

## **Press release concerning material being considered by the Czech Government**

### **"National Strategy for rare diseases 2010-2020"**

Rare diseases are complex, mainly genetic (or congenital) diseases with low incidence among the population which have an impact on the patient's quality of life and social integration and can even be life-threatening. Although the individual diseases are rare, taken together their morbidity and mortality rate in the EU is as high as 8 %. In most rare diseases there is no effective causal treatment, yet it is possible to improve the person's quality of life and life expectancy by means of suitable care.

Among current shortcomings in the field of rare diseases are insufficient identification of rare diseases in the International Classification of Diseases, deficiencies in their timely diagnosis and treatment, imbalance and insufficient quality of services provided, inadequate research, shortcomings in data collection and lack of effective care for patients with rare diseases.

In the Czech Republic there is as yet no unified approach to rare diseases. The proposed National Strategy sums up the issue of rare diseases from the EU's and the Czech Republic's point of view and proposes major targets and measures for improving the situation in the Czech Republic. These targets and measures will be specified in more detail in the National Action Plan, which establishes sub-tasks, instruments, responsibilities, dates and indicators for fulfilling individual tasks.

**The National Strategy is intended 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.**

The purpose of the National Strategy is also to make use of expert cooperation with other countries, to enable Czech patients to take part in international clinical studies of new medicines, including treatment abroad in strictly identified cases when it is not possible to obtain adequate specialised care in the Czech Republic.

On 11 November 2008, the European Commission adopted the "Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's challenges" and, within that communication, the Council Recommendation on an action in the field of rare diseases, which was adopted by the Council on 9 June 2009 during the Czech Republic's Presidency of the EU Council.

### III.

## **"National Strategy for rare diseases 2010-2020"**

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## **"National Strategy for rare diseases 2010-2020"**

### **1. Introduction**

Rare diseases are clinically heterogenous diseases, which have their very low prevalence in common. Incorrect or late diagnosis of those diseases often leads to irreversible damage to the patient's health or even death.

Among the current shortcomings in the area of rare diseases are insufficient identification, serious deficiencies in the diagnosis and treatment of such diseases, imbalance in and inadequate quality of services provided, very limited choice of medicines for treatment, shortcomings in data collection at national and regional level and lack of effective care for patients with rare diseases, including a lack of awareness of such diseases among medical professionals and the general public. One consequence of the chronic and often progressive nature of those diseases is that shortcomings often come to light indirectly in the social and legal fields.

In the Czech Republic there is as yet no standard approach to rare diseases. The National Strategy for Rare Diseases 2010-2020 (hereinafter the National Strategy) sums up the issue of rare diseases 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 Czech Republic.

Among the National Strategy's main objectives are early diagnosis and access to appropriate treatment of rare diseases, coordination and centralisation of effective treatment for patients with rare diseases, improvement in the provision of information for professionals and the general public, coordination at national and international level with patients' organisations and improved identification of rare diseases in the International Classification of Diseases (ICD). More detailed tasks, instruments, responsibilities, deadlines, possible costs and financial indicators for performing different activities will be specified in the National Action Plan for rare diseases (hereinafter the National Action Plan).

The National Strategy 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.

### **2. Usual characteristics of rare diseases**

Rare diseases are clinically heterogenous, mainly genetic (or congenital) polysystemic diseases with very low incidence (prevalence) among the population. They have an impact on the patient's quality of life and social integration, and can even be life-threatening. Diseases are defined by the EU as rare when they affect fewer than five persons in 10 000 (i.e. fewer than one patient in 2 000 individuals). The serious nature of rare diseases stems from the fact that there are more than

8 000 such diseases, which means that the overall number of patients is considerable despite the low prevalence of individual clinical cases. A list of rare diseases known thus far, across all medical fields, can be found on the website of Orphanet ([www.orpha.net](http://www.orpha.net)), the European research consortium, which receives long-term support from the European Commission .

