‘NATIONAL ACTION PLAN FOR RARE DISEASES 2012–2014’

GOVERNMENT RESOLUTION NO 633 OF 29 AUGUST 2012.

MINISTRY OF HEALTH – 2012, WWW.MZCR.CZ

GOVERNMENT OF THE CZECH REPUBLIC

RESOLUTION

OF THE GOVERNMENT OF THE CZECH REPUBLIC
No 633 of 29 August 2012

on the National Action Plan for Rare Diseases 2012–2014

The Government

I. **approves** the National Action Plan for Rare Diseases 2012–2014, as contained in Part III of Document No 712/12 (the 'National Action Plan');

II. **enjoins**

1. the Minister for Health

a) to coordinate the implementation of activities contained in the National Action Plan,

b) to submit to the Government, by 31 December 2014,

ba) a report on the implementation of the National Action Plan,

bb) the National Action Plan for Rare Diseases 2015–2017,

c) to coordinate the activities of the Inter-ministerial and Interdisciplinary Working Group on Rare Diseases in accordance with the activities contained in the National Action Plan,

2. members of the Government to implement, within the scope of their competence, the activities contained in the National Action Plan;

III. **recommends that** regional governors, in collaboration with local government authorities and nongovernmental organisations, reflect the treatment of the issue of rare diseases in regional development plans.

To be implemented by:

Members of the Government

Cc:

Regional governors,
Mayor of Prague

Prime Minister
Petr Nečas, m.p.

National Action Plan for Rare Diseases 2012–2014

Rare diseases (RDs) are a clinically heterogeneous group of approximately 6 000 to 8 000 different diseases with the common characteristic of very low prevalence in the general population (i.e. fewer than one in 2 000 individuals). Incorrect or late diagnosis of those diseases often leads to irreversible damage to the patient's health or even death. In particular current shortcomings in the field of RDs include insufficient identification by the general practitioners, serious deficiencies and delay in the diagnosis and treatment of such diseases, inadequate quality of the services provided, limited spectrum of medicines for their treatment, shortcomings in national and regional data collection, lack of effective care for patients with RDs, and a lack of awareness of such diseases among medical professionals and the general public. Another important consequence of the chronic and often progressive nature of RDs is that shortcomings often come to light indirectly in the social and legal fields. The Council of the European Union, in its Recommendation of 8 June 2009 on an action in the field of RDs, recommended that EU Member States improve their identification, promote the development of respective health policies, and expand European cooperation, coordination and regulation in this field.

On 14 June 2010, the Czech Government approved (under Government Resolution No 466) the 'National Strategy for Rare Diseases 2010–2020' (the 'National Strategy'), which sums up the issue of RDs from both the EU's and the Czech Republic's point of view, describes the main objectives and lays down measures for improving the situation in the country. The main objectives of the National Strategy are to ensure that all patients with RDs have access to high-quality care, including timely diagnosis and the best method of treatment based equality and solidarity, including improvement in the effectiveness of care for those patients.

The 'National Action Plan for Rare Diseases 2012–2014' (the 'National Action Plan') stipulates priority tasks and activities that will be implemented in the years 2012 to 2014 so that all of the National Strategy's objectives and measures, simplified for greater clarity and transparency, are gradually being fulfilled. Secondary tasks, tools, responsibilities, deadlines, financial resources and indicators of the fulfilment status of individual tasks of the National Action Plan have been set so that they are feasible and have a minimum financial impact on the central government budget and public health insurance funding.

