Innovative approach towards the national organization of rare disease management – Slovenian National plan

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"The Council Recommendation on European Action in the field of Rare Diseases", adopted by the EU Council in June 2009, recommends Member States to adopt national plans or strategies for rare diseases before end of 2013.

Definition: A NP/NS is as a set of integrated and comprehensive health and social policy actions for rare diseases (with a previous analysis of needs and resources), to be developed and implemented at national level, and characterized by identified objectives to be achieved within a specified timeframe.
"The Council Recommendation on European Action in the field of Rare Diseases", adopted by the EU Council in June 2009, recommends Member States to adopt national plans or strategies for rare diseases before end of 2013.

1. Plans and Strategies in the Field of Rare Diseases
2. Adequate Definition, Codification and Inventorying of Rare Diseases
3. Research on Rare Diseases
4. Centres of Expertise and European Reference Networks for Rare Diseases
5. Gathering the Expertise on Rare Diseases at European Level
6. Empowerment of Patients’ Organisations
7. Sustainability
Short history

EUROPLAN background

"The Council Recommendation on European Action in the field of Rare Diseases", adopted by the EU Council in June 2009, recommends Member States to adopt national plans or strategies for rare diseases before end of 2015.

EUROPLAN recommendations will provide tools to Member States for developing a plan or strategy, linking with a common framework at the European level.

This "double level" approach is respectful of national decisions but is expected to ensure a coherent and consistent progress in EU rare diseases care.


( Financed within the Programme of Community action in the field of Public Health )
The main goal is to provide National Health Authorities with supporting tools for the development and implementation of National Plans and Strategies for rare diseases following the European Council Recommendation.
Short history

- Built to provide integrated, comprehensive, long-term strategy taking into account the principles and guidelines of the key policy documents.

- Aim 1: To produce a complete, coherent and feasible operational proposal for NP/NS
Short history

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- Aim 1: To produce a complete, coherent and feasible operational proposal for NP/NS

The first round of establishing national plans was a joint effort!
Short history

EUROPLAN: A Project to Support the Development of National Plans on Rare Diseases in Europe

Fig. 1. Conformity among priorities of the Council Recommendation of 8 June 2009 on an action in the field of rare diseases (2009/c 151/02) and existing National Plans/Strategies in EU Countries in 2012.
National activities

NAČRT DELA NA PODROČJU REDKIH BOLEZNI
V REPUBLIKI SLOVENIJI 2021-2030

Ljubljana, november 2021
# National activities

## Personalizirana - inovativna medicina za RB

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<tr>
<th>Diagnostika za preprečevanje in zdravljenje</th>
<th>Vrhunska zdravstvena oskrba</th>
<th>Integracija RB v družbo</th>
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<td>Genomsko prediagnostično in neonatni presežek</td>
<td>Deljenje informacij za skupen napredek</td>
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<td>Presijanje za RB z začetkom v odrasli dobi v primarnem zdravstvu</td>
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<td>Nacionalne mreže v ERM</td>
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<td>Dostopnost do zdravilni sirot</td>
<td>Nova zdravljavnica in namenjena gensko zdravljenje RB</td>
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## Integracija RB v družbo

- Deljenje informacij za skupen napredek
- Integrirana socialna oskrba in izobraževanje

Ljubljana, november 2021
# National activities

## Pilar 1 - Personalized - innovative medicine for RD

**Objective 1.** Diagnostics for prevention and treatment

## Pilar 2 - Integration of RD into society

**Objective 2.** Superb health care

**Objective 3.** Sharing information for common progress

**Objective 4.** Integrated social care and education

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| Center za nediagnosticirane RB | Genetsko prepoznavanje in neonatalno presenjanje | Presenjanje za RB z dece, ki je v odrašci dobi začetek v primarnem zdravstvu | Multidisiplinarni tim za obravnavo RB in za nemoten prehod v odrašca dobo | Nacionalne mreže v RB | Nacionalni koordinacijski center za RB | Dostopnost do zdravil sirot | Nova zdravljenje in napredno geniko zdravljenje RB | Integrirana nacionalna baza za RB | Spodbujanje povezovanja bolnikov | Glas bolnikov za oblikovanje politik | Izobraževanje za sodelovanje |
Objective 1. Diagnostics for prevention and treatment

Activity 1. Center for undiagnosed rare diseases

- A significant proportion of RD patients remains undiagnosed
(Despite the fact that we have introduced advanced genomic technologies for
diagnosis of genetic diseases in our health care system)
Objective 1. Diagnostics for prevention and treatment

Activity 1. Center for undiagnosed rare diseases

- A significant proportion of RD patients remains undiagnosed (Despite the fact that we have introduced advanced genomic technologies for diagnosis of genetic diseases in our health care system)

Activity 2. Genomic prenatal and neonatal screening

- aim: to develop modern options for primary and secondary prevention of genetic diseases

- project is planned in 2024 – to assess the possibility of using genomic screening tests, both from a professional as well as from social and ethical points of view.
Objective 1. Diagnostics for prevention and treatment

Activity 3. Adult-onset rare disease screening in primary healthcare

- aim: to enable the early detection of patients and people with a high risk of development RD in adulthood

- secondary aim: to empower the healthy population and primary healthcare to identify genetic risks.