Rare diseases manifest themselves most often shortly after birth, affecting 4-5 % of newborn babies and infants (for example some congenital deformities, genetic metabolic diseases, genetically determined diseases and rare tumours), but can appear later during childhood or adulthood. Some 80 % of rare diseases are genetic in origin, yet in most patients the etiology of their disease remains undetected. Absent or late diagnosis, particularly in the case of patients with treatable diseases, leads to irreversible damage to health. In such cases, not only patients but also their families become traumatised and lose confidence in the health system. Early, accurate diagnosis of rare diseases is contingent on available methods of diagnosis and on the doctors concerned having received the appropriate training.

As regards health policy, it is vital to develop neonatal screening for rare diseases for which effective treatment is already available, given that early diagnosis for newborn babies, specialist advice on genetics and various forms of prenatal diagnosis are among the most effective methods of prevention in the field. The treatment of patients with rare diseases is effective only if performed by a multi-disciplinary health team in specialist centres that, according to EU recommendations, are to be created at national and regional levels. Those centres, which are to deal with the diagnosis and treatment of rare diseases and provide advice for doctors in the field, will be described in more detail in the National Action Plan. We propose to put in place around 10 to 20 national centres targeting groups of related diseases and transnational cooperation.

A list of rare diseases and essential information on those diseases for professionals and the general public will be published at [www.vzacnenemoci.cz](http://www.vzacnenemoci.cz) and are set out in the seven main European languages on the European Orphanet website.

### **3. The EU and rare diseases**

In response to the "Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's challenges", the topic of rare diseases has been discussed in the working groups of the European Council and the European Parliament. Subsequently, during the Czech Republic's Presidency of the EU ([www.eu2009.cz](http://www.eu2009.cz)), the European Council adopted the "Council Recommendation on an action in the field of rare diseases" (<http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF>).

The content of those documents stems from the recommendations made by the European Commission's multi-disciplinary working group, DG SANCO's Rare Disease Task Force ([www.rdtf.org](http://www.rdtf.org)), the Orphanet project ([www.orpha.net](http://www.orpha.net)) and the European Organisation for Rare Diseases (Eurordis)([www.eurordis.org](http://www.eurordis.org)). The documents' content is also in line with the ongoing European Project for Rare

Diseases National Plans Development (EUROPLAN [www.europlanproject.eu](http://www.europlanproject.eu)) as part of the "Community Action in the Field of Public Health" programme.

The EU's main goals in the field of rare diseases are:

- 1) to improve the identification of rare diseases
- 2) to support the development of health policy in this field and to ensure that care is more effective
- 3) to develop European cooperation, coordination and regulation in this field.

The Council's recommendations for action in the area of rare diseases include:

- establishing national plans for rare diseases in the Member States in order to ensure that patients with rare diseases have universal access to high-quality care, including diagnostics, treatment and orphan drugs on the basis of equal treatment and solidarity throughout the EU;
- establishing appropriate definition, codification and cataloguing of rare diseases;
- performing research into the causes and treatment of rare diseases;
- setting up coordination centres and European reference networks for rare diseases;
- gathering expertise on rare diseases at European level;
- enhancing cooperation with organisations of patients with rare diseases;
- promoting sustainable activities in the field of rare diseases.

In accordance with those recommendations, the EU Member States have the possibility to use specialised consultancy across borders, take part in international clinical studies of new medicines or treat patients abroad in cases where it is not possible to obtain specialised care in the EU Member State.

Further information can be found on the EU's website:

[http://ec.europa.eu/health/ph\\_threats/non\\_com/rare\\_diseases\\_en.htm](http://ec.europa.eu/health/ph_threats/non_com/rare_diseases_en.htm).

#### **4. The Czech Republic and rare diseases**

To date, there are no statistics available in the Czech Republic on the prevalence of rare diseases, given that such diseases are not yet appropriately classified in the ICD-10. It is estimated that there are some 20 000 patients with such diseases. Around 80 % of rare diseases are genetic in origin, and the remainder are the result of infections, auto-immunity, allergies and negative environmental impact. Given that most diseases develop at the prenatal stage due to their genetic nature, over 75 % of rare diseases manifest themselves in children aged under 10. Causal treatment is

available only for a small number of those diseases, whereas with most such diseases long-term palliative care is given. Unfortunately, therefore, around one third of patients with rare diseases do not currently live past the age of five. Consequently, the only possibility for helping families with a child suffering from a rare disease is to ensure – with the help of targeted prenatal diagnosis, or even, now, pre-implant diagnosis – the birth of a healthy second child.

