European Rare Diseases
Research Alliance

Resource on the State of the Art of Rare Disease Activities

2025 Report for Sweden

National Mirror Group

Sweden has a National Mirror Group (NMG), which was launched on 15th October 2024. The NMG is coordinated by The Swedish Research Council. This NMG has overseen the collection of data from Sweden via a data contributing committee. Contributors are listed at the end of this report.

Definition of a Rare Disease

Sweden 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.). However, since 2020 the term used in the official definition of a rare disease in Sweden is 'rare health condition'. The translated official definition is as follows:

> Health condition refers to a disease, injury, disorder, or alteration in bodily function. Rare health conditions usually entail long-term consequences for living conditions and specific challenges resulting from their rarity. They often require special expertise and coordinated efforts from healthcare and other parts of society. As a guideline, a prevalence of fewer than 5 in 10,000 inhabitants may be regarded as low prevalence (See also Decision No.1295/1999/EC of the European Parliament and of the Council.)

Some in Sweden raise the need for a definition of ultra rare conditions.

Status Quo of any National Plan or Strategy for Rare Disease

What is the status quo?

Sweden does not have, nor has it ever had, a National Plan or Strategy for Rare Disease. The National Board of Health and Welfare has submitted a proposal for a national strategy for rare disease to the Swedish Government. The proposal is currently under consideration and has not yet been decided upon by the Swedish Government.

Co-funded by the European Union

ERDERA has received funding from the European Union's Horizon Europe research and innovation programme under grant agreement N°101156595.
Views and opinions expressed are those of the author(s) only and do not necessarily reflect those of the European Union or any other granting authority, who cannot be held responsible for them.

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

Whilst Sweden does not have a National Plan or strategy for rare disease, the proposed National Strategy for Rare Disease does address research. The proposed strategy promotes alignment with many European initiatives and includes the following topics related to rare disease research:

- National RD research investment, opportunities and funding calls
- International/transnational RD research investment, opportunities and funding calls
- Registries or registry catalogues for rare diseases
- Biobanks/biosample catalogues for rare diseases
- Ontologies, codification or data standardisation
- Diagnostics research ('solving unknown conditions')
- Basic research (e.g. cell lines, animal models etc)
- Clinical/Translational research
- Sociological (e.g. Quality-of-Life-related) or socio-economic research

---

Rare Disease Research Programmes and Funding

Sweden does not have dedicated national funding schemes specifically for Rare Disease. However, the Swedish Research Council and Vinnova (the Swedish Innovation Agency) participate in ERDERA Joint Transnational Calls (JTC). To date, these agencies alternate in funding JTCs. Vinnova has provided approximately €800, 000 in funding to JTCs in the European Joint Programme on Rare Disease (EJPRD) and ERDERA.

Rare Disease Registration and Biobanking

Sweden has a national registry for rare disease. The Rare Disease registry is located and governed in the Skåne region but it has a national steering committee. Further information is available here. The registry uses ICD10 coding.

There are rare disease specific registries in Sweden, examples include the Registry for metabolic diseases and the Registry for primary immunodeficiencies.

There is a national biobank in Sweden but not specifically or exclusively for rare disease biosamples. There are individual biobanks in Sweden that collect rare disease biosamples. The metadata for biobanks in Sweden is available here.

Organisation of Rare Disease Care

Centres of Expertise

There is no policy in place in Sweden to designate Centres of Expertise for Rare Disease.

ERNs

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

Newborn Screening

26 conditions are screened for in Sweden's newborn screening programme. Further information is available here.

The National Board of Health and Welfare uses a model for assessing, implementing, and monitoring national screening programmes. The model is designed to be applicable regardless of which condition the newborn screening programme aims to detect and address. Ethical, health economic, and medical aspects are all considered within the model. Feasibility, and thereby the conditions for equitable healthcare, are also important factors. Before the recommended screening can be initiated, the National Board of Health and Welfare must amend its regulations. The disease must be added to Section 1 of the Board's regulations (HSLF-FS 2019:13) on which diseases may be detected and diagnosed through tissue samples in the PKU biobank. The Board must also make an addition to its regulations (HSLF-FS 2019:12) on exemptions from the requirement for authorisation to perform genetic testing in public health examinations, in order for examinations concerning the specific disease not to require special authorisation. The model consists of three main components: 15 assessment criteria, a systematic working process, and a description of the organisation that plays a central role in the assessment of screening programmes. Further information can be found here.

Sweden is involved in international collaborations regarding newborn screening including screen4care (Akademiska hospital, Uppsala) and International Porphyria Network (Karolinska university hospital, Stockholm).

Diagnostics

In all regions in Sweden with university healthcare, regional Centres for Genomic Medicine (GMCs) have been established. These centres serve as the primary contact points for performing gene sequencing of patient samples, as well as for research collaborations and clinical studies. Each GMC is responsible for implementing precision medicine at the university hospital and for ensuring that hospitals and primary care facilities in neighbouring regions also benefit from the initiative. There are no specific provisions in place for reimbursement of tests, as all regions are autonomous.

