insurance Companies of the Czech Republic as far as refunds of RD diagnostics and cross-border cooperation are concerned. Professional recommendations have been issued with respect to informed consent before genetic examination and the implementation of cytogenetic examinations for rare congenital developmental defects. Good laboratory practice recommendations for specific RD were updated and the amendment to Decree 134/1998 governing the list of medical services with their scores with respect to genetic lab test was commenced. In terms of accessibility, the network of genetic departments is adequate in the Czech Republic, with minimal regional differences.

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

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

In the period from 2012–2014, centres of highly specialized medical care for RD were established pursuant to Act No. 372/2011 Sb. on medical services and their provision (such as the National Coordination Centre for Patients with Rare Diseases, centres for patients with cystic fibrosis, inherited metabolic diseases, epidermolysis bullosa congenita (EB), centres for pulmonary hypertension, rare congenital and heart defects, etc.). A review of the existing network of functional centres for RD patients was conducted in cooperation with health insurance companies and representatives of professional societies. Calls by the Ministry of Health of the Czech Republic for the establishment of centres of highly specialised care for these patients in individual medical fields were being prepared in cooperation with representatives of professional societies.

In 2014, pursuant to Section 112 of Act No. 372/2011 Sb. on medical services, a Call by the Ministry of Health was published in the Journal of the Ministry of Health of the Czech Republic in 3/2014 to submit applications for the grant of the Status of a centre of highly specialized care for patients (adults and/or children) with rare congenital and acquired haematopoiesis disorders and the Statute of a centre of highly specialized care for patients (adults and/or children) with haemophilia and other homeostasis disorders. The Status of a Centre of highly specialised care for patients with haematological diseases shall be granted for a period of five years. The territory in which highly specialised haematological care is to be provided is the Czech Republic. Centres of highly specialised care for children with haemophilia and other

homeostasis disorders shall ensure complex diagnostic and curative care, including surgical operations and the treatment of inhibitors to coagulation factors.

The centralisation of care for patients with RD and the granting of Status of Centres of highly specialised medical care shall proceed in the upcoming period. The EU Council Recommendation ruled that these centres should have an international connection (such as the European project E-rare2, Horizon2020) and they should be involved in international initiatives or European grant projects in the given field.

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

The application of the latest findings in RD diagnostics and treatment in practice was supported through grants and support was also provided for expanding the range of existing patient registers and establishing new ones, improving screening and diagnostics, and efforts to introduce standards of care for individual rare diseases. An overview of grants and projects for 2013–2014 is given in the annex.

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

In the period from 2012 to 2014, a great number of medicinal products for the treatment of rare diseases reached the reimbursement system, either as highly innovative medicinal products or as permanently reimbursed medicinal products. The reimbursement of orphan medicinal products is usually bound to specialized centres ensuring the effectiveness and economy of the prescription of these drugs, as well as their prescription at the highest professional level. Pharmacotherapy of rare diseases is also influenced by the setting of prices and reimbursements of medicinal products from health insurance. The regulation of price and reimbursements of medicinal products is governed by the act on public health insurance. The applicable legislation does not directly specify the term “rare diseases” or “orphan medicinal products”. The assessment of orphan medicinal products is practically carried out based on the same criteria as the assessment of other medicinal products.

Drugs for RD are distributed to patients by the same mechanism as other medicinal products. On average, ensuring their availability takes two years after approval by the European Commission (registration and setting the price and reimbursement). The decision-making on reimbursements of medicinal products used in out-patient care is within the powers of SÚKL – The State Institute for Drug Control. However, SÚKL does not make decisions about reimbursements of medicinal products used during hospitalization. There are other possibilities to reimburse a medicinal product, apart from the determination of reimbursement by SÚKL in administrative proceedings. Drugs are included in treatment recommendations of individual professional medical societies in compliance with their

registration. This might include, for example, reimbursement based on the agreement of a medical facility with the producer of a specific drug, another possibility is reimbursement pursuant to Section 16 of the Act on Public Health Insurance, which may be applied to products which are not normally covered by health insurance, if their administration is the sole method of treatment. The application of Section 16 of this act is the responsibility of health insurance companies.