The Czech Republic has recently developed a number of activities in the field of care for patients with rare diseases, with a view to ensuring equal access to health services, high-quality health care and patient safety.

#### **4.1. Finance and rare diseases**

Rare diseases are usually diagnosed by a specialist laboratory and by means of auxiliary examination. The process of diagnosis takes place in several stages, is time-consuming and draws on the expertise of highly educated specialists.

The treatment of some rare diseases is costly; for example, some medicines used for enzyme replacement treatment currently cost up to CZK 20-30 million per year per patient, whereas the treatment of certain other rare diseases, such as certain genetic metabolic diseases, costs tens to hundreds of thousands of CZK per year.

There are currently no valid data available on the exact cost of treating rare diseases. Only around 200 of the 8 000 rare diseases are codified in ICD-10, which means that such diseases cannot be statistically monitored. The ICD-11 revision, with the complete classification of rare diseases, will not be published until 2012. There are currently no available data on levels of funding from health insurance companies for different diagnoses, in particular the treatment of secondary symptoms of rare diseases. Given that the national coordination centre for rare diseases has yet to be set up, general practitioners have no opportunity, when carrying out their different diagnostic assessments, to seek advice or help from specialists. As such, patients with rare diseases are often treated on the basis of incorrect diagnoses, their condition gradually worsens because of inappropriate treatment and their costs therefore increase disproportionately.

According to the European recommendations drawn up by the Council and by Eurordis patients' organisations ([www.eurordis.org](http://www.eurordis.org)), the cost of treating complications arising from the late diagnosis of a rare disease can actually be reduced by centralising care for patients with such diseases. Centres of reference with the necessary inter-disciplinary capacity and sufficient experience in the early diagnosis and long-term therapy of rare diseases ensure that patients can be given high-quality and effective treatment. The European recommendations state that such centres should also have international links and should be involved in international initiatives or European grant projects in the field.

Both pre-implant diagnosis, which does not necessarily entail the interruption of pregnancy, and 'classic' prenatal diagnosis in the early stages of pregnancy lead to

considerable financial savings. Both methods enable affected families to have healthy children.

As regards the early diagnosis of rare diseases, it is possible – thanks, for example, to the spread of neonatal screening – to diagnose diseases before the clinical symptoms develop and, by means of early, targeted treatment, to forestall organ-related complications. This view is strongly backed by a 10-year cost-benefit study into cystic fibrosis, according to which the cost of treating patients with cystic fibrosis diagnosed by means of neonatal screening is significantly lower than that of treating patients diagnosed with cystic fibrosis at a later stage. (Economic implications of newborn screening for cystic fibrosis: a cost of illness retrospective cohort study. Lancet 2007).

A further economic benefit of both forms of prenatal genetic prevention is that the time in which mothers remain outside work as a result of the onerous, long-term care of the child is significantly reduced.

In the light of the above, it is not currently possible to estimate medium-term health care costs in this field. Such a prediction can only be made once the statistical system for monitoring rare diseases has been updated to cover all rare diseases. It will then be possible to monitor rare diseases statistically in the context of the proposed centralised and coordinated care as part of the National Action Plan.

The cost of the proposed objectives and corrective measures, which will be specified in more detail in the National Action Plan, will be covered under existing budgetary chapters and domestic and foreign subsidies. For example, the CZK 5-10 million to set up the National Coordination Centre and to concentrate care in specialised centres will be covered in full by resources approved for Chapter 335 - Ministry of Health. The three-year National Action Plan will be funded from different sources to be specified in detail in the plan (public health insurance, domestic and foreign subsidies, etc.).