- ongoing project in the primary healthcare: assessing the possibility of a systematic use of family history tool at the primary level of health care for discovering important genetic predisposition for genetic diseases.
Objective 2. Superb health care

Activity 4. Multidisciplinary teams for RD patient management

- RD patients are mostly treated within the speciality clinics, according to the main symptoms and signs that appear in a certain disease
- many RDs affect different organ systems

- multidisciplinary teams should be organized in such a way that patients are seen by different medical professionals within one team evaluation

- team: team coordinator, 2 MD, specialized for RD, 2 registered nurses, a clinical phychologist, social worker and nutritionist
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Activity 5. National networks in European reference networks

ERNs in Slovenia

<table>
<thead>
<tr>
<th>Full membership</th>
<th>Associated membership</th>
<th>National hub</th>
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<td>ERN-PND</td>
<td>ERN-IHACO</td>
<td>ERN-BOND</td>
</tr>
<tr>
<td>ERN-EURO-NMD</td>
<td>ERN-GUARD-HEART</td>
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<td>MetaERN</td>
<td>ERN-CRANO</td>
<td>ERN-EuroBloodNet</td>
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<td>ERN-NeuONET</td>
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<td>ERN-RTA</td>
<td>ERN-LUNG</td>
<td>ERN-ITHACO</td>
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<tr>
<td>Endo-ERN</td>
<td>ERN-RARE-LIVER</td>
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<tr>
<td>ERN-PeadICAN</td>
<td>ERN-EyeCARE</td>
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<td>ERN-REN</td>
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<td>ERN-ERGOCAM</td>
<td>ERN-GERONTIS</td>
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University Medical Centre Ljubljana

Institute of Oncology, Ljubljana
Objective 2. Superb health care

Activity 6. National Coordination Center for Rare Diseases

- to enable the coordinated implementation of the national plan in the future, to identify new needs in the field of RD in Slovenia, to connect all institutions for RD and ensure the sharing of good practices and provision of common standards in patient care

- to be a direct link to the Ministry of Health, and to collaborate in the creation of new health policies AND to ensure a connection with ORPHANET

- Planned after 2025
Objective 2. Superb health care

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- Planned after 2025

Activity 7. Access to orphan drugs

Activity 8. New treatments for RD and advanced gene therapy for RD

- Pilot project planned in 2024 – to assess all aspects in the field AND joining consortium of all stakeholders and institutions being in the field of RD
Objective 3. Sharing information for common progress

Activity 9. Integrated national database for RD

- standardized, accessible and interoperable data sources, including comprehensive overview of the individual disease progression, epidemiology, to be linked with other databases, HIS
- enable international exchange of data, involvement in national and international clinical research
- financial indicators monitoring and optimization
- National Registry for non-malignant RD
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Activity 10. Encouraging the association of all RD patients
- some RD are so rare, that there are only 1 or 2 patients with the disease in Slovenia and it cannot be expected for them to establish their own association
- National patient alliance should provide assistance and information – continuous additional funding is planned in National Plan
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Objective 4. Integrated social care and education

Activity 11. Patients' voice for policy making

- aim 1: to provide an inclusive environment for the development of social policies and services, actively involving patients and their organizations in identifying the needs of patients and their families

- aim 2: to increase autonomy, improve the quality of life, education, employment and participation of RD patients in Slovenian society.

- collaboration between the National patient alliance and National coordination center for RD (Activity 6)

- Planned after 2025 and continuous after that
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- Planned after 2025 and continuous after that

Activity 12. Education for cooperation

- The aim is to provide innovative education for healthcare professionals, patients and the general public

- collaboration with the Natinal coordination center for RD (Activity 6)

- Planned after 2026 and continuous after that
National activities


Člani delovne skupine Ministarstva za zdravje:
- dr. Robert Medved, Ministarstvo za zdravje, vodja;
- mag. Bernarda Kociper, Ministarstvo za zdravje, članica in namestnica vodje;
- prof. dr. Borut Peterlin, Klinični inštitut za medicinsko genetiko, UKC Ljubljana, član;
- Eeva Murko, Nacionalni inštitut za javno zdravje, članica;
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- Alenka Marič Cevzar, Zavod za zdravstveno zavarovanje Slovenije, članica;
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- Tea Čemigoj Putinjak, Združenje za redke bolezni Slovenije, članica;
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- doc. dr. Blaž Korišnik, UKC Ljubljana, član;
- prof. dr. Robert Ekar, UKC Maribor, član;
- prof. dr. Nataša Marcčun Varda, UKC Maribor, članica;
- doc. dr. Mojca Žerjav Tanšek, UKC Ljubljana, članica.
Thank you

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