

**2012 REPORT ON THE STATE OF THE ART  
OF RARE DISEASE ACTIVITIES IN EUROPE  
OF THE  
EUROPEAN UNION COMMITTEE OF EXPERTS  
ON RARE DISEASES**



**STATE OF THE ART OF RARE DISEASE ACTIVITIES IN  
CZECH REPUBLIC**

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More information on the European Union Committee of Experts on Rare Diseases can be found at [www.eucerd.eu](http://www.eucerd.eu).

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## ACRONYMS

### General

CAT - Committee for Advanced Therapies at EMA  
CHMP - Committee for Medicinal Products for Human Use at EMA  
COMP - Committee on Orphan Medicinal Products at EMA  
DG - Directorate General  
DG Enterprise - European Commission Directorate General Enterprise and Industry  
DG Research - European Commission Directorate General Research  
DG Sanco - European Commission Directorate General Health and Consumers  
EC - European Commission  
ECRD - European Conference on Rare Diseases  
EEA - European Economic Area (Iceland, Switzerland, Norway)  
EMA - European Medicines Agency  
ERN - European reference network  
EU - European Union  
EUCERD - European Union Committee of Experts on Rare Diseases  
EUROCAT - European surveillance of congenital anomalies  
EUROPLAN - European Project for Rare Diseases National Plans Development  
EURORDIS - European Organisation for Rare Diseases  
FDA - US Food and Drug Administration  
HLG - High Level Group for Health Services and Medical Care  
HTA - Health Technology Assessment  
IRDiRC – International Rare Diseases Research Consortium  
JA - Joint Action  
MA - Market Authorisation  
MoH - Ministry of Health  
MS - Member State  
NBS - New born screening  
NCA - National Competent Authorities  
NHS - National Health System  
PDCO - Paediatric Committee at EMA  
RDTF - EC Rare Disease Task Force  
WG - Working Group  
WHO - World Health Organization

### Pilot European Reference Networks

Dyscerne - European network of centres of expertise for dysmorphology  
ECORN-CF - European centres of reference network for cystic fibrosis  
Paediatric Hodgkin Lymphoma Network - Europe-wide organisation of quality controlled treatment  
NEUROPED - European network of reference for rare paediatric neurological diseases  
EUROHISTIONET - A reference network for Langerhans cell histiocytosis and associated syndrome in EU)  
TAG - Together Against Genodermatoses – improving healthcare and social support for patients and families affected by severe genodermatoses  
PAAIR - Patients' Association and Alpha-1 International Registry Network  
EPNET - European Porphyrin Network - providing better healthcare for patients and their families  
EN-RBD -European Network of Rare Bleeding Disorders  
CARE-NMD -Dissemination and Implementation of the Standards of Care for Duchenne Muscular Dystrophy in Europe project  
ENERCA - European network for rare and congenital anaemia – Stage 3

# GENERAL INTRODUCTION TO THE REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE OF THE EUROPEAN UNION COMMITTEE OF EXPERTS ON RARE DISEASES

The 2012 Report on the State of the Art of Rare Disease Activities in Europe was produced by the Scientific Secretariat of the European Union Committee of Experts on Rare Diseases (EUCERD), through the EUCERD Joint Action: Working for Rare Diseases (N° 2011 22 01), which covers a three year period (March 2012 – February 2015).

The report aims to provide an informative and descriptive overview of rare disease activities at European Union (EU) and Member State (MS) level in the field of rare diseases and orphan medicinal products up to the end of 2011. A range of stakeholders in each Member State/country have been consulted during the elaboration of the report, which has been validated as an accurate representation of activities at national level, to the best of their knowledge, by the Member State/country representatives of the European Union Committee of Experts on Rare Diseases. The reader, however, should bear in mind that the information provided is not exhaustive and is not an official position of either the European Commission, its Agencies or national health authorities.

The report is split into five parts:

Part I: Overview of rare disease activities in Europe

Part II: Key developments in the field of rare diseases in 2011

Part III: European Commission activities in the field of rare diseases

Part IV: European Medicines Agency activities and other European activities in the field of rare diseases

Part V: Activities in EU Member States and other European countries in the field of rare diseases

Each part contains a description of the methodology, sources and validation process of the entire report, and concludes with a selected bibliography and list of persons having contributed to the report.