## 4.2. Orphan drugs

Medicinal products for rare diseases, also known as orphan drugs, are often very expensive. The situation in this regard has improved considerably in recent years, with such products being developed as medicine has progressed, and then introduced into medical practice. This process is coordinated by the *Státní ústav pro kontrolu léčiv* - SUKL [www.sukl.cz](http://www.sukl.cz) (State Institute for Drug Control) in Prague, in conjunction with the European Medicines Agency (EMA; [www.ema.europa.eu/](http://www.ema.europa.eu/)) and its specialist committee, the Committee for Orphan Medicinal Products ([www.ema.europa.eu/htms/general/contacts/COMP/COMP.html](http://www.ema.europa.eu/htms/general/contacts/COMP/COMP.html)).

The availability of orphan drugs in the Czech Republic is among the highest in the EU. 27 of the 62 orphan drugs registered in the EU via what is known as the centralised procedure are currently categorised in the Czech Republic. Efforts are being made there to ensure that most registered orphan drugs are covered by public health insurance. As regards the setting of prices and the reimbursement of orphan

drugs in the Czech Republic, the procedure is the same as for any other medicinal product.

Under Czech law it is possible to give preference to what are known as highly innovative medicinal products. The status of 'highly innovative medicinal product' can be acquired by both traditional medicinal products and orphan drugs provided they meet the conditions laid down in Decree No 92/2008 on the list of countries for the reference basket, the means of evaluating the amount, conditions and form of reimbursement of medicinal products and food products for special medical purposes and the requirements for making an application for reimbursement. Most orphan drugs covered by public health insurance have a fixed reimbursement amount that is below the maximum price for the final consumer.

In the case of new orphan drugs covered by public health insurance, it will be necessary to ensure the fiscal (budgetary) balance of the public health insurance system. To this end, an instrument should be created that will ensure the expected financial stability in proportion to the abovementioned principles of access to care. The National Action Plan will cover this matter in more detail.

#### **4.3. International projects**

The Czech Republic is taking part in a number of international projects in the field of rare diseases, namely:

1. Orphanet ([www.orpha.net](http://www.orpha.net)), which concerns the development of a Europe-wide database on rare diseases for doctors, patients and their families.
2. EuroGentest ([www.eurogentest.org](http://www.eurogentest.org)), dealing with the harmonisation and standardisation of genetic diagnostic services at European level relating to rare diseases.
3. ECORN ([www.ecorn-cf.eu](http://www.ecorn-cf.eu)) on specialist e-Health information for patients with cystic fibrosis, a typical example of a rare disease.
4. EuroCareCF ([www.eurocarecf.eu](http://www.eurocarecf.eu)) on research into diagnosis and treatment, including the creation of a Europe-wide register of patients with cystic fibrosis.
5. ERDNIM ([www.erndim.unibas.ch](http://www.erndim.unibas.ch)), on the organisation of external quality control in the field of biochemical genetics.
6. RAPSODY (Rare Disease Patient Solidarity; [www.rapsodyonline.eu](http://www.rapsodyonline.eu)) - the SUKL took part in this project, holding a national seminar of patients with rare diseases in March 2007 on the theme of the specialised centres, and in so doing became involved at international level. Subsequently, in July 2007, it hosted an international seminar in Prague on the same theme, which was attended by 80 representatives of patients' organisations, specialist doctors, heads of health institutes and the European Commission.
7. EUROPLAN ([www.europlanproject.eu](http://www.europlanproject.eu)), which is aimed at putting together general recommendations for national plans for rare diseases in the EU Member States.

8. CHERISH ([www.cherishproject.eu](http://www.cherishproject.eu)), which is aimed at improving the diagnosis of mentally handicapped children.

9. ENCE/CF/LAM/LTX ([www.ence-plan.eu](http://www.ence-plan.eu)), which, using three rare diseases as its model, is aimed at laying the foundations for the creation of European centres of reference (expertise).

10. NEUROPED (European Network on Rare Pediatric Neurological Diseases - [www.neuroped.eu](http://www.neuroped.eu)), which coordinates European cooperation on research into rare neurological diseases in children.