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

In the period from 2012–2014, patient organisations were established, fulfilling an important role as far as psychological and social support is concerned. Many patient organizations and activities reached beyond the framework of a mere exchange of experiences and they became true partners for patients and their loved ones to whom they provide counsel from doctors, lawyers, psychologists and social workers. Patient organisations also offer great help to doctors, nurses, professional societies and the healthcare system by raising awareness on prevention and screening possibilities, thus contributing to making an early diagnosis of certain diseases while they are still well curable.

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

Due to the pressure of medical professionals, provisions on the introduction and use of the ICF - International Classification of Functioning, Disability and Health was removed from medical legislation. The reintroduction of a statutory obligation of the use of ICF is being discussed.

6.2. Educating the professional public about the ICF system

Doctors active in the field of labour and social affairs are ready for the introduction of ICF. Most of them have attended respective workshops and have an international certificate.

6.3. Health and social services

With respect to the area of health and social services, it needs 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. An interministerial working group for addressing the issue of cross-sectional social and health services was set up in order to facilitate cooperation between the Ministry

of Health and the Ministry of Labour and Social Affairs aiming 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.

The organisation of summer camps for children and their families, e.g. by the Czech Haemophilic Association, aimed at the training of the intravenous application of coagulation factors for stay-at-home treatment, represents an activity beneficial for ill children. Prophylactic treatment by coagulation factors in haemophilic patients contributes substantially to improving their quality of life. Similar activities have been undertaken for other RD in cooperation with patient organisations, such as cystic fibrosis and muscular dystrophy.

DEBRA ČR also organises one-week recuperative stays with intensive educational content for patients and family members and provides contributions to patients for seaside stays. At the conference supported by DEBRA ČR, patients and their families were provided not only with news in the field of care for EB patients and with updated information on the activities of DEBRA ČR, but also with information and help in the social and legal fields and an offer of psychological counselling.

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

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

In the period from 2012–2014, centres for medical care for RD patients cooperated on other research projects (such as the cardiovascular research program on Invasive approaches to the rescue or regeneration of the cardiac muscle as part of the PRVOUK project). A project to support the research and development of RD - a pilot study by the Institute for Inherited Metabolic Disorders entitled “The Optimization of neonatal screening for inherited metabolic disorders in 2011–2013” was conducted under the auspices of the Internal Grant Agency (IGA) of the Ministry of Health of the Czech Republic.

Grants by IGA of the Ministry of Health of the Czech Republic were also used to fund the Centre for Hereditary Ataxia. Under the (IGA) of the Ministry of Health of the Czech Republic (2013–2015), the Brno University Hospital is undertaking a joint project of the Center of Molecular Biology and Gene Therapy IHOK, the Children’s Dermatology department of the Paediatric Clinic, the Clinical genetics department of the Brno University Hospital and the I. Institute of Pathology and Anatomy of St. Anne’s University Hospital, entitled “The Comprehensive diagnostics of selected genetic dermatoses”.

Participation in the E-rare international European projects has not been ensured. These are an initiative by the European Commission for international research in the field of RD, where national teams are funded from national resources within international consortia. The area of

RD was included in the sectoral commissions of the newly established Agency for Medical Research. No calls relating specifically to RD have been put up to date. In cooperation with the sector for research and development of the Ministry of Health of the Czech Republic, the area of RD was approved as one of research priorities of the Agency for Medical Research.

International EU grants are one of the areas in which participation in research consortia took place (such as the Eurogentest.org, Orpha.net, Techgene.eu, Treat-NMD.eu, Care-NMD.eu, RD-connect.eu, RD-neuromics.eu a Eurenomics.eu 7 - EU framework programme) in the area of RD. With the support of the European Agency for Health and Consumers (now CHAFEA), international registries of RD patients were created with active participation of the Institute for Inherited Metabolic Disorders of the General University Hospital and the First Faculty of Medicine at Charles University.

