European Rare Diseases
Research Alliance

Resource on the State of the Art of Rare Disease Activities

2025 Report for Slovenia

National Mirror Group

Slovenia does not currently have a National Mirror Group (NMG). Slovenia's plan is to activate an existing group as their NMG and to increase membership to include all rare disease stakeholders. A proposal has been shared with Slovenia's state health council and they are now awaiting approval of the proposal.

The collection of data for this survey has been overseen via a data contributing committee. Contributors are listed at the end of this report.

Definition of a Rare Disease

Slovenia adopts the formal European Union (EU) definition of a rare disease (i.e. those with a prevalence of no more than 5 patients per 10,000 persons. This definition is laid down in Regulation EC no. 141/2000 on Orphan Medicinal Products, Directive 2011/24/EU on Cross Border Healthcare as well as in the Council Recommendation on an action in the field of rare diseases of 8 June 2009.). The National Plan espouses this definition and there are no instances when a different definition is used.

Status Quo of any National Plan or Strategy for Rare Disease

What is the status quo?

Slovenia has a National Work Plan for Rare Disease that is described as live and in date. The plan is timebound for the period of 2021-2030. Prior to the current plan, there was one previous edition of Slovenia's National Work Plan for Rare Disease. In addition to the work plans, which are prepared for a longer period (e.g. from 2011 to 2020; from 2021 to 2030), Slovenia has specific action plans, which are active for a shorter time period, but do relate to the work plan. The previous action plan was between 2022-2023. The next action plan is being consolidated and is in the final phase, to be published publicly soon.

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Elaboration and Adoption/How was the original plan elaborated?

Slovenia's National Work Plan for Rare Disease for 2021-2030 was presented to the Minister of Health on 4th January 2022 and was approved by the Health Council on 18th January 2022. The Register of Rare Non-Malignant Diseases was legally defined in 2018 but the National Work Plan, as a whole, was not enshrined in any laws or regulations.

There was a dedicated group in charge of overseeing the drafting and adoption of Slovenia's National Work Plan for Rare Disease. This group included the following stakeholder types:

- Patients/people with lived experience of rare condition
- Slovenia's National of Rare Disease Patient Organisations
- Health Ministry/Competent National Authority in charge of Health or Care
- Researchers/clinicians from rare disease centres
- ERN coordinators or representatives

The group consisted of 13 members and included two patient representatives (hemophilia patient and Spinal muscular atrophy (SMA) type II patient), six clinicians/researchers, two representatives of the Health Insurance Institute of Slovenia (Zavod za zdravstveno zavarovanje Slovenije), one representative of the National Institute of Public Health (research) and two representatives of the Ministry for health.

Is there funding for the Plan? How is it Implemented and/or Monitored or Evaluated?

There is a dedicated budget for Slovenia's National Work Plan for Rare Disease. Through the Action Plan for 2022 and 2023, more than €4 million per year was provided for the implementation of the Rare Diseases Work Plan (2021 to 2030).

The same group that oversaw the drafting/adoption of Slovenia's National Work Plan for Rare Disease was responsible for overseeing the implementation of the plan. The National Work Plan has been implemented on the basis of biennial Action Plans, which will set out the specific objectives of each activity, the timeframes for their implementation and the indicators for monitoring the success of the planned activities. The leader of each action in the Action Plan will report regularly to the Coordination Group. The Coordination Group prepare a biennial report on the implementation of the work plan and a biennial action plan. However, only one biennial plan was prepared for the duration of 2022-2023. This group is not currently functioning and does not meet. No formal monitoring or evaluation of the National Work Plan has taken place.

Research in National Plans or national research strategies relevant to rare disease

Slovenia's National Work Plan for Rare Disease does not specifically address rare disease research. The plan is orientated towards clinical work, diagnosis, infrastructure, support, communication and knowledge transfer. The plan does reference the European Joint Programme on Rare Disease (EJP RD). The plan also addresses the following topics related to rare disease research:

- Registries or registry catalogues for rare diseases
- Ontologies, codification or data standardisation

- Diagnostics research ('solving unknown conditions')

Rare Disease Research Programmes and Funding

There are no programmes or funding calls specifically for rare disease but rare disease projects are funded from general research programmes in Slovenia. In 2024, Slovenia allocated €549 million from the state budget to research and development activities. The agency in charge is the Slovenian Research and Innovation Agency. There has been no policy decision to allocate a portion of the national research budget specifically to rare disease research.

