

Annex 1:
2nd National Action Plan for Rare Diseases (2015 – 2017)

Goals	Tasks	Deadline	Outputs	Administrator	Cooperation	Assumptions
1. Improving awareness	1.1. Operation, development and updating of web portals for RD; Establishing telephone helplines	ongoing	RD web portals Helpline "116"	NKCVO	MU Brno, RD Centers, PO MPS, HIC ČAVO Czech Telecommunications Authority	MH Appropriations NG
	1.2. Awareness of the medical professionals and general public on RD	ongoing	Seminars, conferences, campaigns, cooperation with media; research into RD awareness	NKCVO	Professional societies RD Centers PO, MH HIC	MH Appropriations NG
2. Education	2.1. The medical professionals (pre- and post-graduate education of doctors, nurses, other healthcare workers, life-long / continuing medical education in the field of RD)	ongoing	Expanded pre- and post-graduate education in the area of RD; Creation of a system of post-graduate education in the area of RD at all CZ medical faculties; Editions of expert publications / monographs / textbooks	NKCVO RD Centers, Professional organizations, IPVZ MH	Medical faculties MŠMT Accredited facilities for medical and non-medical training	MH Appropriations NG
3. Prevention	3.1. Preconception and prenatal RD screening, DNA diagnostics	ongoing	Proposed methodology for preconception and prenatal RD screening, DNA testing	NKCVO, KCNS VFN, Professional organizations	MH HIC	MH Appropriations Health insurance
4. Improved screening and genetic diagnosis	4.1. Expanded nationwide neonatal screening to include early RD diagnosis (sensory defects, additional hereditary metabolism disorders, etc.)	2015-2017	Nationwide neonatal screening of hearing loss, Improved laboratory methodology for neonatal screening; implementation of	Professional organizations ČLS JEP, NKCV, KCNS,	MPS HIC	IGA MH Appropriations Health insurance NG

	4.2. Development of fetal cardiology centers 4.3. Associations of expert genetic facilities for diagnosis of RD in cardiology		next generation sequencing Proposal for optimization of neonatal screening for metabolic disorders, congenital heart defects	MH		
	4.4. Quality and accessibility of diagnostic laboratories for RD	2015-2017	Information for laboratories preparing for ISO 15189 accreditation and update of Law No. 373/2012 Coll. Information on the accessibility of genetic testing methods for RD at the NKCVO web portal and SLG.cz	MH Society for Medical Genetics of the Czech Medical Association of J.E. Purkyně	MU Brno Professional organizations	MH Appropriations
5. Improving access to- and quality of care	5.1. Centralization, coordination and integration of care for patients with RD	2015-2017	Analysis of existing RD centers; <i>de iure</i> granting of the status of Centers of Expertise for other RD; Development of cross-border care (diagnostics, treatment)	MH HIC	Professional organizations NKCVO MPS RD Centers PO	NG Health insurance
	5.2. Recommended approaches for the diagnosis and treatment of selected RD (including approach to nursing-related issues), standards of care	ongoing	Recommended approaches, methodologies, standards of care, pilot projects on selected groups of RD	Professional organizations	HIC PO	MH Appropriations NG
	5.3. Effective and timely pharmacotherapy for RD (using orphan medicinal products) following internationally accepted standards and development of professional recommendations	2015-2017	Analysis and proposal of effective pharmacotherapy for RD (improving accessibility and efficacy of care, monitoring efficacy of treatment)	SÚKL MH NKCVO	Professional organizations, HIC MPS	MH Appropriations
6 Improving quality	6.1. Introduction of the ICF system	ongoing	Study of the feasibility of its		MPSV	

