European Rare Diseases
Research Alliance

Resource on the State of the Art of Rare Disease Activities
2025 report for Austria

National Mirror Group

Austria has a National Mirror Group (NMG), which was launched on 30th August 2024. The NMG is coordinated by St. Anna Kinderkrebsforschung GmbH (CCRI). This NMG has overseen the collection of data from Austria via a data contributing committee. Contributors are listed at the end of this report.

Definition of a Rare Disease

Austria 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 Diseases

What is the status quo?

Austria currently has an active, open-ended, National Plan. Austria's first National Plan for Rare Diseases was officially conceptualised for the period 2014-2018 but has now become an open-ended plan. It was initially designed as a strategy rather than a plan but it was extended by the beginning of 2019 and is now an open-ended plan that includes regular monitoring of implemented measures.

Elaboration and Adoption/How was the original plan elaborated?

The Austrian National Plan for Rare Diseases (NAP.se) was developed and written by the National Coordination Center for Rare Diseases, which was established by the Austrian Ministry of Health in April 2011. Two committees were established accompanying the activities of the Center, namely the

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Experts Committee on Rare Diseases, which includes representatives of a variety of stakeholders in the rare disease field in Austria, and the Strategic Platform, where delegates of the decision-makers in the Austrian health care and social system are represented. These two committees participated actively in the development of the NAP.se. Upon finishing the draft, the input of all relevant decision-makers was obtained, and the NAP.se was approved by and published on behalf of the Austrian Ministry of Health as well as the Ministry of Labour, Social Affairs and Consumer Protection, and the Ministry of Science, Research and Economy. However, the National Plan is not enshrined in law. Taken together, a broad acceptance of the plan among stakeholders in health care, social services and research with respect to rare diseases can be assumed. The following stakeholder groups were included in a dedicated group that were in charge of overseeing the drafting/adoption of Austria's National Plan:

- Patients/people with lived experience of rare condition
- Austria'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
- ERN coordinators or representatives

How is it Implemented and/or Monitored or Evaluated?

The same group that drafted/adopted Austria's National Plan is in charge of implementing/overseeing the National Plan. The group was specifically established for drafting and implementing Austria's National Plan for Rare Disease. The implementation/oversight group meets regularly.

Formal and comprehensive evaluation and monitoring of the National Plan takes place by another group. Regular monitoring of the National Plan is carried out by the Ministry of Health and the National Coordination Center for Rare Diseases, as well as regular reporting to the advisory bodies, which include all involved stakeholders. A formal evaluation took place in 2021 which resulted in the prioritisation of some measures to be implemented.

The Austrian National Plan for Rare Disease has a dedicated budget, in the sense that the drafting of the original National Plan was financially supported by the Ministry of Health. However, the implementation of the National Plan is carried out by different stakeholders within the health care sector, so the exact funding mechanisms are unknown. To work on a few selected and prioritised measures, the Federal Ministry of Labour, Social Affairs, Health and Consumer Protection provides financial support to the National Coordination Centre for Rare Diseases.

Rare Disease Research Programmes and Funding

The Austrian National Plan specifically addresses rare disease research and is one of 9 key areas included within the plan. The following topics related to rare disease research are specifically addressed:

- Registries or registry catalogues for rare diseases
- Ontologies, codification or data standardisation
- Diagnostics research ('solving unknown conditions')

Austria does have specific research programme/funding calls dedicated to rare diseases that are publicly funded. These funding calls are not regular but sporadic. The approximate value of national investment in rare disease funding calls over the past 5 years is €22 million (plus an additional estimated €25 million for rare cancers research). No general decision was made to allocate a portion of the national research budget specifically to rare disease research, but it was decided to dedicate fixed amounts (from the Austrian Science Fund (FWF)) to participate in calls and fund research projects within European initiatives such as ERA-Nets and European Joint Programs and Partnerships including ERDERA.

Rare Disease Registration and Biobanking

Currently, neither national nor regional Rare Disease registries exist in Austria at the pan-rare-disease-level. There are rare disease specific registries in Austria and they are clinician-led. Only larger registries dealing with large groups of both rare and non-rare diseases (like the Austrian Cancer Registry) are in public hands. Disease specific rare disease registries presently listed in Orphanet include: Cystic Fibrosis, Multiple System Atrophy (MSA), brain tumours, alpha 1-antitrypsin deficiency, mastocytosis, non-Hodgkin lymphoma (NHL) in children and adolescents, Acute Lymphoblastic Leukemia (ALL) in children and adolescents, Atypical Haemolytic Uraemic Syndrome (aHUS), alternating hemiplegia and rare epilepsies, T-cell lymphomas, inborn errors of metabolism, multiple myeloma, echinococcosis, Chronic myelogenous leukemia (CML), squamous cell carcinoma of the head and neck, acromegaly, epidermolysis bullosa, myelodysplastic syndromes, GIST, haemophilia, Upshaw-Schulman syndrome, histiocytic disorders, Behcet disease, cancer predisposition syndromes, congenital anomalies. Further information is available here. It is noted that the list of rare disease registries is not exhaustive. A Nationwide mapping process for clinical services for rare disease is currently underway and this information will be updated. Pro Rare Austria requested in 2024 a national register for Austria for Rare Disease. However, it is assumed that there is still no such registry.

Austria has biobanks that are not specifically for rare diseases but do collect some rare disease samples. For example, the Austrian node (BBMRI.at) of the BBMRI-ERIC (the European Biobanking Research Infrastructure) includes rare disease samples. According to the BBMRI-ERIC Directory Rare Disease collections/biobanks, there are currently 65 biobanks in Austria containing rare disease samples. The metadata is available to view here and is also connected to the ERDERA Virtual Platform. Some rare disease researchers have their own collections of rare disease samples.

