

Resource on the State of the Art of Rare Disease Activities

2025 Report for Lithuania

National Mirror Group

Lithuania has a National Mirror Group (NMG), which was launched on 30th May 2025. The NMG is coordinated by Vilnius University Hospital Santaros klinikos. This NMG has overseen the collection of data from Lithuania via a data contributing committee. Contributors are listed at the end of this report.

Definition of a Rare Disease

Lithuania 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. However, there is a different definition when it comes to reimbursement for medicines. According to Lithuania's legal regulations, treatment with Orphan drugs is only reimbursed for conditions with a prevalence of less than 1 per 200 000 inhabitants. This means that not every patient with a rare disease is able to access certain treatments.

Status Quo of any National Plan or Strategy for Rare Disease

What is the status quo?

Lithuania has a National Plan for Rare Disease that is described as being a live and open-ended plan. The plan was adopted through the Order of the Ministry of Health in 2012 and is Lithuania's first National Plan for Rare Disease. Lithuania's second National Plan for Rare Disease is currently in development.

Elaboration and Adoption/How was the original plan elaborated?

A dedicated, multi-stakeholder, committee was put in place to oversee the drafting and adoption of Lithuania's National Plan for Rare Disease. This group included the following stakeholder types:

- Patients/people with lived experience of rare condition
- Lithuania's National Alliance of Rare Disease Patient Organisations

- Health Ministry/Competent National Authority in charge of Health or Care
- Researchers/clinicians from rare disease centres

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

There is no dedicated funding for Lithuania's National Plan itself but there are dedicated funds for specific actions within the plan.

A different group was responsible for overseeing the implementation of Lithuania's National Plan for Rare Disease. This group included the following stakeholder types:

- Patients/people with lived experience of rare condition
- Health Ministry/Competent National Authority in charge of Health or Care
- Research funders
- Researchers/clinicians from rare disease centres
- ERN coordinators or representatives

This group was set-up specifically to address rare disease, not just for the purpose of implementing the National Plan for Rare Disease. The group is still functioning and meets regularly. This group carried out formal monitoring of the National Plan. Annual reporting to the Rare Disease Policy Group is carried out.

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

Lithuania's National Plan for Rare Disease states that rare disease research is important. However, the plan was not signed by the Ministry of Science and Education and does not include any specific responsibilities with regards to rare disease research.

Rare Disease Research Programmes and Funding

There are no specific rare disease programmes/funding calls in Lithuania but rare disease projects are funded from the general research programmes.

Rare Disease Registration and Biobanking

Lithuania has neither national nor regional registries for rare disease. There are currently no plans to develop a national registry for rare disease. However, ORPHA coding was introduced into the national e-health system. There are rare disease specific registries within Lithuania and these are hosted in two hospitals that have rare disease Centres of Expertise.

National/regional biobanks exist in Lithuania but not specifically for the collection of rare disease biosamples. The metadata for biobanks in Lithuania are available to view [here](#).

Organisation of Rare Disease Care

Centres of Expertise

Rare Disease Centres of Expertise are hosted in two university hospitals in Lithuania: Vilnius University Hospital Santaros klinikos and Lithuanian University of Health Sciences hospital Kauno klinikos. Rare Disease Centres of Expertise are established according to the decisions of these hospitals. However, criteria requirements are provided by the Ministry of Health.

ERNs

Lithuania is involved in multiple European Reference Networks. For the latest details on participating HCPs, click [here](#).

Newborn Screening

12 conditions are included within Lithuania's Newborn Screening programme. Expansion of screening in Lithuania is based on the general procedure for inclusion of any service into the health system, therefore, the criteria for inclusion is HTA-based.

Diagnostics

In Lithuania, reimbursement of genetic tests is based on the Orders of the Ministry of Health. Reimbursement is applied for a wide range of testing from single gene testing to Whole Exome Sequencing.

There are no agreements in place to facilitate cross border genetic testing for rare disease with any specific country. However, cross-border healthcare services are instituted and reimbursed according to the Order of the Ministry of Health. There is a committee that decides on the application of cross-border services (including testing abroad) and reimbursement is implemented according to procedures.

Lithuania has implemented approaches to address undiagnosed patients/people with currently undiagnosable conditions including reimbursing the reanalysis of next-generation sequencing data. Lithuania is involved in the diagnostics research workstream within ERDERA.

There is a policy in place to ensure national providers provide genetic counselling for patients with a suspected or confirmed rare disease. However, there are no general genetic counsellors, so genetic counselling services are provided by clinical geneticists within a specific speciality.

National Alliances of Rare Disease Patient Organisations

Nacionalinis retu ligu aljansas is Lithuania's National Alliance of Rare Disease Patient Organisations. Further information can be found [here](#). The National Alliance is involved in Lithuania's National Mirror Group (NMG) and is involved in setting the strategic direction for rare disease research.

Information Resources for Rare Disease

National Orphanet Engagement

There is an operational national Orphanet team in Lithuania hosted by Vilnius University Hospital Santaros klinikos. The team is funded by the hosting organisation and from funding from the OD4RD2 grant.

Helplines

There is no national helpline/information portal dedicated to rare disease in Lithuania.

Training and Education

There are rare disease training activities available in Lithuania. Training activities include the following topics:

- Diagnostics
- Raising awareness of rare disease
- Data management
- Standards and quality of genetics/genomics data in clinical practice and laboratories

Rare disease education is mostly provided by two medical universities: Vilnius University, Faculty of Medicine, and Lithuanian University of Health Sciences. Topics relating to rare disease are included in pre-graduate courses for medical and nursing students and there are specialised courses for residents in training, as well as continuous medical education courses. In addition, educational conferences that include rare disease are regularly organised and provide CME credits.

Orphan Medicinal Products (OMPs)

OMPs are reimbursed in Lithuania according to the procedure described in the Order of the Ministry of Health for very rare diseases and conditions with a prevalence of less than 1/ 200 000 inhabitants only. For every patient, a physician must complete required forms and apply for reimbursement to a committee. Additionally, there is a short list of OMPs that would be reimbursed, also based on the decision of the committee for every patient.



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The Data Contributing Committee of Lithuania, 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:

- Birute Tumiene, Vilnius University Hospital Santaros klinikos, a member of a Rare Disease Policy group under Ministry of Health, in consultation with other members.