In 2012–2014, the following tasks and activities, summarised in the table below, are proposed in the field of rare diseases:

1. Improved awareness

There is a general lack of awareness about RDs among both the professional and lay public in the Czech Republic. Priority tasks and activities to improve awareness in this field include the operation of a general website for RDs (www.vzacnenemoci.cz) and other related websites, participation in the European Orphanet project (www.orpha.net) and the expansion of its dedicated Czech portal (www.orphanet.cz), cooperation on the development of a national registry of rare disease diagnostic examinations (http://www.registr-raritnich-vysetreni.cz/), including the sub-group of rare birth defects on the website www.vrozene-vady.cz, greater awareness among professionals about the extensive encyclopaedic information and links in Orphanet, which could make a significant contribution to enhanced comprehensive care for patients with RDs. In order to improve awareness, recommendations and informative materials will also be utilised from the web based resources of the European Committee of Rare Diseases Experts (EUCERD.eu), in whose activities the Czech Republic is involved, and from the umbrella organisation of European patient associations (Eurordis.org), which cooperates with the Czech Rare Diseases Association (ČAVO;

www.vzacna-onemocneni.cz) and the Coalition for Health (www.koaliceprozdravi.cz). In order to improve awareness of RDs, numerous seminars, conferences, campaigns and media events will be held.

2. Education

Deficiencies in the diagnosis and treatment of RDs currently belong to one of the main problems to be addressed. Priority tasks and activities in this area therefore include the incorporation of specific RD-related issues into undergraduate and postgraduate education in collaboration with individual Czech medical schools. Postgraduate training should focus primarily on general practitioners. Training will also be provided to other health care workers who provide services to patients with RDs. Planned activities include educational seminars and conferences; relevant educational programmes, including e-learning and standard educational procedures which will be targeted at selected RDs. Domestic and foreign educational materials from e.g. Orphanet, Eurordis, EUROCAT, ICBDSR, EUCERD websites will be used in these educational activities. The experience and suggestions of RD patient organisations will also be solicited and utilised.

3. Prevention

Prevention of RDs should primarily focus on the development of their preconception- and prenatal diagnosis in accordance with the recommendations adopted in the framework of international cooperation (EUCERD.eu, Eurogentest.org, ESHG.org) and in cooperation with, *inter alia,* with various professional associations of ČLS JEP [Czech Medical Association of Jan Evangelista Purkyně], comprising the Czech Society of Medical Genetics, Czech Society for Ultrasound Examinations in Obstetrics and Gynaecology, Czech Paediatric Society, including Society of Practical Paediatricians and all health insurance companies.

4. Improving screening and diagnosis

At present, in accordance with a guideline published by the Czech Ministry of Health (the MoH) Bulletin (Věstník MZ) No 6/2009, a dried blood drop is tested for 13 diseases in two neonatal screening laboratories with nationwide coverage. The effectiveness of the screening programme is comparable to other European countries, reporting a detection rate of approximately 1:1200 newborns. For instance in comparison with the situation in the United States of America, the number of RDs in the Czech screening panel is significantly lower. Therefore, there is an opportunity to expand the number of RD screened within the frame of a research project implemented by the Internal Grant Agency of the MoH entitled ‘Optimising the Neonatal Screening of Inherited Metabolic Disorders 2011–2013’, which will assess which further RDs could be added to the updated screening panel. Importantly, respective change to the MoH guideline will also be proposed. Another type of nationwide neonatal screening is represented by the screening of hearing loss, which has recently been included in the List of Medical Procedures (Sazebník Výkonů). In order to implement neonatal hearing loss screening, the creation of the corresponding guideline needs to be completed and published in the MoH Bulletin. Neonatal wards also need to be equipped with an appropriate technology and their personnel needs to be duly trained. Selective screening (i.e. the examination of patients or persons at high risk of a genetic disease) takes place as part of standard medical and preventive care at different sites throughout the country. With an outlook of promoting selective screening, planned activities will include the creation of an informative website on the availability of diagnostic procedures for each RD. Nevertheless, an important prerequisite for the quality of genetic diagnosis performed on the germ line genome is the accreditation of genetic testing laboratories, including external quality control schemes, which are legally enshrined in Act No 373/2011 Art 28. Genetic laboratory accreditation process has been launched in 2012 and from the perspective of the National Action Plan it is desirable to expand the promotion of awareness among genetic laboratories

about the necessity of laboratory accreditation. At the same time, there is a need to develop a specialised database of accredited laboratories / centres operating in the field of medical genetics which are establishing diagnoses of RDs (www.slg.cz, www.registr-raritnich-vysetreni.cz), and to foster cross-border information exchange of such portals with www.orpha.net, www.eddnal.com and the US Genetests.org.