The present document contains the information from Parts II and V of the report concerning Czech Republic. A list of contributors to the report and selected sources are in annex of this document. For more information about the elaboration and validation procedure for the report, please refer to the general introduction of the main report<sup>1</sup>.

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<sup>1</sup> <http://www.eucerd.eu/upload/file/Reports/2012ReportStateofArtRDActivities.pdf>

# RARE DISEASE ACTIVITIES IN CZECH REPUBLIC

## Definition of a rare disease

Stakeholders in the Czech Republic accept the definition outlined in the European Regulation on Orphan Medicinal Products of a prevalence of no more than 5 in 10'000 individuals.

## National plan/strategy for rare diseases and related actions

In October 2010, the Czech Republic released for the first time a ten-year strategy (2010-2020) for rare diseases. The strategy was approved by the government on 14 June 2010. The Czech strategy intends to “ensure the effective diagnosis and treatment of rare diseases, ensure that all patients with rare diseases have access to the indicated, high-quality health care, and ensure their subsequent social integration on the basis of equal treatment and solidarity”, and is “fully compliant with the European Council's recommendation mainly concerning improved identification of rare diseases, support for the development of health policy and the development of European-level cooperation, coordination and regulation in this field”. The Strategy outlines existing efforts and proposes major targets and measures for improving the situation in the Czech Republic, which are to be subsequently specified in more detail in the context of a three-year National Action Plan that will establish “sub-tasks, instruments, responsibilities, dates and indicators for fulfilling individual tasks”. The first meeting of the working party for the preparation of the National Action Plan convened on 12 November 2010 in Prague and since then a dedicated taskforce (“Meziresortní a mezioborová komise pro vzácná onemocnění – Interministerial and interdisciplinary commission for rare diseases”, henceforward “Taskforce”), under scientific coordination of Prof. Milan Macek (Czech National Orphanet Coordinator and Representative of the Czech Republic on the EUCERD) comprised of leading rare diseases experts, biotech industry, lawyers, the State Institute for Drug Control, medical statisticians and health insurance representatives, has convened every other month. This Taskforce has created dedicated working parties with the aim to establish the basis for the National Action Plan by 2013. A group of stakeholders was gathered in 2011 (including the Ministry of Labour and Social Affairs, experts and payers) and a deadline for elaborating the plan has been established for June 2012.

The Czech ten-year strategy<sup>2</sup> reveals the budgetary sources for the plan, which will include “existing budgetary chapters and domestic and foreign subsidies” such as the Ministry of Health and the country's public health insurance. A budget for the strategy has not yet been announced and is in the process of substantiation by the Taskforce. The Ministry of Health has been trying to assure funding within the frame of the EEA Norway Grants scheme (2008-2014) for the National Coordination Centre at University Hospital Motol<sup>3</sup> and via annual Ministry of Health targeted appropriation schemes, both which have deadlines in June 2012.

Care for rare diseases is to be concentrated in 10 to 20 centres. The establishment of a National Coordination Centre for rare diseases in the Prague-Motol Teaching Hospital of Charles University Prague<sup>4</sup> in collaboration with the University Hospital of Masaryk University Brno<sup>5</sup> for the sake of regional representation) coincides with the approval of the strategy and creation of the Taskforce. Besides diagnostics and treatment, the strategy will encompass research, public information, training for health professionals (both paediatric and adult specialists), and quality of life for patients in collaboration with the Ministry of Social Affairs. In terms of the establishment of centres for rare diseases, there is an agreement to have one coordinating centre for rare diseases and one in the Moravia region, each accounting approximately for half of the entire Czech population of 10 million inhabitants.

## Centres of expertise

There are specialised centres for rare diseases, two of which are the national centre for the diagnosis and treatment of Gaucher disease, and for cystic fibrosis. The value of these hubs has been acknowledged by many of the country's major stakeholders including the State Institute for Drug Control, the Czech general insurance company, the Ministry of Health, patient groups, researchers and physicians. Treatment with orphan medicinal products is reimbursed in these centres and these centres manage the provision of very expensive orphan medicinal products. The organisation of additional specialised centres will be a part of the Czech strategy.