of life and social inclusion	(ICF - International Classification of Functioning, Disability and Health)		implementation in the CZ health care system	ÚZIS	Professional organizations HIC, PO	MH Appropriations
	6.2. Educating of medical professionals on the ICF system	ongoing	Seminars, conference, education programs, methodology	IPVZ Professional organizations ČLK	MH, MPSV, ÚZIS, PO	MH Appropriations
	6.3. Social services	ongoing	Proposal for a systematic solution for long-term health-social care for RD, feasibility of its implementation	MH MPSV	HIC, Regional and municipal authorities, ČLK, ČLS JEP, PO, MPSV	Health insurance Financing social services
7. Support for basic and translational research	7.1. Targeted support for science and research in the area of RD - on the national level;	2015-2017	Research, projects, studies	Medical faculties NKCVO RD Centers Professional organizations	CZ Academy of Sciences PřF MH Charles University SMEs	MH Appropriations IGA
	7.2 Development of cooperation between faculty hospital, extra-faculty hospital scientific facilities, medical and non-medical facilities, SMEs					
	7.3. Targeted support for science and research in the area of RD - on the international level	2015-2017	Research, projects, studies connecting the Czech Republic to the ERA-Net E-rare2 project, etc.	NKCVO RD Centers Professional organizations	WHO	MH Appropriations NG 3 rd EU Public Health Program; Horizon 2020; E-rare2
8. Standardization and development of data and biological samples collection	8.1. National collection of data in the area of RD, improving methodology for collection of this data, support for participation in international projects	ongoing	Data collection, statistics, analyses, Institute of Health Information and Statistics, international cooperation with	ÚZIS CZ Office for Personal Data Protection	Masaryk Univ. Brno, RD Centers, Professional organizations	

	for data collection in the area of RD		Orphanet	KCNS,		
	8.2. Legal framework for the collection of data and biological samples	2015-2017	Project for data collection according to ICN-11 Collection of data and biological samples in RD	ÚZIS MH	Professional organizations ÚZIS	
9. Support and strengthening the role of patient organizations	9.1. Cooperation with and coordination of CZ patient organizations, further cooperation with Eurordis and other international patient organisations	ongoing	Projects, seminars, conferences, cooperation with media	PO SÚKL	MPS Professional organizations WHO CZ	MH Appropriations
	9.2. Support for patient organizations' activities	ongoing	Seminars, conferences, cooperation with media	PO	Professional organizations WHO CZ	MH Appropriations NG 3rd EU Public Health Program
10. Inter-ministerial and inter-departmental cooperation in CZ	10.1. Coordination of the fulfillment of tasks stemming from the National Strategy 2010-2020 and corresponding National action plans.	ongoing	Activity of MPS RD (Taskforce)	MH	MPS (Taskforce)	
11. International cooperation	11.1. Cooperation on an international level in the area of RD, exchanging of experience, data, information gathering	ongoing	International cooperation, projects	NKCVO	RD Centers Professional organizations PO WHO CZ ÚZIS	MH Appropriations
		2015-2017	International Cooperation	NKCVO	PO	MH Appropriations

	11.2. Cooperation within the European Commission EuroPlan/Eurordis/E-rare2 projects/CEGRD			MH		NG
	11.3. Classification of RD for the planned update of ICN-11 in cooperation with Orphanet	ongoing	ICD-11	ÚZIS MH WHO CZ	MPS, Professional organizations, Orphanet, PO	MH Appropriations

Abbreviations used:

RD – rare diseases

NKCVO - National coordinating center for rare diseases at the Motol University Hospital

KCNS -Coordinating center for neonatal screening at the General University Hospital, Prague

E-rare - ERA-Net for Research Programmes on Rare Diseases of the European Commission

MH – Ministry of Health

MPSV – Ministry of Labor and Social Affairs

MŠMT – Ministry of Education, Youth and Sports

MPS - Interministerial and interdepartmental taskforce for RD

PO - Patient Organizations

NG – Norway Grants

SÚKL – State Institute for Drug Control

ÚZIS – Institute of Health Information and Statistics

WHO CZ - Office of the World Health Organization in the Czech Republic

MU Brno – Masaryk University Brno

EU – European Union

Eurordis - European association of patient organizations for rare diseases

HORIZON 2020- current framework program of the European Commission

IGA - Internal Grant Agency

ČLS JEP – Czech Medical Association of J.E. Purkyně

ČLK – Czech Medical Chamber

HIC – health insurance companies

AVČR – Academy of Science of the Czech Republic
PřF – Natural Sciences Faculty of Charles University Prague
CEGRD – Commission Expert Group on Rare Diseases (former EUCERD.eu)