5. Improving the availability and quality of care

It is necessary to improve the effectiveness of the diagnosis and treatment of RDs, in order to ensure that all such patients have equal access to indicated and high-quality health care, and to introduce organisational measures that will remove barriers in patient access to specialised care. According to the aforementioned Recommendation of the EU Council, improvements in the care of patients with RDs can be improved and the cost of treating complications arising from their late diagnosis can effectively be reduced by centralisation of care for such patients. Thus, RD centres with the necessary inter-disciplinary capacity and sufficient experience in their early diagnosis and therapy will ensure that patients can be rendered high-quality and effective treatment. The Recommendation also states that such centres should have international links and should be involved in international initiatives or European research projects in this field. In the Czech Republic, a basic network of centres for RDs already exists, comprising the National Coordinating Centre for RDs, Centres for the Care of Children and Adults with Cystic Fibrosis, the Centre for Congenital Epidermolysis Bullosa and the Centre for Inherited Metabolic Disorders. Further to an analysis of already existing centres providing care to patients with RDs, a network of other specialised care RDs centres will be established within the frame of the National Action Plan. We will reflect current developments in this area at the EU level where at present, the establishment of European reference networks (ERNs) is being elaborated in accordance with Directive 2011/24/EU on the Application of patients' rights in cross-border healthcare. We will follow criteria and conditions which are currently being developed at the European level. The National Action Plan sets out plans for the development of "virtual" diagnostic algorithms for patients with RDs, both based on the visualisation of expertise (the interlinking of experts at different institutions via e-health) and the creation of a national diagnostic 'telephone help-line' for general practitioners to direct patients with unclear diagnoses to specialised centres. To improve health care for patients with RDs, best practices for their diagnosis and treatment need to be formulated. Within the upcoming two-year period effective and timely pharmacotherapy, using costly medicines for RDs (orphan drugs), in accordance with internationally recognised standards and best practice guidelines in terms of their broader national availability and effectiveness of care, will be longitudinally analysed and fostered .

6. Improving the quality of life and social integration

One of the objectives of the National Strategy for Rare Diseases for the years 2010 to 2020 is to improve the quality of life and social integration of persons with RDs. Planned activities in this field include the introduction of, and awareness among the professional public of, the introduction of the International Classification of Functioning, Disability and Health (ICF), including specific improvements in health and social services. The ICF, like the International Classification of Diseases and Related Health Problems (ICD), is part of the set of classification systems created by the World Health Organisation (WHO). The ICF classification is intended for the evaluation of the degree of disability (i.e. the extent to which certain common activities are restricted or prevented due to a disorder), the assessment of medical fitness to work (if a natural person is disabled), the assessment of special educational needs, and the prescription and reimbursement of medical devices. Moreover, it is aimed at health insurance companies, the identification of health status as a basis for the assessment of benefits in the domain of social security and employment services. It also covers, the assessment of long-term adverse health status in matters of social security and

employment, including introduction of monitoring of the evaluation of health status of patients with RDs in terms of their employment possibilities. The ICF was approved in 2001 at the 54th WHO World Health Assembly and recommended for international use. Act No 372/2011 introduces the ICF into the Czech health care system with effect as of 1 April 2013. Under Section 53(2)(f) of that Act, medical records according to ICF contain the classification of patients whose medical condition indicates that certain physical, mental or social functions are impaired in the long-term and/or are of permanent nature. This act also stipulates conditions for the assessment of patients who are partially or fully disabled, including the quantification / degree of their disability. Therefore, in the forthcoming period, professionals must be properly informed about the ICF system and its application into clinical practice.