<sup>2</sup> [http://ec.europa.eu/health/rare\\_diseases/docs/national\\_czech\\_en.pdf](http://ec.europa.eu/health/rare_diseases/docs/national_czech_en.pdf)

<sup>3</sup> [http://www.mfcr.cz/cps/rde/xchg/mfcr/xsl/Financial\\_Mechanism\\_EEA.html](http://www.mfcr.cz/cps/rde/xchg/mfcr/xsl/Financial_Mechanism_EEA.html)

<sup>4</sup> [www.ublg.lf2.cuni.cz](http://www.ublg.lf2.cuni.cz)

<sup>5</sup> [www.fnbrno.cz](http://www.fnbrno.cz)

Another important and internationally recognised institution is the Institute for Inherited Metabolic Disorders which deals centrally with these diseases at national level. Another important centre for epidermolysis bullosa has been formed as a Czech branch of Debra International at the University Hospital Brno. However, all these centres mentioned above operate based on their professional reputation and could be thus considered as *de facto* centres.

The Czech National Strategy for Rare Diseases foresees the concentration of care for rare diseases in 10 to 20 different centres, with a National Coordination Centre at the Motol Teaching Hospital in Prague, which will coordinate at the regional level with University Hospital Brno. This centre will assure its funding through the Norway Grants scheme by June 2012. Its major aim, beyond structuring the rare diseases field in the country, would be to identify additional *de facto* centres of expertise and propose their transformation into *de iure* centres by the Taskforce. First steps were taken and at their last meeting (November 2011) the Taskforce decided to officially establish three pilot *de iure* rare diseases centres for a) cystic fibrosis<sup>6</sup>, b) metabolic diseases<sup>7</sup> and c) epidermolysis bullosa<sup>8</sup>, based on a compiled criteria drawn from the published EUCERD and EURORDIS centres of expertise recommendations.

Establishment of centres gives them government recognition, but still does not imply a dedicated budget line. Treatment and diagnostics will thus continue to be provided within standard health insurance procedures administered by a group of health insurance companies. However, in the future and following gradual reform of health care funding (after 2012) it is planned that respective rare diseases or related rare disease diagnostic groups will be concentrated into dedicated centres. In this regard it is expected that the major condition for health care reimbursement would be centre-based care. In addition, it is expected that in duly justified instances (e.g. very rare diseases) care will be assured within European Reference Networks (i.e. in line with the EU Directive on cross-border healthcare sections 54 and 55). Currently, cross-border healthcare falls into the domain of the Centre for International Reimbursements<sup>9</sup>.

#### **Pilot European Reference Networks**

Teams in the Czech Republic participate, or have participated, in the following European Reference Networks for rare diseases: Dyscerne, ECORN CF, EPNET/EPI, ENERCA Paediatric Hodgkin Lymphoma Network, NEUROPED, PAAIR and Care-NMD.

#### **Registries**

The Czech Republic contributes to some European registries such as ECFS.eu for cystic fibrosis<sup>10</sup> and TREAT-NMD for muscular dystrophies, European Porphyria Network (EPNET), EUROCAT for rare birth defects<sup>11</sup>, as well as the SCNIR international registry. The National Registry for Cancer<sup>12</sup> also contains information on the distribution of rare cancers in the Czech Republic. The National Action Plan Taskforce is now in the process of establishing the National registry of rare diseases, which will serve as a “confederated database” of all other disseminated registries in centres to be established.

#### **Neonatal screening policy**

Neonatal screening is now available for 13 disorders<sup>13</sup>. Neonatal screening is routinely performed for phenylketonuria, congenital adrenal hyperplasia, congenital hypothyroidism, hyperphenylalaninemia, maple syrup urine disease, isovaleric aciduria, glutaric aciduria type I, medium-chain acyl-CoA dehydrogenase deficiency, long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency, carnitine palmitoyl transferase 1 deficiency, carnitine palmitoyl transferase 2 deficiency, carnitine acylcarnitine translocase deficiency and cystic fibrosis. There are follow-up clinical services available for all screened disorders and an *ad hoc* working group comprising representatives of the Czech Ministry of Health and screening institutes meets at a tri-monthly basis. Neonatal screening is fully reimbursed by the General Insurance Company<sup>14</sup> from 2010. In early 2011 the

