European Rare Diseases
Research Alliance

Resource on the State of the Art of Rare Disease Activities

2025 Report for Luxembourg

National Mirror Group

Luxembourg does not currently have a National Mirror Group (NMG) in place. There are plans to launch an NMG in early 2026. A data contributing committee has overseen the collection of data for Luxembourg for this report. Contributors are listed at the end of this report.

Definition of a Rare Disease

Luxembourg 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?

Luxembourg currently has a live, in-date, time-bound, National Plan for rare disease. The plan covers the period between 2025-2029. Previously, there was a National Plan for Rare Disease between 2018-2023.

Elaboration and Adoption/How was the original plan elaborated?

Luxembourg's second National Plan for Rare Disease was elaborated in 2024, following the same strategy as their first plan, and was approved by Luxembourg's Ministry of Health and Social Security. To develop the second National Plan, a dedicated steering committee was set up by Luxembourg's Directorate of Health. The committee included key stakeholders involved in the rare disease field in Luxembourg, many of whom were involved in the development of the first plan. Other governmental institutions were also involved in this committee including the Ministry of Health and Social Security, the Ministry of Family affairs, and the Ministry of Higher Education and Research. Additionally, the following stakeholder types were included within the steering committee:

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- Patients/people with lived experience of rare condition
- Luxembourg's National Alliance of rare disease patient organisations
- Health Ministry/Competent National Authority in charge of Health or Care
- Research Ministry/ Competent National Authority in charge of Research
- Social or Welfare Ministry/ Competent National Authority in charge of social affairs
- Researchers/clinicians from rare disease centres
- European Reference Network (ERN) coordinators or representatives

The objectives of the committee were to define the overall strategy of the second National Plan. The second plan is intended to continue the efforts of the first plan, whilst considering the key recommendations from the final evaluation of the first National Plan. Luxembourg's second National Plan for Rare Disease was first submitted to the Directorate of Health, then to the Ministry of Health and Social Security and was launched in September 2025.

Luxembourg's National Plan is divided into five main themes: coordination of the national plan, access to diagnosis of rare diseases, healthcare pathways in rare diseases, awareness and training, and promoting innovation, digitalisation and research in rare diseases. In order to develop the objectives and goals for each of these categories, the following working groups were formed:

- Coordination of the national plan
- Diagnostic and screening of rare disease
- Access to medications and innovative treatments
- Healthcare pathways and continuity of care in rare diseases
- Socio-administrative support
- Educational and professional pathways of patients
- Communication, sensibilization and training programs
- Digitalization of health data, codification and registry
- Research in rare diseases

Each group gathered the relevant national stakeholders concerning the specific topics. These groups include representatives from Luxembourg's National Alliance of Rare Disease Patient Organisations, the Luxembourg National Centre of Genetics (National Health Laboratory), ERNs, Hospitals, the Directorate of Health, the university of Luxembourg, the Agence eSanté, the National Health Fund, the National Employment Agency, the Ministry of Education, Children and Youth, and the Ministry of Research and Higher Education.

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

A dedicated budget exists for Luxembourg's National Plan for Rare Disease. A yearly budget is proposed, approved and made available to carry on pilot projects originating from the National Plan for Rare Disease. The amount/year is not fixed and depends on the state of the art of pilot projects.

The National Steering Committee (called the CNMR) was specifically set-up to implement and oversee Luxembourg's first National Plan for Rare Disease. The committee met regularly. The following stakeholder types were included within this group:

- Patients/people with lived experience of rare condition

- Luxembourg's National Alliance of rare disease patient organisations
- Health Ministry/Competent National Authority in charge of Health or Care
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- Social or Welfare Ministry/ Competent National Authority in charge of social affairs
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A committee (CNMR2) for the second National Plan for Rare Disease is currently being set-up.

Formal and comprehensive evaluation took place for Luxembourg's first National Plan for Rare Disease. In Luxembourg, each national plan goes through a half-way through and a final evaluation. In the case of the first National Plan for Rare Disease, two evaluations took place, involving international experts in the field of rare disease. For the final evaluation, a team of experts including patient alliances, medical/genetics and European actions were involved. A detailed report was produced and published, and their recommendations are included in the second National Plan for Rare Disease. A similar process for evaluation has been included in the second plan.

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

Luxembourg's current National Plan for Rare Disease does address rare disease research. Research objectives are included within the plan but the responsibility of implementing those objectives is with the Ministry of Higher Education and Research. Research related activities within the national plan are not financially supported. Luxembourg's Directorate of Health funds the National Plan, but not specifically actions relating to research. A collaboration with the Ministry of Higher Education and Research will need to be put in place in order to implement and fund research actions.