Within the frame of health care and social services, the Czech Ministry of Labor and Social Affairs (MoLSA) and MoH will address the issue of long-term health and social care in order to create conditions for the application of the principles of equity in health and social services. A major emphasis is placed on the transfer of this process to the level of local government and the maximum possible application of long-term care services in a natural, e,g, family, setting. Among other things, it will be necessary to map related services for RDs patients in their natural setting, including social services.

7. Support for science and research

The development of basic and applied research in the field of RDs should be supported and this type of research activities should be included among national research priorities. The Czech Republic's participation in international projects and involvement in the exchange of experiences, data and information (e.g. ORPHANET, EURORDIS, ERNDIM, EUROCAT, ICBDSR, E-rare ERA–Net for Research Programmes on Rare Diseases) should also be encouraged.

8. Harmonisation and development of data collection and biological sampling

Current shortcomings in the field of RDs include their inadequate identification and deficiencies in data collection both nationally and regionally. To date, there are no statistics available in the Czech Republic on the exact prevalence of RDs, given that such diseases are not yet appropriately classified in the International Classification of Diseases (ICD). In the process of harmonisation and development of data collection and biological sampling, it is necessary to support the development of national data collection in the field of RDs (in the pre-natal, peri-natal and post-natal periods) as part of the activities of the Institute of Health Information and Statistics (IHIS; UZIS.cz) and the National Register of Reproductive Health which is currently being developed by MoH. The planned activities include a study on the feasibility of national registers for individual RDs in collaboration with the Czech Data Protection Authority (uoou.cz) from the perspective of data security for small datasets, including collaboration with international projects (e.g. Epirare, EUCERD, Orphanet, SSIEM.org, FP7 RDConnect project in the field of international registers, including EuroBiobank.eu).

9. Support and strengthening of the role of patient organisations

The main tasks of the NAP include the involvement of patient organisations in national inter-ministerial and interdisciplinary cooperation in the field of RDs and the strengthening of their role. Such cooperation will focus in particular on working with overarching organisations comprising the Coalition for Health (www.koaliceprozdravi.cz) and the Czech Association for Rare Diseases (www.vzacna-onemocneni.cz). It is necessary to support the activities of patient organisations for example by the development of information websites, training of patients with RDs, the holding of dedicated seminars, fundraising and cooperation with the press in raising awareness of and promoting the issue of RDs.

10. Inter-ministerial and inter-disciplinary collaboration

In 2010, the MoH established an Inter-ministerial and Inter-disciplinary Working Group on Rare Diseases, comprising representatives of the MoH, the MoLSA), the State Institute for Drug Control (sukl.cz), General Health Insurance company (vzp.cz), the Union of Czech Health Insurance Companies, expert societies of the ČLS JEP, the WHO Office in the Czech Republic, the IHIS, patient organisations and other experts. In the planned period, this working group, coordinating tasks and activities under the National Strategy for Rare Diseases 2010–2020 and the National Action Plan 2012–2014, will continue its work.

11. International cooperation

In the field of RDs, international cooperation and the exchange of experience, data, information and best practices are essential. Planned activities will include efforts to involve RDs centres in the ERNs currently under development (see Section 7). International cooperation of RDs patient organisations with EURORDIS.org will be established in collaboration with the Czech Association for Rare Diseases and the Coalition for Health (see Section 9). Cooperation will take place under numerous international programmes and projects (e.g. EUROPLAN, EUROCAT, ICBDSR, EuroGentest, Orphanet, ESHG, ESHRE, ERNDIM, ICBDSR, and the European Commission / NIH project IrDIrC). Planned activities include the preparation of cooperation with partners from the EEC using Norway Grants in the fields of public health, support of RD patient organisations, including RD-related research/diagnostics.