Rare Disease Registration and Biobanking

Slovenia has a national register for rare disease. The Register of Rare Non-Malignant Diseases was established by law in 2018, and then officially began in 2024. The register is run by the Paediatric Clinic of the University Medical Centre Ljubljana. The register is included in Slovenia's National Work Plan for rare disease. The register includes the ICD10 and Orphacode coding systems.

Slovenia also has rare disease specific registries. National disease specific registries are managed/lead via university medical centres or dedicated hospitals. Some of the national disease-specific registries are:

- National registry of patients with Fabry disease
- Register of patients with cystic fibrosis
- Register of persons with blood clotting disorders
- Register of patients with neuromuscular diseases
- Slovenian National Registry of Patients with Primary Immunodeficiency
- Cancer Registry

Slovenia does not have a specific biobank for Rare Disease biosamples.

Organisation of Rare Disease Care

Centres of Expertise

Slovenia has a policy in place for designating Centres of Expertise for rare disease, both at the cross-rare disease level and for specific rare diseases. The policy does not include specific criteria but designation is given to individual departments and clinics that have acquired tertiary status, which is well defined within Slovenia, and are willing to participate in European Reference Networks (ERN) with their expertise.

ERNs

There are 13 HCPs who are full members of ERNs, four HCPs who are associated with national centres, and six ERNs are covered by the Slovenian ERN HUB. For the latest details on participating HCPs, click here.

Newborn Screening

In Slovenia, newborn screening includes tests for: congenital hypothyroidism, 18 different congenital metabolic disorders, Spinal Muscular Atrophy (SMA) and 20 different severe congenital immune disorders. In 2022, as part of Slovenia's National Work Plan for Rare Disease, the Ministry of Health allocated €1,926,868.24 per year for the introduction of an additional newborn screening program. In addition to the existing screening for phenylketonuria, hyperthyroidism, and a number of rare congenital metabolic diseases, SMA was added to the newborn screening programme, as well as severe congenital immune disorders, cystic fibrosis, and congenital adrenal hyperplasia. Between 45-50 diseases are covered within Slovenia's newborn screening programme.

Slovenia collaborates with other countries on newborn screening decisions via European Reference Networks (ERNs).

Diagnostics

Three laboratories within University Medical Centres carry out genetic testing. The cost of tests are completely covered by the general health insurance in Slovenia. Within Slovenia, there is a Centre for Undiagnosed Rare Diseases established within UMC Ljubljana. There is a policy in place in Slovenia to ensure genetic counselling is provided for patients with a suspected or confirmed rare disease.

National Alliances of Rare Disease Patient Organisations

Združenje za redke bolezni Slovenije (Rare Diseases Association of Slovenia) is Slovenia's National Alliance of rare disease patient organisations. Further information can be found here. Slovenia's National Alliance is involved in in setting the strategic direction for rare disease research. Rare Diseases Association of Slovenia brings together patient organisations and facilitates dialogue between patients, families, clinicians, and researchers. This allows the association to represent patient needs, which is crucial for directing research towards relevant priorities. The Rare Diseases Association of Slovenia is involved in the preparation of the National Work Plan for Rare Diseases (2021–2030). Through this participation, the association contributes to setting research priorities and monitoring implementation.

Section for Rare Diseases within the Patient Organisations Association of Slovenia are collaborating with different research institutes and clinics (National Institute of Chemistry, specifically their newly established Centre for Gene and Cell Therapy Technologies (CTGCT)), with University Medical Centres (special focus to the Paediatric clinic at UMC Ljubljana and with the Centre for rare diseases at this clinic), and Jožef Stefan Institute. They are also a partner within the Horizon WIDERA (Widening Participation and Strengthening the European Research Area) project together with the National Institute of Chemistry. New research calls are currently being discussed and proposals are being prepared.

