

WORK PLAN FOR THE FIELD OF RARE DISEASES IN THE REPUBLIC OF SLOVENIA

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Introduction

The expression 'rare diseases' derives from the observation that many diseases, particularly metabolic, have several common characteristics when diagnosed and treated, although they may differ in their clinical picture and manner of treatment. These diseases are not only rare, but frequently also chronic, advancing and degenerative. Some 80 per cent of cases are of genetic origin, and in 75 per cent, they affect children and are frequently incurable. The majority of rare diseases have a great impact on the quality of life of the affected person and the people closest to them.

There is no common international definition of rare diseases. The action programme of the European Union on rare diseases, including genetic diseases, considers diseases as rare if they affect between 5 and 10,000 people. The above-mentioned definition is also emphasised in the Council Recommendation of 8 June 2009 on a European action in the field of rare diseases (hereinafter referred to as the Recommendation). Among others, the Recommendation states that there are between 5,000 and 8,000 of these diseases, which affect between 6 and 8 per cent of population.

In spite of the lack of accurate epidemiological data on the situation in the Republic of Slovenia, the statistics clearly indicate that this is also an important field of healthcare in this country. The rarity of these diseases frequently causes problems in their identification, and because they are chronic, they frequently require comprehensive medical treatment. The work plan is thus an opportunity for the better harmonisation of efforts by all the partners involved to achieve health care that is comprehensive, accessible, timely and focused on the patient¹.

The work plan considers the needs of stakeholders in the field of rare diseases and the above-mentioned Recommendation, which also recognises the need for a strategy or a plan in this field in individual EU member states.

¹ A patient is an ill person or any other user of health care services in relation to health care workers and health care co-workers or health care service providers, regardless of their health status (Source: *Patient Rights Act*).

Target population

The work plan is intended for patients² with rare diseases and their relatives, and all partners in the health care system whose work can affect the care of these patients, organisations of patients with rare diseases and all residents of the Republic of Slovenia.

Time frame

The work plan will be drafted for the period up to 2020 and serves as a starting point for the preparation of annual action plans.

Goals:

1. definition and monitoring of rare diseases,
2. improvement of early diagnostics and access to suitable medical treatment (orphan drugs, rehabilitation),
3. improvement of mechanisms for a comprehensive approach to rare diseases,
4. improvement of access to information for patients, health care and other expert workers and general public.

1. Definition and monitoring of rare diseases

For the needs of this document, the default **definition** of rare diseases is **up to 5 cases per 10,000 people**. Until the publication of the new International Classification of Diseases i.e. ICD-11, which is anticipated in 2015, the international classification of diseases **ICD-10 and codes at the portal for rare diseases and orphan drugs ORPHANET** (<http://www.orpha.net>) apply. For a suitable classification of rare diseases, cooperation between the competent institutions in the Republic of Slovenia and Slovenian representatives in international bodies, particularly in the European Union and World Health Organisation (hereinafter referred to as WHO) is necessary.

² A patient is any recipient of health care services (Source: <http://en.wikipedia.org/wiki/Patient>).

The monitoring of patients with rare diseases enables better treatment, especially with regard to consistency of those providing health activities and in terms of access to epidemiological data. An agreement on the most suitable method of monitoring patients has to be reached. A discussion on the establishment of **a national register for rare diseases** is particularly important; this must also consider organisational feasibility, personal data protection, development and the maintenance costs of a register. The application of electronic protocols for monitoring the effects of treatment is also recommended. Data collection operators and users who will have access to these data have to be selected.

Treatment quality indicators are also needed, which, in addition to prevalence and incidence data, will aid the preparation of diagnostic and therapeutic protocols and standards, and the planning of the implementation of very costly treatment strategies. The data collection operators and users who will have access to these data will also have to be selected. As a rule, users are personnel who take decisions on the treatment of patients at an individual or aggregate (regional, national) level.

Activities	To classify and codify rare diseases accordingly. To establish a national register for rare diseases. To determine indicators for monitoring treatment efficiency.
Purpose	To improve health care consistency between various providers. To acquire data on the incidence and prevalence of individual diseases. To improve the planning of diagnostic and therapeutic activities. To acquire data for the constant improvement of treatment.

2. Improvement of early diagnostics and access to suitable medical treatment (orphan drugs, rehabilitation)

Early diagnosis is of key importance for the early beginning of treatment and rehabilitation, and thus for the improvement of the quality of life of persons with rare diseases. The establishment of **national reference centres** with qualified staff and suitable diagnostic equipment is important for early diagnosis. Reference centres should be integrated in **international networks of reference centres**, because it is difficult to achieve excellence in the field of rare diseases for a small country such as the Republic of Slovenia.

About 80 per cent of rare diseases are genetic, which is why patients and their families

need efficient access to genetic testing and information.

The study and enablement of optimum routes for cross-border cooperation (telemedicine, travel of experts or patients) would be sensible, because patients frequently receive certain treatments abroad.

The possibility of financing **development and research, and above all, useful projects** in the field of rare diseases has to be envisaged.

A cost-efficient **screening system** for rare diseases is important for early diagnosis. Screening for phenylketonuria and congenital hypothyreosis in the neonatal period and the screening of pregnant women for Down syndrome is already established in the Republic of Slovenia. The continuous assessment of technical possibilities, the related selection of diseases and cost-effectiveness is reasonable. Available data has to be reviewed, and a related policy has to be prepared for implementing screening tests for the entire population or special subgroups while considering all factors (technical, ethical, organisational, financial, social) in the Republic of Slovenia.

It is important that **a system of clinical guidelines** based on evidence is introduced for efficient treatment and rehabilitation.