Table: National Action Plan for Rare Diseases 2012–2014						
Targets	**Tasks**	**Deadline/Milestones**	**Outputs/Indicators**	**coordinator**	**cooperation**	**Expectations**
1. Improved information on RDs	1.1. Operation and updating of RD websites	ongoing	RD websites	Motol NCC	Brno MU, RD centres, POs, IWG, HIC,	Grant schemes NG
	1.2. RD awareness among professionals and the general public	ongoing	Seminars, conferences, campaigns, media	Motol NCC	Expert societies RD centres POs, MoH HIC,	Grant schemes NG
2. Education in the field of RDs	2.1. Professionals – undergraduate and postgraduate training of doctors, general medical staff, non-medical health workers, lifelong learning in this field	ongoing	Expert seminars, conferences, accredited training programmes, e-learning, standard procedures/methodologies, recommended curricula for the undergraduate and postgraduate teaching of RDs	Motol NCC, RD centres, Expert societies, Czech Medical Chamber (ČLK), MoH	Faculties of medicine Institute of Postgraduate Medicine (IPVZ) Accredited centres	Grant schemes NG
3. RD prevention	3.1. Pre-conception and pre-natal screening for RDs	ongoing	Draft methodology of preconception and prenatal screening for RDs	Motol NCC, CCNS GTH Expert societies	MoH HIC,	Grant schemes Health insurance
4. Improving RD screening and diagnosis	4.1. Expansion of nationwide NS to include the early diagnosis of RDs (e.g. sensory defects, other inherited metabolic diseases, etc.)	2012–2014	Nationwide NS of hearing, Methodology for the NS of hearing, Proposal for the optimisation of the NS of metabolic disorders, Update of NS methodology	ORL Society of the ČLS JEP, Motol NCC, CCNS GTH MoH	Expert societies IWG, HIC, maternity hospitals	IGA Grant schemes Health insurance

	4.2. Quality and availability of diagnostic laboratories for RDs	2012–2014	Information for laboratories preparing for accreditation according to ISO 15189 and Act No 373/2012. Information on the availability of laboratory diagnostic methods for RDs on the website of the Motol NCC and SLG.cz	MoH Medical Genetics Society of the ČLS JEP	Brno MU Expert societies	Grant schemes
5. Improving the availability and quality of care for patients with RDs	5.1. Centralisation and coordination and integration of care for patients with RDs	2012–2014	Proposal for a national telephone 'help line' Analysis of existing centres Establishment of a basic network of centres for RDs Proposal for a network of highly specialised centres for RDs Development of cross-border care	Motol NCC MoH HIC,	Expert societies IWG, RD centres POs	Grant schemes NG Health insurance
	5.2. Best practices in the diagnosis and treatment of selected RDs (including nursing practices)	ongoing	Best practices, methodologies, standards, pilot projects for selected groups of RDs	ČLS JEP MoH	Expert societies HIC, POs	Grant schemes NG
	5.3. Effective and timely RD pharmacotherapy (orphan drugs) following up on internationally recognised standards and best practices	2012–2014	Analysis and proposal of effective RD pharmacotherapy (in terms of the improved availability and effectiveness of care, monitoring of the efficacy of treatment)	SIDC MoH	Expert societies, HIC, IWG,	Grant schemes

6. Improving the quality of life and social integration of persons with RDs	6.1. Introduction of the ICF system (ICF - International Classification of Functioning, Disability and Health)	gradually	ICF feasibility study	MoH	MoLSA IHIS Expert societies HIC, POs	Grant schemes
	6.2. ICF system training for health professionals	ongoing	Seminars, conferences, training programmes, methodology	Institute of Postgraduate Medicine (IPVZ) Expert societies ČLK	MoH, MoLSA, IHIS, POs	Grant schemes
	6.3. Health and social services	ongoing	Proposal for a systemic solution for long-term RD health and social care, feasibility study	MoH MoLSA	HIC, Regional and municipal authorities, Czech Medical Chamber (ČLK), ČLS JEP, POs	Health insurance Financing of social services
7. Support for basic and applied research in the field of RDs	7.1. Targeted support for science and research in the field of RDs – at national level	2012–2014	Research, projects, studies	Motol NCC RD centres Expert societies	MoH	Grant schemes IGA
	7.2. Targeted support for science and research in the field of RDs – at international level	2012–2014	Research, projects, studies, involvement of the Czech Republic in the E-rare project etc.	Motol NCC RD centres Expert societies	WHO	Grant schemes NG Second Programme of Community Action on Public Health FP7