Sweden participates in initiatives to address undiagnosed patients/people with currently undiagnosable conditions. Undiagnosed Diseases Network Sweden (UDN Sweden) is part of the Undiagnosed Diseases Network International (UDNI). UDN Sweden is a multicentre study aimed at achieving equitable national implementation of whole genome sequencing for children with undiagnosed rare syndromes involving congenital anomalies and/or intellectual disability. The purpose of the project is to prepare the healthcare system to meet the growing need for whole genome sequencing. In addition, the project aims to determine whether more patients can be diagnosed by combining different methods of modern genetic technology. The multidisciplinary team within UDN Sweden consists of experts in diagnostics with extensive experience in the clinical and genetic characterisation of patients with intellectual disabilities and/or congenital anomalies.

There is a policy in place in Sweden to ensure providers provide genetic counselling for patients with a suspected or confirmed rare disease.

National Alliances of Rare Disease Patient Organisations

Rare Disease Sweden is the National Alliance of Rare Disease patient organisations in Sweden. Further information can be found here. Rare Disease Sweden is involved in Sweden's National Mirror Group (NMG) for rare disease.

Information Resources for Rare Disease

National Orphanet Engagement

There is an operational Orphanet team within Sweden. The team is hosted and funded by the Karolinska University Hospital. Further information is provided here.

Helplines

There is a helpline in Sweden dedicated to rare disease that is available for anyone to use. The helpline receives a mixture of public and private funding. The helpline is managed by Rare Disease Sweden. The National Board of Health and Welfare through Ågrenska has an information centre for rare diseases. The Centres of Rare Diseases (CSD) at university hospitals also have helplines.

Training and Education

There are rare disease training activities within Sweden.

Orphan Medicinal Products (OMPs)

The exact number of orphan medicinal products available in Sweden varies, but by the end of 2022 there were a total of 140 medicines that at that time or previously had orphan designation and were available on the Swedish market. Several factors influence this number: some medicines have lost their orphan designation but remain in use, and there is also a difference between all authorised orphan medicines and those actually made available by pharmaceutical companies in Sweden.

Below is a list of early/expanded access programmes within Sweden:

1. **Compassionate Use Programme (CUP) – EU Framework**
   Sweden follows EU Regulation (EC) No 726/2004, Article 83, which allows patients access to medicines prior to marketing authorisation if: the medicine is intended for patients with a chronic or seriously debilitating disease, or a life-threatening condition, no satisfactory treatment is available, and the medicine is likely to receive marketing authorisation within a reasonable timeframe. In Sweden, pharmaceutical companies may apply to the Swedish Medical Products Agency (Läkemedelsverket) to establish a compassionate use programme. Examples include certain advanced therapies and oncology medicines that have been made available through this mechanism.
2. **Licence Procedure (Named Patient Use)**
   The most common route for early access in Sweden is the licence procedure. An individual physician applies to the Swedish Medical Products Agency for a licence for a specific patient (or a defined group of patients). The medicine must either be authorised in another country or have adequate documentation on quality and safety. At EU level, this mechanism is often referred to as named patient use. This is the practical pathway for early access in Sweden, particularly for rare diseases.
3. **Clinical Trials (including Open-Label Extension Studies)**
   Patients may also gain access to new medicines through participation in clinical trials or open-label extension studies following completion of a trial.

Sweden is involved in collaborations with other countries to support access to medicines and therapies for Rare Disease. FINOSE is a Nordic collaboration for the evaluation of medicines, which was launched in 2018 as part of an initiative of the HTA authorities in Finland, Norway, and Sweden. The collaboration was expanded to include Denmark in 2023 and Iceland in 2024. With the expansion of the FINOSE group, the collaboration changed its name to the Joint Nordic HTA Bodies (JNHB) in June 2024. The collaboration involves the authorities conducting joint evaluations of medicines. The work includes an assessment of the treatment effect of the new medicine compared with that of existing treatments, as well as a health economic evaluation. Decisions on pricing, reimbursement, and recommendations for use are made nationally in each country. By working together and sharing knowledge, they aim to produce high-quality assessment reports that provide a solid basis for national decision-making.

ERDERA
European Rare Diseases
Research Alliance

Disclaimer: The data collection activities which enabled this 2025 national report were supported by the ERDERA. ERDERA has received funding from the European Union's Horizon Europe research and innovation programme under grant agreement N°101156595. Views and opinions expressed are those of the author(s) only and do not necessarily reflect those of the European Union or any other granting authority, who cannot be held responsible for them, nor should this document be viewed as an official national 'position'.

Co-funded by
the European Union

The Data Contributing Committee of Sweden, 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:

- Louise Rügheimer, The Swedish Research Council
- Anna-Carin Christoffersson, Vinnova, the Swedish Innovation Agency
- Lena Lövqvist, The National Board of Health and Welfare