11. EMQN ([www.emqn.org](http://www.emqn.org)), which organises the external quality control system for molecular genetics laboratories for genetic diagnosis of rare diseases.

12. EUROCAT ([www.eurocat-network.eu](http://www.eurocat-network.eu)), which coordinates European and national data collection in the monitoring of the presence of congenital deformities.

#### 4.4. Neonatal screening for rare diseases (NS)

The objective of NS is the rapid diagnosis and early treatment of newborn babies suffering from certain genetic metabolic and endocrine diseases classified as rare diseases. All newborn babies in the Czech Republic have, since 1 October 2009, undergone neonatal screening for the following 13 diseases (of the 45 thus far detected):

1. congenital hypothyroidism
2. congenital adrenal hyperplasia
3. phenylketonuria and **hyperphenylalaninemia**
4. maple syrup disease
5. medium-chain Acyl CoA dehydrogenase deficiency
6. long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency
7. very-long-chain Acyl CoA dehydrogenase deficiency
8. carnitine palmitoyltransferase I deficiency
9. carnitine palmitoyltransferase II deficiency
10. **Carnitine**-acylcarnitine **translocase** deficiency
11. glutaric aciduria type I
12. isovalric acidemia
13. cystic fibrosis

Previously, neonatal screening in the Czech Republic was carried out for only three diseases: congenital hypothyroidism, phenylketonuria and congenital adrenal hyperplasia. As such, the 2009 extension of the programme brought the Czech Republic into line with the developed European countries. The provision of NS is covered in full by public health insurance.

NS provision is coordinated by the National Neonatal Screening Coordination Centre set up in Prague's *Všeobecná fakultní nemocnice* (University General Hospital). Information on NS for specialists and the general public has, since October 2009, been available on the following website: [www.novorozeneckyscreening.cz](http://www.novorozeneckyscreening.cz).

#### 4.5. Specialised centres of reference for rare diseases

Until now, there has been no network of specialised centres for rare diseases and no single concept and definition of the care offered by such centres. However, there are specialised centres of reference, such as the National Reference Centre for the Research and Treatment of Gaucher's disease, the National Centre for the Diagnosis and Treatment of Cystic Fibrosis, the Centre for the Diagnosis and Treatment of Fabry Disease, the Centre for Pulmonary Hypertension, the Centre for Hereditary Ataxia and the Complex Oncology Centres, which treat rare tumours. A national centre has been set up at Brno University Hospital for treating skin diseases such as epidermolysis bullosa and a Centre for Cochlear Implants in Children at the Motol University Hospital for severe hearing deficiencies. In the field of genetic diagnostics, a National Reference Laboratory for DNA Diagnosis, with a list of over 400 diagnosed diseases ([www.uhkt.cz/nrl/db](http://www.uhkt.cz/nrl/db)), has been set up at the Institute of Haematology and Blood Transfusion in Prague.

A Neonatal Screening Coordination Centre has been set up at Prague University Hospital and a national coordination centre for rare diseases has been set up at Motol University Hospital.

The creation of other centres and the setting of conditions to be met in such centres will be discussed and then described in detail in the National Action Plan, in line with the international recommendations issued by Eurordis ([www.eurordis.org/IMG/pdf/position-paper-EURORDIS-centres-excellence-networksFeb08.pdf](http://www.eurordis.org/IMG/pdf/position-paper-EURORDIS-centres-excellence-networksFeb08.pdf)), including future recommendations from the **newly established** European Union Committee of Experts on Rare Diseases (EUCERD; [www.eucerd.eu](http://www.eucerd.eu)).