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<sup>6</sup> [www.cfklub.cz](http://www.cfklub.cz), [www.cfregistr.cz](http://www.cfregistr.cz), [ublg.lf2.cuni.cz](http://ublg.lf2.cuni.cz)

<sup>7</sup> [www.udmp.cz](http://www.udmp.cz)

<sup>8</sup> [www.debra.cz](http://www.debra.cz)

<sup>9</sup> <http://www.cmu.cz/en?Itemid=224>

<sup>10</sup> [www.cfregistr.cz](http://www.cfregistr.cz)

<sup>11</sup> [www.vrozene-vady.cz](http://www.vrozene-vady.cz)

<sup>12</sup> <http://www.linkos.cz/odbornici/onkologie/nor.php>

<sup>13</sup> <http://www.novorozenecky-screening.cz/>

<sup>14</sup> [www.vzp.cz](http://www.vzp.cz)

National coordination centre for neonatal screening was officially established by the Czech Ministry of Health<sup>15</sup> and its operation is funded by targeted annual appropriation schemes.

### **Genetic testing**

In terms of diagnostic services, there are over 62 molecular genetic laboratories<sup>16</sup> in the country offering DNA testing and 30 cytogenetic laboratories<sup>17</sup>. Together, they offer diagnostic tests for more than 518 different rare diseases<sup>18</sup>. Genetic counselling exists for all families at risk and 45 such facilities are currently registered<sup>19</sup>, which mostly collaborate with molecular genetic and cytogenetic laboratories. Clinical genetics services are available throughout the entire country, with every major district having such services, both at private and/or state based levels. Genetic services are carried out in compliance with all international professional standards and are fully covered by the national health insurance system. In November 2011 a new law On Specific Health Care Services 373/2011 Sb was passed. It stipulates (Part 6; section 28) that germinal genome is allowed to be tested within the context of rare diseases in genetic laboratories that are accredited according to the ISO 15189 norm, in accordance with "OECD guidelines for quality assurance in molecular genetic testing"<sup>20</sup>. Moreover, new law 372/2011 Sb stipulates provisions regarding informed consent procedures in the domain of health care services.

### **National alliances of patient organisations and patient representation**

In December 2011 foundations were established for the creation of an overarching Czech National Alliance for Rare Diseases<sup>21</sup>. This alliance will link together other patient support groups via a democratic, bottom-up, activity spearheaded by several larger patient support groups. Its registration with the Ministry of Interior is expected to be completed by May 2012, including establishment of its bylaws and governance structure. This association plans to join EURORDIS and participate in its activities. Creating an alliance for rare disease patient groups is a provision of the national strategy being developed, together with the Coalition for Health Association<sup>22</sup>. The Coalition for Health Association includes all diseases, while the Czech National Alliance for rare diseases will collaborate on topics of common interest. However, there are 43 patient organisations in the Czech Republic. Some groups benefit from aid from the Ministries of Health and of Labour and Social Affairs; the system will be streamlined under the National Action Plan, since representatives of the Coalition for Health are members of its Taskforce. Following official registration of the Czech National Alliance for Rare Diseases its representatives will be included in the Taskforce.

### **Sources of information on rare diseases and national help lines**

#### ***Orphanet activities in the Czech Republic***

Since 2006 there is a dedicated Orphanet team in the Czech Republic, currently hosted by the University Hospital Motol and the Second Faculty of Medicine of Charles University Prague. The team was designated as the Czech national Orphanet team by the Ministry of Health in 2010. This team is in charge of collecting data on rare disease related services (specialised clinics, medical laboratories, ongoing research, registries, clinical trials and patient organisations) in their country for entry into the Orphanet database. The team also maintains the Orphanet Czech Republic national website<sup>23</sup> in the Czech language providing an entry point to the database.

#### ***Official information centre for rare diseases***

There is no official information centre for rare diseases in the Czech Republic other than Orphanet: however its creation is envisaged by the National Action Plan Taskforce, together with a dedicated website<sup>24</sup>.