These guidelines also include the application of **orphan drugs**. The regulatory framework for the classification of new drugs among those for which financing is provided within the framework of compulsory or supplementary health insurance is constantly changing. Regardless of the manner of adopting decisions, or the competent authority, it is important to keep the identified characteristics of drugs used in the treatment of rare diseases. In addition to the costs, ethical aspects also have to be considered in their assessment. By establishing decision-making guidelines for competent bodies, policy in this field has to be defined more clearly.

Drugs used in the treatment of rare diseases are frequently particularly expensive, and the consideration of an additional financial source would be sensible.

Additional forms of financing will enable patient equality and access to health care services for patients with rare diseases, which are two fundamental principles of the Slovenian health care system.

A qualitative network of rehabilitation services has to be established, because special treatments for the majority of rare diseases are not yet available.

Activities	<ul style="list-style-type: none"> To establish national reference centres and their integration into international reference networks. To improve efficiency of access to genetic diagnostics and genetic counselling. To establish a system of clinical guidelines for diagnostics and the treatment of rare diseases or groups of rare diseases. To monitor and, if necessary, adapt the policy of implementing screening tests for rare diseases. To form guidelines for the placement of drugs for rare diseases in programmes of compulsory or supplementary health insurance, and to study the option of acquiring additional sources for funding very expensive drugs. To dedicate funds to development and useful research projects in the field of rare diseases. To develop rehabilitation programmes for patients with rare diseases.
Purpose	<ul style="list-style-type: none"> To improve diagnostics and the early diagnostics of rare diseases. To improve access to treatment (including with orphan drugs) and other medical treatments. To improve the prognosis for the outcome of disease.

3. Improving mechanisms for a comprehensive approach to rare diseases

Rare diseases are usually chronic conditions which require long-term and comprehensive treatment. In some cases, patients require drugs or medical technical devices, and the provision of these is sometimes quite lengthy. **Patients' special needs have to be recognised**, in particular with regard to their right to suitable, qualitative and safe care. While recognising specific needs, the possibility of relaxing the procedures for realising guaranteed rights has to be studied.

The implementation of **primary prevention** has to be supported with the inclusion of programmes of preconception genetic counselling and diagnostics, dietary programmes (e.g. folic acid), information centres for teratogenes, perinatal care for pregnant women with chronic diseases, the prevention of infections in pregnant women, control of exposure to the harmful effects of the environment, and other risk factors during pregnancy.

The operations and active inclusion of non-governmental organisations – associations of patients with rare diseases in the preparation and monitoring of implementation of the work plan has to be supported. Non-governmental organisations (NGOs) are an important source of support and exchange of experience between patients. They can also be a source of valuable information on problems which patients face and of proposals for their alleviation. NGOs are partners within the health care system, not only in the formation of its policies, but also in their implementation. The initiative to establish an umbrella association for sufferers of rare diseases in the Republic of Slovenia seems logical.

Access to suitable social services to enable the **social inclusion of patients** (e.g. inclusion in schools, vocational education, etc.) has to be provided.

Activities	<ul style="list-style-type: none"> To recognise the special needs of patients with rare diseases. To study the possibility of reducing recognised administrative barriers to the provision of patients' rights. To propose special forms of support and the inclusion of children and young people with rare diseases in the school system. To encourage professional qualifications of patients with regard to vocational education and employability. To propose measures for the support of families with persons with rare diseases, including home help. To support the establishment and development of patients' associations as the main discussion partners.
Purpose	<ul style="list-style-type: none"> To improve the timeliness, quality and safety of patients' care. To improve the quality of the lives of patients with rare diseases. To enable patients with rare diseases to provide for their own social security.

4. Developing information for patients, health care and other expert workers and the general public

Patients and health care workers dealing with rare diseases must frequently put a lot of effort into acquiring information on diagnostic procedures, possible treatment methods, qualified providers and other important data related to a disease. Rare diseases have similar characteristics, and patients and experts are faced with similar challenges. It would be logical to establish **a national centre for rare diseases** where useful information for patients and their relatives, health care and other expert workers would

be collected. Experience abroad has shown that it is usually better to appoint an organisation responsible for the described activities from among existing institutions rather than to establish a new one. Publicly accessible information can assist in raising public awareness regarding rare diseases.

The role of such a centre, which could contribute to the development of the discipline by monitoring centres which offer highly specialised health care services at home and abroad, has to be discussed; it could serve as an aid and support to patients by providing them with information directly, or by directing them to NGOs. An important task of such a centre could be participation in the establishment of a reference centre network in the Republic of Slovenia and networking with recognised reference centres abroad.

The role of NGOs, within the framework of which patients can exchange experience, acquire information and form joint proposals for the improvement of care, is also of key importance. NGOs can greatly contribute to raising public awareness.

It is often difficult to recognise rare diseases. Diagnostic procedures can sometimes take several years. An opportunity to improve the situation lies in the **organisation of education** for health care and other expert workers who treat rare diseases. Educational content must also be made available to patients, their families and the general public.

Activities	<p>To establish a national centre for rare diseases. To establish educational modules and educational material for various target groups (health care and other expert workers, patients, public). To inform on rare diseases (organisation of a rare disease day).</p>
Purpose	<p>To improve access to information on rare diseases for patients, health care and other expert workers and public.</p>

Monitoring and reporting

All stakeholders that are recognised in the process of forming the work plan will also participate in further implementation, monitoring and adjustment within the framework of a coordination group. The work plan will be implemented on the basis of annual action plans which will define certain specific goals of individual activities, schedules for their

implementation and indicators for monitoring the efficiency of implementing anticipated activities. Those undertaking individual activities per the action plan will report regularly to the coordination group. The coordination group will prepare an annual report on the implementation of the work plan and annual action plan.

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