#### 4.6. Future development

As regards pre-symptomatic diagnostics, neonatal screening is expected to be extended to the whole population in respect of treatable genetic metabolic diseases and the rapid and extensive arrival of new technologies for analysing the human genome. It is predicted that the complete sequencing of a person's genome will be technically possible in the years to come. As a result of such a test (carried out once in a person's life), it will no longer be necessary to determine all indicative and diagnostic criteria, and the number of DNA tests will be significantly reduced, although confirmatory assays will not be affected. Knowledge of genome information and the wider availability of such information will require entirely different specialisations beyond the current level of clinical and biochemical genetics. It will be necessary to set up teams of experts in genomics, molecular pathologies, biostatistics, bioinformatics and medical ethics, which will be capable of interpreting a person's genome information effectively and on the basis of evidence (based on current medical information and "evidence-based medicine") and of harnessing that information to the person's benefit. In this regard, several centres enjoying support from various sources should gradually be created in order to introduce evidence-based technical genome analysis, to create teams of experts with the required specialisations and gradually train experts in the field.

In the field of treating rare diseases, progress is expected to be made in developing approaches aimed at targeted biological treatment and treatment affecting mutant protein conformation, the development of protein replacement therapy, RNA interference and other methods of genome treatment. Given that such treatment is very expensive, mechanisms must be set up in advance for assessing the effectiveness of these new procedures and for ensuring funding for the highly specialised care provided by dedicated centres, in line with current and future international recommendations in the area.

## **5. Objectives and proposed measures**

### **5.1. Objectives**

To ensure that all patients with rare diseases have access to high-quality care, including diagnosis, treatment and orphan drugs, on the basis of equal treatment and solidarity and to improve the effectiveness of the diagnosis and care provided to patients with rare diseases.

### **5.2. Measures proposed:**

#### **5.2.1. Improved information on rare diseases**

- A rare diseases website – the creation of such a website has received support as part of the Health Ministry's grants programme and will be operational in 2010.
- Patients – patients' organisations set up; they will participate in working group meetings.
- Health workers – professional establishments, medical practitioners' organisations.

The main activity in this field will be to create the rare diseases website and to support our participation in the Orphanet project, which will ensure that patients and doctors receive essential information translated into all EU languages, including Czech.

#### **5.2.2. Education in the field of rare diseases**

- Patients – website, experts' seminars, joint initiatives of patients' organisations.
- Professionals – undergraduate and postgraduate training of doctors, general medical staff, non-medical health workers, lifelong learning in this field

#### **5.2.3. Improved diagnosis and screening of rare diseases**

- Full-scale neonatal screening in the Czech Republic should, where possible, gradually be extended to the early diagnosis, for example, of sensory deficiencies (hearing and eyesight), and other already treatable genetic metabolic diseases.
- Selective screening of (symptomatic) patients with rare diseases and healthy (asymptomatic) carriers of genetic conditions for rare diseases, in line with international recommendations or in the context of relevant research projects.

- Diagnostic laboratories – supporting their registration in the database of the National Reference Laboratory for DNA diagnosis at the Institute of Haematology and Blood Transfusion in Prague (<http://www.uhkt.cz/nrl/db>), accrediting such laboratories pursuant to Standard ISO 15189 in cooperation with the *Český institut pro akreditaci o.p.s* - [www.cai.cz](http://www.cai.cz) (Czech Accreditation Institute, a non-profit organisation), implementing the recommendations contained in the OECD Guidelines for Quality Assurance in Molecular Genetic Testing from 2007 ([www.oecd.org/dataoecd/43/6/38839788.pdf](http://www.oecd.org/dataoecd/43/6/38839788.pdf)) in diagnostic practice, setting minimum conditions for human and technical resources of laboratories in cooperation with the relevant professional establishments of the Jan Evangelista Purkyně Czech Medical Association ([www.cls.cz](http://www.cls.cz)), implementing a licencing system for such laboratories by the Ministry of Health and public health insurance in the form of generally binding legal provisions, the creation of national reference laboratories for different groups of rare diseases – setting indicative and diagnostic criteria for molecular genetic diagnostics in line with the development of international recommendations and evidence-based medicine, with reference to current and future international recommendations in this field.