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<sup>15</sup> [www.novorozenecky-screening.cz](http://www.novorozenecky-screening.cz)

<sup>16</sup> <http://www.slg.cz/pracoviste/molekularni-genetika/>

<sup>17</sup> <http://www.slg.cz/pracoviste/cytogenetika/>

<sup>18</sup> This information is provided by <http://www.slg.cz/pracoviste/vysetreni/>

<sup>19</sup> <http://www.slg.cz/pracoviste/klinicka-genetika/>

<sup>20</sup> <http://www.oecd.org/dataoecd/43/6/38839788.pdf>

<sup>21</sup> [www.vzacna-onemocneni.cz/](http://www.vzacna-onemocneni.cz/)

<sup>22</sup> [www.koaliceprozdravi.cz](http://www.koaliceprozdravi.cz)

<sup>23</sup> <http://www.orphanet.cz/national/CZ-CS/index/%C3%BAvod/>

<sup>24</sup> The site [www.vzacnenemoci.cz](http://www.vzacnenemoci.cz) is under construction.

### **Help line**

A help line for rare diseases is under preparation and is planned to be funded within the Norway Grants mechanism with the National Coordination Centre for Rare Diseases in University Hospital Motol.

### **Other sources of information**

Patient organisation web sites are one of the few national sources of information for rare diseases in the Czech language. It is expected that the National Alliance for Rare Diseases will foster their links and best practice. A web based information service on neonatal screening is available<sup>25</sup>.

### **Good practice guidelines**

Best practice guidelines for genetic diagnosis are listed at the National Reference Laboratory for DNA diagnostics at the Institute of Haematology and Blood transfusion for the more common rare diseases<sup>26</sup> and reflect EMQN, CMGS and Eurogentest.org guidelines.

### **Training and education initiatives**

No specific activity reported.

### **National rare disease events in 2011**

Amongst the rare disease events hosted by the Czech Republic in 2011 were the Metabolic Days<sup>27</sup> (Mikulov, 11-13 May 2011), Czech National Genetics Meeting<sup>28</sup> (Třeboň, 7-9 September 2011) and the Czech National DNA diagnostics conference<sup>29</sup> (Prague, 24-25 December 2011).

### **Hosted rare disease events in 2011**

No rare disease events hosted by the Czech Republic in 2011 were reported.

### **Research activities and E-Rare partnership**

#### **National research activities**

Rare diseases research is conducted under several funding bodies: the internal grant agency of the Czech Ministry of Health ([www.mzcr.cz](http://www.mzcr.cz)), the grant agency of the Czech Republic ([www.gacr.cz](http://www.gacr.cz)), and the grant agency of the Charles University Prague ([www.gauk.cz](http://www.gauk.cz)). Currently around 15 different research projects in the field of rare diseases are registered with Orphanet, focusing on around 30 different rare disorders. At least three projects are targeting specific genes. The Czech Republic also participates in many international-level activities including ERNDIM (a consortium for quality assessment in biochemical genetics for rare disease).

#### **Participation in European research projects**

Teams in the Czech Republic participate, or have participated, in the European rare disease research projects, including: CLINIGENE, ENCE PLAN, EUMITOCOMBAT, EURO-PADNET, EUROCARE-CF, EUROPEAN LEUKEMIA NET, EUROAGENTEST, EUROGLYCANET, HUE-MAN, MYORES, NEUROSIS, PNSEURONET, RD PLATFORM, SARS/FLU VACCINE, SCRIN-SILICO and SIOPEN-R-NET. Teams in the Czech Republic contribute to the following European registries: EUROCARE CF, EUROCAT and TREAT-NMD.

#### **E-Rare**

The Czech Republic is not currently a partner of the E-Rare research programme on rare diseases.

#### **IRDiRC**

Czech funding agencies are not yet committed members of the IRDiRC.

### **Orphan medicinal products**

SUKL<sup>30</sup>, the State Institute for Drug Control, is the regulatory body in the Czech Republic responsible for the regulation and surveillance of human medicinal products and medical devices, including orphan medicinal products.

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<sup>25</sup> <http://novorozenecky-screening.cz>

<sup>26</sup> <http://www.uhkt.cz/nrl/nrl-dna/blp>

<sup>27</sup> [www.dedicnemetabolickeporuchy.cz/](http://www.dedicnemetabolickeporuchy.cz/)

<sup>28</sup> [www.trebon2011.slg.cz/](http://www.trebon2011.slg.cz/)

<sup>29</sup> [www.dnakonference.cz](http://www.dnakonference.cz)

### **Orphan medicinal product committee**

There is no permanent committee for orphan medicinal products in the Czech Republic.