8. Harmonisation and development of data collection and biological sampling in connection with RDs	7.1. National RD data collection, improvements in the data collection methodology support of participation in international RD data collection projects	ongoing	Data collection, statistics, analyses IHIS methodology, international cooperation	IHIS Office for Personal Data Protection CCNS GTH	Brno MU, RD centres, Expert societies	
	8.2. Legal framework of data collection and biological sampling	2014	Project of data collection pursuant to ICD-11 Adjustment of data collection and biological sampling in the field of RDs	MoH	IHIS Expert societies	
9. Support and strengthening of the role of organisations of patients with rare diseases	9.1. Collaboration with patient organisations in the Czech Republic, the development of cooperation with the Eurordis European alliance of patient organisations.	ongoing	Involvement of POs in the Inter-ministerial and Interdisciplinary Working Group on Rare Diseases RD website Projects Seminars, conferences, media	POs SIDC	IWG, Expert societies WHO CZ	Grant schemes
	9.2. Support for the activities of patient organisations	ongoing	Websites, seminars, conferences, media	POs	Ministries Expert societies WHO CZ	Grant schemes NG Second Programme of Community Action on Public Health EU
10. Inter-ministerial and interdisciplinary collaboration	10.1. Coordination of tasks arising from the National Strategy for the Prevention of Rare Diseases 2010–2020 and the respective action plans.	ongoing	IWG activities	MoH	IWG,	
11. International cooperation in the	11.1. International cooperation in the field of RDs, exchange of experiences, data,	ongoing	International cooperation, projects	Motol NCC	RD centres	Grant schemes

field of RDs	information				Expert societies POs WHO CZ IHIS	
	11.2. Cooperation within the scope of the European Commission's Eurordis project	2012	National Action Plan for RDs	MoH	IWG, Ministries, POs	Grant schemes
	11.3. Standard classification of rare diseases for the planned ICD-11 revision and in cooperation with the Orphanet consortium.	ongoing	ICD-11	IHIS MoH WHO CZ	IWG, Expert societies, Orphanet, POs	Grant schemes

Abbreviations:

RD – rare disease
Motol NCC – National Coordination Centre for Rare Diseases at Motol Faculty Hospital Prague
CCNS GTH – Coordination Centre for Neonatal Screening at the General Faculty Hospital Prague
E-rare – European Commission ERA-Net for Research Programmes on Rare Diseases
MoH – Ministry of Health
MoLSA – Ministry of Labour and Social Affairs
MoEYS - Ministry of Education, Youth and Sports
IWG – Inter-ministerial and Interdisciplinary Working Group on Rare Diseases
PO – patient organisation
NAP – National Action Plan for Rare Diseases 2012–2014
NG – Norway Grants
SIDC – State Institute for Drug Control
IHIS – Institute of Health Information and Statistics
OPDP – Office for Personal Data Protection
WHO CZ – Office of the World Health Organisation in the Czech Republic
Brno MU – Masaryk University, Brno
EU – European Union
Eurordis – European association of patient associations for rare diseases

FP7 EU – Seventh Framework Programme of the European Union
IGA – Internal Grant Agency
ČLS JEP – Jan Evangelista Purkyně Czech Medical Association
ČLK – Czech Medical Chamber
HIC – health insurance companies
Orphanet – European website on rare diseases