#### **5.2.4. Improvement in the quality of treatment and care**

- Centralisation of care for patients with rare diseases:
  - analysis of existing centres, proposal for a network of centres, setting of criteria for admission to the network of centres and definition of the conditions for providing care.
  - setting up a National Coordination Centre for Rare Diseases, with the aim of guaranteeing and coordinating activities in the field of rare diseases, from a specialist perspective. Where applicable, setting up national reference centres for individual diseases or their clinical/diagnostic groups. Such centres should be created either institutionally (i.e. as part of existing or proposed centres, usually within university hospitals) or 'virtually' (i.e. functional centres stemming from institutional cooperation between various centres).
- Recommended procedures for diagnosis and treatment, including nursing procedures, analysis of the current situation, proposals for necessary standards and their gradual integration, along with regular assessment of the effects of treatment.
- Secondary prevention for patients with rare diseases – drafting of an expert recommendation for monitoring selected groups of rare diseases.
- cooperation between all interested parties – namely, doctors, health insurance, the SUKL and the Ministry of Health – in ensuring effective pharmacotherapy pursuant to internationally recognised standards.
  - Orphan drugs – better availability and effectiveness of care, recording of medicinal product consumption, monitoring of the effectiveness and costs of treatment.
  - Cataloguing other medicines needed by patients with rare diseases but not covered by public health insurance.

### **5.2.5. Improvement in the quality of life and social integration of people with rare diseases**

- Introduction of the ICF (International Classification of Functioning, Disability and Health [www.who.int/classifications/icf/en/](http://www.who.int/classifications/icf/en/)) as a means of assessing the impact of disability on the quality of life and of monitoring the effectiveness of treatment in improving the social skills of people with rare diseases; analysis of the needs of such persons pursuant to the ICF.
- Improving inter-ministerial cooperation between the Ministries of Health, Employment and Social Affairs, Agriculture, Education, Youth and Sport, Regional Development and Finance.
- Setting up a social-health network

### **5.2.6. Support for science and research in the field of rare diseases**

National level – ministry-level research

a) effective support for research

IGA MZ - *Interní grantová agentura MZ* [www.mzcr.cz](http://www.mzcr.cz) – Health Ministry Internal Grant Agency

GA ČR - *Grantová agentura ČR* [www.gacr.cz](http://www.gacr.cz) – Czech Grant Agency

TA ČR - *Technologická agentura ČR* [www.tacr.cz](http://www.tacr.cz) – Czech Technology Agency

b) Institutional support for research carried out up to 2012 via research projects

c) regular establishment of thematic topics by the national agencies

- inclusion of research into rare diseases in the Czech Republic's long-term research priorities.
- creation of a system of cooperation with patients' organisations and their involvement in research and clinical studies in the field of rare diseases.

International level

Information on research opportunities abroad in the context of:

- DG SANCO / EUCERD
- DG Research (framework programmes)
- COST ([www.cost.esf.org/](http://www.cost.esf.org/))
- Cooperation with the Technological Centre of the Academy of Sciences of the Czech Republic ([www.tc.cz](http://www.tc.cz)) to investigate international cooperation opportunities
- development of cooperation with the USA – NIH programmes for rare diseases ([www.rarediseases.info.nih.gov](http://www.rarediseases.info.nih.gov)).
- ERA-net on rare diseases, for example cooperation with the E-rare2 project ((ERA-net for research programmes for rare diseases; [www.e-rare.eu](http://www.e-rare.eu)).

### **5.2.7. Harmonisation and development of data collection and biological sampling in connection with rare diseases**

- National data collection in cooperation with the *Ústav zdravotních informací a statistiky - ÚZIS* [www.uzis.cz](http://www.uzis.cz) (Institute of Health Information and Statistics) and connection with EU initiatives in the field

- Legal framework for data collection and biological sampling in accordance with the requirements laid down by the *Úřad na ochranu osobních údajů* [www.uouu.cz](http://www.uouu.cz) (Personal data protection office) and with the relevant national and international legal standards.
- Development of the collection and long-term storage of biological samples (biobanking) in compliance with international recommendations and legal provisions, including financing from various sources – see point 5.2.6.