### **Orphan medicinal product incentives<sup>31</sup>**

According to the *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products*, the Czech Republic has a number of mechanisms in place to encourage orphan medicinal product development. For example *“administrative fees are not charged for applications for the registration of medicinal products or for an amendment, extension or transfer of registration of a medicinal product or for authorisations for parallel import of a medicinal product, if the application concerns a medicinal product included in the register of orphan medicinal products under Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products.*

*“Under §65(2)(b) of Act No 79/1997 Coll. On medicines, amending and supplementing other acts, as amended, the State Institute for Drug Control may refrain from recovering costs where these concern operations which are in the public interest or may have especially important implications for the wider population. These operations include applications for: authorisation/registration of clinical assessments of medicinal products and notification to the submitter of additions to the records in cases concerning the evaluation of an orphan medicinal product, and consultation and opinions on such applications; application for registration of an orphan medicinal product and application for amendment, extension or transfer of registration or application for authorisations for parallel import of an orphan medicinal product and consultation and opinions on applications concerning orphan medicinal products.*

*“Under §26d(1) of Act No 79/1997 Coll. On medicines, amending and supplementing other acts, as amended, the State Institute for Drug Control may, in the case of orphan medicinal products in justified cases meeting the conditions laid down by decree, allow the registration of a medicinal product or the placing on the market of individual batches of a medicinal product even where the data are indicated on the packaging in a language other than Czech.<sup>32”</sup>*

### **Orphan medicinal product pricing policy**

No specific information reported.

### **Orphan medicinal product market availability situation**

In January 2012, 62 orphan medicinal products were registered in the Czech Republic, of which 45 have been launched on the market (Afinitor, Aldurazyme, Arzerra, Busilvex, Cystadane, Duodopa, Elaprase, Evoltra, Exjade, Fabrazyme, Firazyf, Firdapse, Gliolan, Glivec, Ilaris, Increlex, Inovelon, Kuvan, Litak, Lysodren, Mepact, Mozobil, Myozyme, Naglazyme, Nexavar, Nplate, Pedea, Peyona, Prialt, Replagal, Revlimid, Revolade, Somavert, Tasigna, Tepadina, Torisel, Tracler, Trisenox, Ventavis, Vidaza, Volibris, Wilzin, Yondelis, Zavesca).

### **Orphan medicinal product reimbursement policy**

Not all orphan medicinal products are reimbursed; the 45 which are distributed on a centre basis are fully reimbursed (Afinitor, Aldurazyme, Arzerra, Busilvex, Cystadane, Duodopa, Elaprase, Evoltra, Exjade, Fabrazyme, Firazyf, Firdapse, Gliolan, Glivec, Ilaris, Increlex, Inovelon, Kuvan, Litak, Lysodren, Mepact, Mozobil, Myozyme, Naglazyme, Nexavar, Nplate, Pedea, Peyona, Prialt, Replagal, Revlimid, Revolade, Somavert, Tasigna, Tepadina, Torisel, Tracler, Trisenox, Ventavis, Vidaza, Volibris, Wilzin, Yondelis, Zavesca). In some cases the level of reimbursement is according to individual negotiation between centres and marketing holders.

### **Other initiatives to improve access to orphan medicinal products**

The country has compassionate use programme for specific orphan medicinal products, and therapeutic programmes that allow for the use of certain non-authorised medicinal products, usually coordinated by specific centres, on a named-patient basis. Ad hoc committees exist for very expensive orphan medicinal products, which are centre-based.

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<sup>30</sup> [www.sukl.cz](http://www.sukl.cz)

<sup>31</sup> This section is written with information from the *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products. Revision 2005* (pp9-10)

<sup>32</sup> *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products. Revision 2005* (pp9-10)

### Orphan devices

No specific information reported.