The implementation of the project entitled "National Coordination Center for Rare Diseases at the Motol University Hospital", supported by the funding mechanism of EEA/Norwegian funds, commenced. The project activities are supposed to raise awareness and knowledge in the field of RD among both the professional community and the general public, and help implement the introduction of new methods of molecular-cytogenetic and genetic diagnostics for the purposes of making prenatal and postnatal RD diagnoses, improve the pharmacoeconomic estimation of diagnostic and therapeutic costs for selected RD, improve complex patient care using experience from Norwegian healthcare services, and develop undergraduate and postgraduate teaching courses. The project shall be completed in 2016.

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 projects

The collection of data on rare diseases was mainly limited by the insufficient details provided in the International Classification of Diseases (ICD-10), 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 of the Czech Republic (hereinafter "IHIS") worked intensively on addressing 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 of these tools and the intensification of international cooperation in their use. A representative of IHIS took part in seminars abroad 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 in the Czech Republic 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 the 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 in 2012–2014 under the auspices of individual professional societies (e.g. the cardiology registers KARDIO-ICD, KATAB, REPACE, REPLY, TAVI, the register of rare diseases of erythron (red blood cells), and the register of the Hereditary Ataxia Centre). A new joint Czech and Slovak register of congenital defects with pulmonary hypertension was established and data collection was carried out in the register of cystic fibrosis.

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

9.1. Support of the activities and development of cooperation with Eurordis, the European association of patient organisations.

RDCZ is an umbrella organisation for patient organisations for RD, comprising 30 patient organisation members and 18 individual members for particular diagnoses. In the period from 2012 - 2014, the members were invited to get involved in RDCZ activities and educational meetings of RDCZ members were organised in an effort to involve individual patient organisations in the implementation of the National Action Plan for Rare Diseases. Furthermore, there were educational activities of representatives of patient organisations in the Academy of Patient Organisations.

DEBRA ČR is a member of DEBRA International, which convenes a yearly congress for patients and experts. The organisations are in touch with one another throughout the year to exchange experiences.

10. Interministerial and interdisciplinary collaboration

In 2010, the Interministerial and Interdisciplinary Working Group on Rare Diseases was established at the Ministry of Health, comprising representatives of the Ministry of Health, the Ministry of Labour and Social Affairs, the Czech VZP health insurance company, the Association of Health Insurance Companies of the Czech Republic, the professional societies of Jan Evangelista Purkyně Czech Medical Association, the WHO Office in the Czech Republic, IHIS, patient organisations and other experts. Between 2012 and 2014, 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 2012 –2014.

Interdisciplinary cooperation was carried out as part of specialised centres of individual medical specialities, where specialised care is provided to patients with RD. The centralisation of healthcare creates conditions not only for interdisciplinary cooperation, but also for

acquiring and maintaining expert knowledge and increasing the efficiency of management, thereby reducing the funds spent on the diagnostics, treatment and further monitoring of a patient while maintaining or even increasing the quality of the care provided.

11. International RD cooperation

11.1 Cooperation at the international level

As far as cardiology is concerned, in the period from 2012 to 2014, individual centers cooperated with their counterparts abroad, the existing patient organisations cooperated with their counterparts in European countries as well as with umbrella European organisations, Czech experts were involved in the creation of European recommended best practices for the diagnosis and treatment of rare diseases in cardiology and in the field of inherited metabolic diseases.

The EB Centre with the Brno University Hospital is a member of "EB Clinet", an international team of clinical experts, and is involved in creating international best practices for the diagnosis and treatment of EB. The children´s dermatological department of the Pediatric clinic of the Brno University Hospital is a member of the international network of clinical experts on rare congenital skin defects - Genodermatoses network, which convenes regular meetings, organizes meetings for young dermatologists, and drafts guidelines focused on RD in dermatology.

In 2013, the National Coordination Center for Rare Diseases organised an international workshop entitled "Rare genetic diseases: diagnosis and discovery workshop" in cooperation with International rare disease research consortium of the European Union and in 2014, it cooperated with the EU section of the MH of the Czech Republic on the preparation of a conference presenting the results of the 2nd EU Action Programme in the field of public Health together with the European agency CHAFEA (formerly EAHC DG Sanco, Luxembourg). Cooperation also took place with the International Rare Disease Research Consortium, where the Czech Republic has a representative in the Diagnostic Committee.