Luxembourg's National Plan for Rare Disease includes the following topics relating to research:

- National RD research investment, opportunities and funding calls
- Registries or registry catalogues for rare diseases
- Ontologies, codification or data standardisation

Additionally, the National Plan promotes alignment with ERDERA, EJPRD and JARDIN.

All national plans developed in Luxembourg, including the National Plan for Rare Disease, are under the supervision of a National Plans Coordination Service at the Directorate of Health, which aims to harmonise processes and create synergies between common actions.

Luxembourg collaborates with other countries. In particular, due to its geographical position, there are collaborations with France, Belgium and Germany. However, the National Plan does not deliver any core actions relating to collaboration with other countries.

Select Achievements of the National Plan

Below are five key achievements resulting from Luxembourg's National Plan for Rare Disease:

1. The first census of rare diseases in Luxembourg

2. Set-up of a national service of information for patients, families and professionals called Infoline Maladies Rares Luxembourg
3. Set-up of a National Hub within the framework of the European Reference Network (ERN)
4. Extension of the neonatal screening programme, including SMA and other metabolic diseases
5. The elaboration and publication of a booklet on national resources, providing information on medical, psycho-social and administrative tools (Guide d'orientation luxembourgeois pour les personnes vivant avec une MR).

Rare Disease Research Programmes and Funding

There are no specific funding calls for rare disease research in Luxembourg, but specific rare disease projects are funded within general research programmes.

Rare Disease Registration and Biobanking

Luxembourg has neither national nor regional rare disease registries. The creation of a national rare disease registry is included within Luxembourg's current National Plan for Rare Disease. The Directorate of Health and the Ministry of Health and Social Security are working on establishing the legal framework, to allow the set-up and implementation of a national registry for rare disease.

There are biobanks within Luxembourg but not specifically for rare disease biosamples.

Organisation of Rare Disease Care

Centres of Expertise

There is no national policy in place in Luxembourg for designating centres for expertise, although this is currently under discussion. Luxembourg has opted for a national hub for rare diseases, recognised as an affiliated partner of ERNs. This hub is centralised within the Centre Hospitalier de Luxembourg. A medical and social coordination team consisting of a doctor and a social coordinator acts as an intermediary between the hub and patients. Patients and GPs can contact the hub directly or the medical and social coordination team via a helpline (Infoline Maladies Rares Luxembourg).

ERNs

Luxembourg is involved in multiple European Reference Networks. For the latest details on participating HCPs, click here.

Newborn Screening

Luxembourg has a national Newborn Screening program which includes six rare diseases: Phenylketonuria (PKU), Congenital Hypothyroidism, Congenital Adrenal Hyperplasia, MCAD deficiency, Cystic Fibrosis, and Spinal Muscular Atrophy (SMA). These screenings are typically performed on the third day of life. Severe combined immunodeficiency (SCID) will soon be added to the screening panel. In addition to these six conditions, clinical screenings are also carried out including for severe congenital heart diseases and a neonatal hearing test is carried out to detect early auditory impairments.

The national newborn screening program is coordinated by Luxembourg's Directorate of Health of the Ministry of Health and Social Security in collaboration with the Laboratoire National de Santé (LNS). Established in 1968, the program is regularly updated to reflect medical advancements. Decisions to add new conditions to the screening panel are made by the Directorate of Health, based on scientific evidence and public health relevance. The process for considering a new condition includes:

- Scientific and clinical evaluation by experts
- Alignment with the framework of the National Rare Diseases Plan
- Collaboration between the Ministry of Health and Social Security, LNS, and other stakeholders such as patient associations and healthcare professionals

The selection of conditions follows internationally recognised criteria, including the Wilson and Jungner principles published by the World Health Organisation. The criteria consider factors such as disease severity, the availability of effective treatment, and the feasibility of early detection. Discussions are currently underway with national stakeholders regarding the development of a neonatal genetic screening pilot project.

Luxembourg is not involved in any international initiatives concerning newborn screening but does consider data and evidence from neighbouring countries in newborn screening decision making.

Diagnostics

In Luxembourg, all genetic diagnostics are either performed by the Luxembourg National Centre of Genetics (NCG) or sub-contracted by the NCG to competent, cooperating, laboratories abroad. All genetic tests are reimbursed by public health insurance.

The NCG works closely with the ERN Hub in Centre Hospitalier de Luxembourg (CHL). It plays an active role in coordinating the ERN GENTURIS and, through the CHL Hub, has the opportunity to present cases via the CPMS platform to discuss them at a European level with experts in the field.