### Specialised social services

A few patient organisations also offer recreational services, such as summer camps for children or rehabilitation/therapeutic weekends for adult patients. These are usually fully reimbursed by the Ministry of Social Affairs. The *Act on social services for people with disabilities* came into force in 2007, improving the access to social services for rare disease patients: these schemes are reimbursed and are fully funded from social insurance and are coordinated by the Ministry of Social Affairs.

## DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2011 IN CZECH REPUBLIC

### National plan/strategy for rare diseases and related actions

A taskforce (*“Meziresortní a mezioborová komise pro vzácná onemocnění – Interministerial and interdisciplinary commission for rare diseases”*, henceforward “Taskforce”) convenes every other month and has created dedicated working parties with the aim to establish the basis for the National Action Plan by 2013. A group of stakeholders was gathered in 2011 (including the Ministry of Labour and Social Affairs, experts and payers) and a deadline for elaborating the plan has been established for June 2012. The Czech ten-year strategy<sup>33</sup> reveals the budgetary sources for the plan, which will include “existing budgetary chapters and domestic and foreign subsidies” such as the Ministry of Health and the country’s public health insurance. A budget for the strategy has not yet been announced and is in the process of substantiation by the Taskforce. The Ministry of Health has been trying to assure funding within the frame of the EEA Norway Grants scheme (2008-2014) for the National Coordination Centre at University Hospital Motol<sup>34</sup> and via annual Ministry of Health targeted appropriation schemes, both which have deadlines in June 2012.

### Centres of expertise

The Czech National Strategy for Rare Diseases foresees the concentration of care for rare diseases in 10 to 20 different centres, with a National Coordination Centre at the Motol Teaching Hospital in Prague, which will coordinate at the regional level with University Hospital Brno. This centre will assure its funding through the Norway Grants scheme by June 2012. Its major aim, beyond structuring the rare diseases field in the country, would be to identify additional *de facto* centres of expertise and propose their transformation into *de iure* centres by the Taskforce. First steps were taken and at their last meeting (November 2011) the Taskforce decided to officially establish three pilot *de iure* rare diseases centres for a) cystic fibrosis<sup>35</sup>, b) metabolic diseases<sup>36</sup> and c) epidermolysis bullosa<sup>37</sup>, based on a compiled criteria drawn from the published EUCERD and EURORDIS centres of expertise recommendations.

Establishment of centres gives them government recognition, but still does not imply a dedicated budget line. Treatment and diagnostics will thus continue to be provided within standard health insurance procedures administered by a group of health insurance companies. However, in the future and following gradual reform of health care funding (after 2012) it is planned that respective rare diseases or related rare disease diagnostic groups will be concentrated into dedicated centres. In this regard it is expected that the major condition for health care reimbursement would be centre-based care. In addition, it is expected that in duly justified instances (e.g. very rare diseases) care will be assured within European Reference Networks (i.e.

<sup>33</sup> [http://ec.europa.eu/health/rare\\_diseases/docs/national\\_czech\\_en.pdf](http://ec.europa.eu/health/rare_diseases/docs/national_czech_en.pdf)

<sup>34</sup> [http://www.mfcr.cz/cps/rde/xchg/mfcr/xsl/Financial\\_Mechanism\\_EEA.html](http://www.mfcr.cz/cps/rde/xchg/mfcr/xsl/Financial_Mechanism_EEA.html)

<sup>35</sup> [www.cfklub.cz](http://www.cfklub.cz), [www.cfregistr.cz](http://www.cfregistr.cz), [ublg.lf2.cuni.cz](http://ublg.lf2.cuni.cz)

<sup>36</sup> [www.udmp.cz](http://www.udmp.cz)

<sup>37</sup> [www.debra.cz](http://www.debra.cz)

in line with the EC Directive on cross-border healthcare sections 54 and 55). Currently, cross-border healthcare falls into the domain of the Centre for International Reimbursements<sup>38</sup>.

### **Neonatal screening policy**

In early 2011 the National coordination centre for neonatal screening was officially established by the Czech Ministry of Health<sup>39</sup> and its operation is funded by targeted annual appropriation schemes.

### **Genetic testing**

In November 2011 a new law On Specific Health Care Services 373/2011 Sb was passed. It stipulates (Part 6; section 28) that germinal genome is allowed to be tested within the context of rare diseases in genetic laboratories that are accredited according to the ISO 15189 norm, in accordance with "OECD guidelines for quality assurance in molecular genetic testing"<sup>40</sup>. Moreover, new law 372/2011 Sb stipulates provisions regarding informed consent procedures in the domain of health care services.