RDCZ cooperated with the European patient organisation for RD - Eurordis.org and it started cooperation with Frambu, a Norwegian patient organisation. The patient organisation DEBRA ČR took part in DEBRA International conferences.

11.2 Standard classification of RD in cooperation with the Orphanet consortium

The International Classification of Diseases ICD-10, which is currently in use, does not enable coding RD precisely and efficiently enough. The Orphacodes system created by the Orphanet onsortium is the leading international system for coding RD. Orphanet cooperates with the

International Health Organisation and the Orpha Codes system will probably be integrated into the 11th revision of the International Classification of Diseases as a special unit.

In the period 2012–2014, the Institute of Health Information and Statistics of the Czech Republic (IHIS) (ÚZIS ČR) took part in international workshops focused on keeping RD registers and, first and foremost, on the coding of RD in registers and information systems. Currently, IHIS (ÚZIS ČR) is working on RD classification and on the use of Orphacodes together with the database entitled Online Mendelian Inheritance in Man (OMIM) and the classification of Society for the Study of Inborn Errors of Metabolism (SSIEM) in a single data point model for RD and as a pilot in the National registry of congenital defects (NRVV). The implementation of the Orphacodes system in the ČR (prospectively including the translation of specialised terminology into Czech and thereby its codification in the Czech version of the system) and its embedding in the minimum data model shall enable RD classification based on international standards. At the same time, the codification of these diseases in the Czech Republic shall be prepared for the transfer to the 11th revision of the International Classification of Diseases.

National Action Plan for Rare Diseases for 2015–2017

In the National Action Plan for Rare Diseases for 2015–2017, emphasis is placed mainly on support for the timely identification and diagnosis of RD, the centralisation of RD patient care, harmonisation and development of data collection, development of standards of care for RD patients, improving awareness and education on RD, continuity of on-going international cooperation and establishing new contacts, as well as the involvement of Czech centers in common European databases and registers that collect clinical data. It will also be important to participate in international research projects on RD (e.g. E-Rare2, Horizon 2020), national 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 establish homecare centers, social beds, respite centres and rehabilitation facilities and to expand long-term care beds with trained personnel and special equipment. Attention needs to be paid to effective and timely rare disease pharmacotherapy building on internationally recognised standards and best practices, as well as to increased education and awareness of 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 annex.

National Action Plan for Rare Diseases for 2015–2017						
Objectives	**Tasks**	**Deadline**	**Outputs**	**Coordinator**	**Cooperation**	**Conditions**
1. Improving RD awareness	1.1. Operating, developing and updating web portals for RD; Establishment of a phone helpline	on-going	RD web portals Helpline 116XXX	Motol NCC	University Hospital Brno, RD Centers, PO, The Ministry of Labour and Social Affairs, health insurance companies, RDCZ, ČTÚ	Subsidy schemes NF
	1.2. RD awareness among the professional community and the general public	on-going	Seminars, conferences, campaigns, cooperation with the media; Research on RD awareness	Motol NCC	Professional societies RD Centers PO, MH Insurance companies	Subsidy schemes NF
2. Education in the field of RD	2.1. The professional community (undergraduate and postgraduate education for doctors, paramedical personnel and non-medical healthcare professionals,	on-going	The extension of pre-graduate and postgraduate education in the field of RD; The creation of the system of postgraduate education in the field of RD;	Motol NCC RD Centers, Professional societies, The Institute for Postgraduate Medical Education (IPVZ) MH	Faculties of Medicine MŠMT – The Ministry of Education, Youth and Sport Accredited facilities	Subsidy schemes NF