#### **5.2.8. Development of international cooperation in the field of rare diseases**

##### a) Provision of care

- Cross-border cooperation (expert level, organisation of specialist advisory services for Czech patients abroad in strictly defined cases).

##### b) Exchange of experience, data and information

- ORPHANET
- EURORDIS
- ERNDIM
- EUROCAT
- and other current and future EU projects

#### **5.2.9. Cooperation with the World Health Organisation**

Such cooperation is important as regards the development of the proposed standard codification of rare diseases for the planned ICD-11 revision and in cooperation with the Orphanet consortium.

#### **5.2.10. Support and strengthening of the role of organisations of patients with rare diseases**

Cooperation with patients' organisations in the Czech Republic, setting up a representative group with a mandate to act on their behalf at national and international level, including the development of cooperation with Eurordis. More information on their activities on the rare diseases website currently being set up (see above).

#### **5.2.11. Strengthening the role of patients from the Czech Republic with rare diseases in clinical tests of new medicinal products at European level**

This objective is important in ensuring that Czech patients have access to modern treatment and relates to our participation in the Orphanet project and the SUKL's activities in this field.

#### **5.2.12. Cooperation on the European Commission's EuroPlan project**

Cooperation with this project is very important as it will enable us to draw on international experience to create a National Action Plan. The main purpose of the project is to lay down requirements for drawing up national action plans at Europe-wide and national level.

### **5.2.13. Sustainable activities in the field of rare diseases**

- at national level (local, regional and national)
- international level
- multi-source financing

### **5.2.14. Setting up an inter-ministerial working group on rare diseases**

In order to be able to coordinate a systemic inter-ministerial approach to resolving problems relating to rare diseases in accordance with the National Strategy for the Prevention of Rare Diseases 2010-2020, it is essential for the Ministry of Health to set up an inter-ministerial working group. This group should comprise representatives of ministries (Health, Labour and Social Affairs etc), professional organisations belonging to the Czech Jan Evangelista Purkyně Medical Association, health insurance companies, the national medicines inspectorate, the WHO's office in the Czech Republic, the Czech Institute of Health Information and Statistics, patients' organisations, specialists in new analytical methods and technologies, genomics, bioinformatics and medical ethics.

## **6. Conclusions**

Rare diseases are complex, mainly genetic (or congenital) diseases with relatively low incidence among the population, which have a major impact on the patient's quality of life and social integration and can even be life-threatening or reduce the person's lifespan.

Although the individual diseases are rare, taken together their morbidity and mortality rate is as high as 8 % in the EU, so millions of people are affected. In the case of most rare diseases there is no effective causal treatment, yet it is possible to extend and improve the quality of life by means of suitable care. There is great hope for modern methods of therapy and the proposed introduction of orphan drugs into medical practice.

The current shortcomings in the field of rare diseases are the insufficient identification of rare diseases in the International Classification of Diseases, shortcomings in their early diagnosis and treatment, imbalance in and insufficient quality of services provided, inadequate research, shortcomings in data collection and lack of effective care for patients with rare diseases.

In the Czech Republic there is as yet no unified approach to rare diseases. The proposed national strategy sums up the issue of rare diseases from the EU's and the Czech Republic's point of view and proposes major targets and measures for improving the situation in the Czech Republic. These targets and measures will be specified in more detail in the context of the appropriate national action plan, which establishes sub-tasks, instruments, responsibilities, dates and indicators for fulfilling individual tasks. **The National Strategy is intended to ensure the effective diagnosis and treatment of rare diseases, to ensure that all patients with rare diseases have access to high-quality health care and ensure their subsequent social integration on the basis of equal treatment and solidarity.** The purpose of the national strategy is also to make use of expert cooperation with other countries, to enable some Czech patients to take part in international clinical studies of new

medicines, including treatment abroad in strictly identified cases when it is not possible to obtain suitable specialised care in the Czech Republic.

On 11 November 2008, the European Commission adopted the "Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's challenges" and, within that communication, the Council Recommendation on an action in the field of rare diseases, which was adopted by the Council on 9 June 2009 during the Czech Republic's Presidency of the EU Council.