Slovenia's National Alliance does provide capacity building training for patient engagement in rare disease research. Rare Diseases Association of Slovenia co-organises the National rare diseases conference with the Ministry of health. The Patient Organisations Association of Slovenia organises webinars and capacity building events on topics including communication, consultation on the importance of PROMs and PREMs and the role of patients, consultation on public health strategy, and an online course on antimicrobial resistance.

The Patient Organisations Association of Slovenia does circulate surveys amongst members including their latest survey about artificial intelligence.

Information Resources for Rare Disease

National Orphanet Engagement

Slovenia does have an operational Orphanet team, which is hosted at the University Medical Centre Ljubljana. Personnel costs for the Orphanet team are partially covered by the host organisation and partially by the OD4RD (Orphanet) project.

Helplines

Slovenia does have a national helpline dedicated to rare disease, which is available for anyone to use. The helpline receives public funding only and is managed by the University Medical Centre Ljubljana. The website of the National Contact Point for Rare Diseases is a project of the Ministry of Health of the Republic of Slovenia. The goal is to connect institutions, experts, and patients with rare diseases and their families, while also giving patients and experts access to quality information on treating rare diseases in Slovenia. Further information about The National Contact Point for Rare Diseases is available here in Slovene and here in English.

Training and Education

Rare disease training activities are available in Slovenia. These training activities include the following topics:

- Diagnostics
- Rasing awareness of rare disease
- Standards and quality of genetics/genomics data in clinical practice and laboratories
- Registries
- Biobanks
- Clinical research

Currently in development is a Targeted Research Program for innovative education in the field of rare disease which will offer online educational courses. Higher education institutions in Slovenia do offer training about rare disease. For example, The University of Novo mesto, Faculty of Health Sciences, offers a study program for genetic nurses and genetic counsellors for advanced education after completing a first-level higher education degree in the field of nursing and healthcare.

Orphan Medicinal Products (OMPs)

The Health Insurance Institute of Slovenia finances all OMPs that are registered by the EMA and are available in the country.

Slovenia does have in place a compassionate use policy to help patients access medicinal products for rare conditions. The public body responsible for this policy is the Agency for Medicinal Products and Medical Devices of the Republic of Slovenia (JAZMP). Compassionate use of medicinal products in Slovenia is regulated by Article 41 of the Medicinal Products Act (Official Gazette of the Republic of Slovenia, No. 17/14 and amendments) and by European legislation (Article 83 of Regulation (EC) No. 726/2004).

Conditions for compassionate use:

1. The medicinal product must be undergoing clinical trials or the process of obtaining a marketing authorisation through a centralized procedure.
2. There must be a clinical need that cannot be met by medicinal products that have a valid marketing authorisation.
3. The manufacturer must be willing to supply the medicinal product for compassionate use.

An application for compassionate use authorisation may be submitted to the JAZMP by:

1. The applicant whose medicinal product is undergoing the centralized marketing authorization procedure.
2. The sponsor of a clinical trial, if the medicinal product is undergoing clinical trials for the purpose of obtaining a marketing authorisation through the centralised procedure.

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The Data Contributing Committee of Slovenia, which provided this 2025 data (correct as of the end of November 2025) in the context of the Resource on the State of the Art of Rare Disease Activities, is composed of the following individuals:

- Tanja Zdolsek Draksler, JSI representative/ERDERA partner
- Robert Medved, Ministry of Health of Slovenia
- Tea Černigoj Pušnjak, Secretary of the Rare diseases association of Slovenia
- Jurij Furst, Head of the Medicines Department at the Health Insurance Institute of Slovenia (i.e. Zavod za zdravstveno zavarovanje Slovenije) - conducts its business as a public institute, bound by statute to provide compulsory health insurance and compulsory insurance for long-term care
- Luca Lovrecic, National Orphanet Coordinator and clinical geneticist in the centre for undiagnosed rare diseases - UMC Ljubljana
- Lana Stavber, Center for rare diseases at UMC Ljubljana