Luxembourg have set-up several initiatives, at a national level, to address undiagnosed patients/people with currently undiagnosable conditions, including the following:

- **Infoline Maladies Rares Luxembourg:** The Infoline is provided by ALAN Maladies Rares Luxembourg and was mandated by the Directorate of Health. It guides people affected by a rare disease to the appropriate services.
- **Weekly ALAN/Hub consensus meeting:** Cases of patients with a suspicion of a rare disease, who reached out to the national coordination unit, are discussed in weekly ALAN/Hub meetings. The national medical coordinator (CHL), the national psychosocial coordinator (ALAN) and their team of psychologists and social workers, as well as the administrative coordinator of the Hub participate in these meetings. The aim is to investigate patient files to respond to their requests, and, if necessary, offer them a transfer to a specific ERN coordinator. Thereby, access to international experts is given.
- **Monthly multidisciplinary genetics meeting:** Complex cases are discussed in monthly multidisciplinary genetics meetings hosted by the CHL. This initiative aims to reduce the diagnostic odyssey of patients and to offer a comprehensive, multidisciplinary approach to their care.

The long-term goal of the LNS is to offer Whole Genome Sequencing (WGS) to all children in intensive care and all children with a newly recognised neurologic disease. An additional goal is to offer WGS to a limited number of adult patients with rare diseases. The volume of these WGS that can be performed by the LNS depends on the staffing capacity, which is currently estimated between 50 to 100 yearly.

A policy is in place in Luxembourg to provide genetic counselling for patients with a suspected or confirmed rare disease.

National Alliances of Rare Disease Patient Organisations

Luxembourg has a National Alliance of rare disease patient organisations called ALAN asbl. Maladies Rares Luxembourg. Further information can be found here. The National Alliance provides webinars and other forms of learning and capacity-building for patient engagement in rare disease research.

To date, Luxembourg's National Alliance has not been involved in setting the strategic direction of rare disease research. However, the current national plan foresees rare disease research, and this will include consultation with the National Alliance.

Current barriers identified preventing patient engagement in research in Luxembourg include lack of time, awareness of patient organisations and a lack of a coordinated point of information on the different research projects in Luxembourg which seek patient involvement.

Information Resources for Rare Disease

National Orphanet Engagement

Luxembourg has an operational national Orphanet team hosted at the Directorate of Health. The team is funded entirely by the hosting organisation. In Luxembourg, one person participates to the

Management Board of Orphanet. Currently, Luxembourg has a limited contribution to the Orphanet database, particularly in terms of registries and centres of expertise, due to the low number of these resources at a national level.

Helplines

A national helpline is in place in Luxembourg dedicated to rare disease and is available for anyone to use. The INFOline Maladies Rares Luxembourg is run by the National Alliance ALAN asbl. Maladies Rares Luxembourg. It has a dedicated phone number and e-mail address, which is advertised on the National Alliance's website here. Trained social workers and psychologists (employees of ALAN asbl) answer enquiries received through the INFOline. The INFOline and related psychosocial support service has received the label of promising practice on mental health by the Best Practice Portal of the EU Commission in 2023. Further information is available here. The portal is funded through a mix of public and private funding.

Training and Education

There are currently no dedicated training or education activities dedicated to rare disease in Luxembourg.

Orphan Medicinal Products (OMPs)

Luxembourg uses the EMA criteria for designating OMPs. Once a medicine receives OMP designation by the EMA and receives EU-wide marketing authorisation, it is automatically valid in Luxembourg. It is not possible to provide an accurate number of OMPs available in Luxembourg, at this time. The full list of medicines that have a marketing authorisation in Luxembourg are available here and the list of commercialised and reimbursed medicines are available here.

Luxembourg does not currently have any Early Access Programmes or Expanded Access programmes. However, national legislation is currently under revision. The Luxembourg draft law no. 8491, which establishes the Agence Luxembourgeoise des Médicaments et Produits de Santé (ALMPS), includes provisions that allow the agency to manage compassionate use of medicines. These provisions are aligned with Article 5 of Directive 2001/83/EC, which permits the use of unauthorised medicines in exceptional circumstances. In the meantime, some orphan medicines may be available through hospital pharmacies on an individual-level named-patient basis.

Luxembourg is involved in formal initiatives with other countries to support access to medicines and therapies for rare diseases including being a member of Beneluxa since September 2015.

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The Data Contributing Committee of Luxembourg, 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:

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