	lifelong learning in this field)		Publishing of specialised publications			
3. RD Prevention	3.1. Preconception and prenatal RD screening	on-going	Draft methodology for preconception and prenatal RD screening	Motol NCC, KCNS VFN (the Coordination Centre for Neonatal Screening with the General University Hospital in Prague), Professional societies	MH Insurance companies	Subsidy schemes Health insurance
4. Improvement of RD screening and diagnoses	4.1. The extension of population neonatal screening by early diagnosis of RD (sensory impairments, other inherited metabolic disorders etc.) 4.2. The development of fetal cardiology centres 4.3. Association of expert genet.	2015–2017	Population neonatal screening of hearing, methods for neonatal screening of hearing; A proposal for the optimisation of neonatal screening of metabolic disorders	The Czech Society of Otorhinolaryngology The Czech Society of Cardiology; The Czech Medical Association of Jan Evangelista Purkyně Motol NCC, KCNS VFN (the Coordination Centre for Neonatal Screening with the General University Hospital in Prague), MH	Professional societies, IWG Insurance companies	IGA Subsidy schemes Health insurance

	facilities for RD diagnostics in cardiology 2015–2017					
	4.4. Quality and availability of RD diagnostic laboratories	2015–2017	Information for laboratories preparing for accreditation under ISO 15189 and Act No. 373/2012 Sb. Information on the availability of laboratory test methods for RD at the website of Motol NCC and SLG.cz	MH The Society of Medical Genetics of Czech Medical Association of J. E. Purkyně	Brno University Hospital Professional societies,	Subsidy schemes
5. Improving the availability and quality of care for RD patients	5.1. The centralisation, coordination and the 2015-2074 Analysis of the existing MH Specialised integration of care for patients with RD	2015–2074	Analysis of existing facilities; The granting of the status of highly specialized centres for RD; Development of cross-border care	Insurance companies	Professional societies Motol NCC IWG RD Centers PO	NF Health insurance
	5.2. Best practices in the diagnostics	on-going	Best practices, methods,	Professional societies	Insurance companies	Subsidy schemes NF

	and treatment of selected RD (including nursing procedures)		standards, pilot projects for selected groups of RD		PO	
	5.3. Effective and timely RD pharmacotherapy (orphan drugs) building on internationally recognised standards and recommended best practices	2015–2017	The analysis and proposal of efficient RD pharmacotherapy (an improvement in the availability and efficiency, the monitoring of treatment efficiency)	SÚKL MH Motol NCC	Professional societies, Insurance companies IWG	Subsidy schemes
6. Improving the quality of life and social integration for people with RD	6.1. The introduction of the ICF system (ICF – International Classification of Functioning, Disability and Health)	on-going	Feasibility study for ICF	IHIS (ÚZIS ČR)	MLSA Professional societies Insurance companies, PO	Subsidy schemes
	6.2. Educating the professional public about the ICF system	on-going	Seminars, conferences, educational programmes, methodologies	The Institute for Postgraduate Medical Education (IPVZ) Professional societies, CMC (Czech Medical Chamber)	MH, MLSA, IHIS (ÚZIS ČR), PO	Subsidy schemes

	6.3. Health and social services	on-going	Proposal for a system solution for long-term health and social care for RD, feasibility study	MH MLSA	Insurance companies Regional and Municipal authorities, CMC (The Czech Medical Chamber), The Czech Medical Association of Jan Evangelista Purkyně PO	Health insurance Funding of social services
7. Support of basic and applied research in the field of RD	7.1. Targeted support for science and research in the field of RD - at the national level; 7.2 Developing cooperation between faculty and non-faculty scientific facilities and medical and non-medical faculties	2015–2017	Research, projects, studies	Faculties of Medicine Motol NCC RD Centers Professional societies	Czech Academy of Sciences Faculty of Science MH	Subsidy schemes IGA

