

Resource on the State of the Art of Rare Disease Activities

2025 Report for Estonia

National Mirror Group

Estonia has a National Mirror Group (NMG), which was launched on 1st June 2025. The NMG is coordinated by the University of Tartu. This NMG has overseen the collection of data from Estonia. Contributors are listed at the end of this report.

Definition of a Rare Disease

Estonia 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.).

Status Quo of any National Plan or Strategy for Rare Disease

What is the status quo?

Estonia did have a Rare Disease Development Plan but this plan is not described as live or in date. Further information is available [here](#). After the plan was published, no further action was taken. The plan was not enshrined in law or any regulations. There are currently no plans to renew or adopt a new plan.

Elaboration and Adoption/How was the original plan elaborated?

There was no group or body in charge of implementing/adopting the plan.

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

There were no funds or resources committed to the plan and there was no funding to deliver any specific actions. No formal evaluation or monitoring of the plan has ever taken place.

Rare Disease Research Programmes and Funding

Estonia does not have any specific programmes or funding calls reserved for rare disease but specific projects for rare disease are funded from general research programmes.

Rare Disease Registration and Biobanking

Estonia does have a national registry for rare disease, which is part of Estonia's Electronic Health Record system. There are specific rare disease/groups of rare disease registries within Estonia. These registries are carried out as part of research studies and must have the approval from the ethics committee of the University of Tartu to collect data. Estonia's registry has implemented ICD10 and Orpha coding. The registry collects the Common Data Elements for rare disease registries, recommended by the European Commission through the European Platform on Rare Disease Registration (EU RD Platform).

Estonia has both a national biobank for rare disease biosamples and individual rare disease specific biobanks. The biobanks are part of research projects.

Organisation of Rare Disease Care

Centres of Expertise;

There is an official, national, policy in place in Estonia for designating Centres of Expertise. The policy includes both Centres of Expertise at the cross-rare disease level and for specific rare diseases/group of rare diseases.

ERNs

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

Newborn Screening

In Estonia, 23 conditions are screened for as part of their national Newborn screening programme. Further information is available [here](#).

Diagnostics

A list of genetic tests carried out in Estonia is available to view [here](#). Tests are reimbursed by Estonia's Health Insurance Fund. There are policies in place to facilitate cross border genetic testing for rare diseases. An application to do this must be made through the Health Insurance Fund. There are no specific strategies or approaches to address undiagnosed patients/people with currently undiagnosable conditions in Estonia but researchers do carry out regular reanalysis. There is a policy in place to ensure that national providers provide genetic counselling for patients with a suspected or confirmed rare disease.

National Alliances of Rare Disease Patient Organisations

[EPIK \(Estonian Chamber of People with Disabilities\)](#) is the National Alliance for Rare Disease patient organisations in Estonia. EPIK is involved in Estonia's National Mirror group (NMG), as well as being involved in setting the strategic direction of rare disease research in Estonia.

Information Resources for Rare Disease

National Orphanet Engagement

There is an operational, national, Orphanet team in Estonia, hosted by the University of Tartu. The team is funded by an Orphanet grant.

Helplines

A national helpline/portal is in place, dedicated to rare disease in Estonia. Further information is available [here](#). The helpline/portal is available for anyone to use. The helpline/portal does not receive any funding.

Training and Education

There are specific rare disease training activities available in Estonia, particularly for raising awareness of rare disease. Conferences are held annually on Rare Disease Day and offer awareness raising training opportunities.

Orphan Medicinal Products (OMPs)

There are approximately 5-10 OMPs available in Estonia. Estonia does not have Early Access Programmes or Expanded Access programmes.



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The Data Contributing Committee of Estonia, 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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