### **National alliances of patient organisations and patient representation**

In December 2011 foundations were established for the creation of an overarching Czech National Alliance for Rare Diseases<sup>41</sup>. This alliance will link together other patient support groups via a democratic, bottom-up, activity spearheaded by several larger patient support groups. Its registration with the Ministry of Interior is expected to be completed by May 2012, including establishment of its bylaws and governance structure. This association plans to join EURORDIS and participate in its activities. Creating an alliance for rare disease patient groups is a provision of the national strategy being developed, together with the Coalition for Health Association<sup>42</sup>. The Coalition for Health Association includes all diseases, while the Czech National Alliance for rare diseases will collaborate on topics of common interest. However, there are 43 patient organisations in the Czech Republic. Some groups benefit from aid from the Ministries of Health and of Labour and Social Affairs; the system will be streamlined under the National Action Plan, since representatives of the Coalition for Health are members of its Taskforce. Following official registration of the Czech National Alliance for Rare Diseases its representatives will be included in the Taskforce.

### **Sources of information on rare diseases and national help lines**

#### **Help line**

A help line for rare diseases is under preparation and is planned to be funded within the Norway Grants mechanism with the National Coordination Centre for Rare Diseases in University Hospital Motol.

### **National rare disease events in 2011**

Amongst the rare disease events hosted by the Czech Republic in 2011 were the Metabolic Days<sup>43</sup> (Mikulov, 11-13 May 2011), Czech National Genetics Meeting<sup>44</sup> (Třeboň, 7-9 September 2011) and the Czech National DNA diagnostics conference<sup>45</sup> (Prague, 24-25 December 2011).

### **Research activities and E-Rare partnership**

#### **IRDIRC**

Czech funding agencies are not yet committed members of the IRDiRC.

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<sup>38</sup> <http://www.cmu.cz/en?Itemid=224>

<sup>39</sup> [www.novorozenecky-screening.cz](http://www.novorozenecky-screening.cz)

<sup>40</sup> <http://www.oecd.org/dataoecd/43/6/38839788.pdf>

<sup>41</sup> [www.vzacna-onemocneni.cz/](http://www.vzacna-onemocneni.cz/)

<sup>42</sup> [www.koaliceprozdravi.cz](http://www.koaliceprozdravi.cz)

<sup>43</sup> [www.dedicnemetabolickeporuchy.cz/](http://www.dedicnemetabolickeporuchy.cz/)

<sup>44</sup> [www.trebon2011.slg.cz/](http://www.trebon2011.slg.cz/)

<sup>45</sup> [www.dnakonference.cz](http://www.dnakonference.cz)

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## SELECTED BIBLIOGRAPHY AND SOURCES<sup>47</sup>

- Ministry of Health  
[www.mzcr.cz](http://www.mzcr.cz)
- Czech National Strategy for Rare Diseases website  
[www.vzacnenemoci.cz](http://www.vzacnenemoci.cz)
- *Czech National Strategy for Rare Diseases*  
[http://ec.europa.eu/health/rare\\_diseases/docs/national\\_czech\\_en.pdf](http://ec.europa.eu/health/rare_diseases/docs/national_czech_en.pdf)
- Czech site concerning national rare disease activities  
[www.vzacna-onemocneni.cz/](http://www.vzacna-onemocneni.cz/)
- Neonatal screening website  
<http://novorozenecky-screening.cz/>
- *SUKL State Institute for Drug Control*  
[www.sukl.cz](http://www.sukl.cz)
- Orphanet Czech Republic national website  
<http://www.orphanet.cz/national/CZ-CS/index/%C3%BAvod/>

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<sup>46</sup> The contributors and validators of the report have contributed information which is accurate to the best of their knowledge. However, readers should take note that the contents of this report are illustrative and not exhaustive.

<sup>47</sup> All websites and documents were last accessed in May 2012. A more detailed list of sources is available in the full report:  
<http://www.eucerd.eu/upload/file/Reports/2012ReportStateofArtRDActivities.pdf>