	7.3. Targeted support for science and research in the field of RD - at the international level	2015–2017	Research, projects, studies, involvement of the Czech Republic in the E-Rare project, etc.	Motol NCC RD Centers Professional societies	WHO	Subsidy schemes NF 2nd Action Programme of the Association of Public Health FP7
8. Harmonisation and development of RD data collection and biological sampling	8.1. National data collection in the field of RD, improvements in data collection methodologies and support of participation in international RD data collection projects	on-going	Data collection, statistics, analyses, IHIS (ÚZIS) methodology, international cooperation	IHIS (ÚZIS ČR) The Office for Personal Data Protection KCNS VFN (the Coordination Centre for Neonatal Screening with the General University Hospital in Prague)	Brno University Hospital, RD Centers, Professional societies	
	8.2. Legal framework for data collection and biological sampling	2017	Project of data collection pursuant to ICN XI Regulation of RD data collection and biological sampling	IHIS (ÚZIS ČR) MH	Professional societies	
9. Supporting and strengthening the role of RD patient organisations	9.1. Collaboration with patient organisations in the Czech Republic, the	on-going	Projects, seminars, conferences, cooperation with the media	PO SÚKL	IWG Professional societies WHO CZ	Subsidy schemes

	development of cooperation with Eurordis, the European association of patient organisations					
	9.2. Supporting the activities of patient organisations	on-going	Seminars, conferences, cooperation with the media	PO	IWG Professional societies WHO CZ	Subsidy schemes NF 2nd Action Programme of the Association of Public Health The EU
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	on-going	The activity of IWG for RD	MH	IWG	
11. International RD cooperation	11.1. RD cooperation at International level, exchange of experiences, data and information	on-going	International cooperation, projects	Motol NCC	RD Centers Professional societies PO WHO CZ IHIS (ÚZIS ČR)	Subsidy schemes

	11.2. Cooperation within the European Commission's EuroPlan/Eurordis project	2015	International cooperation	Motol NCC MH	PO	Subsidy schemes
	11.3. Standard classification of rare diseases for the planned ICD-11 revision and in cooperation with the Orphanet consortium.	on-going	MKN-11	IHIS (ÚZIS ČR) MH WHO CZ	IWG, Professional societies, Orphanet, PO	Subsidy schemes

Acronyms used:
RD – rare diseases
Motol NCC - the National Coordination Centre for Rare Diseases at the Motol University Hospital
KCNS VFN - the Coordination Centre for Neonatal Screening with the General University Hospital in Prague
E-rare - ERA-Net for Research Programmes on Rare Diseases of the European Commission
MH – The Ministry of Health
MLSA – The Ministry of Labour and Social Affairs
MŠMT – The Ministry of Education, Youth and Sports
IWG – Interdepartmental and interdisciplinary working group for rare diseases
PO – patient organisations
NF – Norwegian Funds
SÚKL – State Institute for Drug Control
IHIS (ÚZIS ČR) – Institute of Health Information and Statistics
WHO CZ – Country Office of the World Health Organisation in the Czech Republic
MU Brno – Masaryk University Brno
EU – The European Union
Eurordis – European association of patient associations for rare diseases
FP7 EU – 7th framework programme of the European Commission
IGA – Internal Grant Agency
CzMA JEP – Czech Medical Association of Jan Evangelista Purkyně
CMC (ČLK) – Czech Medical Chamber
Health insurance companies
CAS – Czech Academy of Sciences
SF - Science Faculties

THE GOVERNMENT OF THE CZECH REPUBLIC

RESOLUTION No. 76

BY THE GOVERNMENT OF THE CZECH REPUBLIC
dated 4 February 2015

regarding the Report on the Fulfilment of Tasks from the National Action Plan for Rare Diseases for 2012–2014 and the National Action Plan for Rare Diseases for 2015–2017

The Government hereby

I. **approves** the Report on the Fulfilment of Tasks from the National Action Plan for Rare Diseases for 2012–2014 and the National Action Plan for Rare Diseases for 2015–2017, attached in Section III of document ref. No. 69/15

II. **imposes** on the Minister of Health the duty to submit to the following to the Government by 31 December 2017

1. The Report on the Fulfilment of Tasks from the National Action Plan for Rare Diseases for 2015–2017;
2. a draft National Action Plan for Rare Diseases for 2018–2020.

To be performed by:
The Minister of Health

Prime Minister
Mgr. Bohuslav Sobotka, by his own hand