Organisation of Rare Disease Care

Centres of Expertise

Austria has adopted both a national and regional approach for designating centres of expertise for rare disease. General information about centres of expertise in rare disease in Austria can be found on the website of the Ministry of Health here.

ERN participation

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

In addition, 1 of the 24 ERNs is coordinated by an Austrian centre:

PaedCan ERN	St. Anna Kinderspital & St. Anna Kinderkrebsforschung	Professor Dr. Ruth Ladenstein

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Newborn Screening

30 conditions are screened for in Austria's Newborn Screening programme. Further information is provided here. Austria's newborn screening program was established in the 1960s by the Ministry of Health and the Ministry of Science and Research. The service is located at the Department of Paediatrics and Adolescent Medicine of the Medical University of Vienna. Newborn screening is part of the 'Parent-Child-Pass' examination program. Testing is free of charge, and virtually all newborns (about 80, 000 per year) are tested for an array of inherited metabolic and endocrine diseases. About 100 children are tested positive per year. Additionally, about 50 are detected with a maternally transferred Vitamin B12 deficiency. The Austrian Newborn Screening is financed by the Medical University of Vienna via the Ministry of Science and Research. The conditions included in the Newborn Screening programme are:

- *Hormonal (endocrine) disorders*
    - Congenital adrenal hyperplasia
    - Congenital hypothyroidism
- *Amino acidopathies and urea cycle disorders*:
    - Phenylketonuria/Hyperphenylalaninemia
    - Tyrosinemia type 1
    - Maple syrup urine disease
    - Arginosuccinic aciduria
    - Homocystinuria
    - MAT I/III

  - Citrullinemia

- *Organic acidopathies*:
  - Methylmalonic aciduria
  - Propionic acidemia
  - Isovaleric acidemia
  - Glutaric aciduria type 1
  - Glutaric aciduria type 2 / Multiple acyl-CoA dehydrogenase deficiency

- *Fatty acid oxidation disorders (ß-Oxidation):*
  - Medium-Chain-Acyl-CoA dehydrogenase deficiency
  - Very-Long-Chain-Acyl-CoA dehydrogenase deficiency
  - Long-Chain-Acyl-CoA dehydrogenase deficiency
  - Mitochondrial trifunctional protein deficiency

- *Disorders of the carnitine cycle:*
  - Carnitine palmitoyl transferase 1A deficiency
  - Carnitine palmitoyl transferase II deficiency
  - Carnitine uptake deficiency
  - Carnitine-acylcarnitine translocase deficiency

- *Disorders of Cobalamin Metabolism*
  - Remethylation Disorders
  - Cobalamin Deficiencies
  - Vitamin B12 Deficiency

- Biotinidase deficiency
- Galaktosemia
- Cystic fibrosis
- Spinal Muscular Atrophy
- Primary Immune Defects

The respective pediatric working groups for different specialties of the Austrian Society of Pediatrics and Adolescent Medicine are required to review guidelines and literature to recommend the implementation or termination of screening for specific diseases in a written consensus. Technical and personnel issues are assessed by the technical and medical directors of the Newborn Screening programme, and the budget is approved by the vice rector for financial affairs of the Medical University of Vienna. Before implementation, there is typically a one-year assessment period to prove feasibility as well accurate qualitative parameters of the screening.

Generally, decisions around newborn screening are based on international experiences and published data in an overall view on the situation, demands and possibilities in Austria. A direct collaboration exists between the Austrian and the German Newborn Screening programme since the technical and

medical leaders of the Austrian Screening are members of the German Society for Neonatal Screening. Additionally, there exist close exchange with other newborn screening teams of European countries (e.g. Switzerland, Hungary, Slovenia, Greece). However, a direct or formal impact from external institutions concerning the Austrian screening programme does not exist.

Diagnostics

The Genetic Engineering Act (Gentechnikgesetz, GTG) provides the legal framework for genetic analyses in Austria including the provision of genetic counselling. The landscape of human genetic testing in Austria is very heterogeneous. On the one hand, there are a total of six Centers for Medical Genetics: one each in Innsbruck, Salzburg, Linz, and Graz, as well as two in Vienna. Their requirements are defined in the Quality Standard for Genetic Counselling and Diagnostics from 2015. These centers have contracts with the statutory health insurance for analyses and genetic counselling. They must be headed by a specialist in medical genetics and hold authorization to provide specialist medical training. In addition, numerous pathology and clinical chemistry laboratories, as well as specialized laboratories affiliated with departments such as neurology, internal medicine, and gynaecology, also offer human genetic diagnostics. Furthermore, private laboratories provide such services, billing patients or referring institutions directly.

A list/registry of laboratories providing specific genetic tests is available. The 'Genanalyseregister' provides a list of all approved laboratories which may provide human genetic analyses. Some laboratories list the parameters they offer in detail, but this is not mandatory. Provisions are in place for reimbursement of some tests. For predictive BRCA testing, official criteria must be met, and the sample must be sent to designated laboratories under specific contracts. For other indications, reimbursement depends on the laboratory performing the analysis. If the laboratory has a contract with insurance providers, costs are usually covered when there is a clinical indication. In some cases, however, laboratories must first submit a request for cost coverage to the insurer. For predictive testing, certain laboratories also require prior counselling by a specialist in human genetics. If patients are hospitalised, the costs are covered by the hospital.

Austria does not have specific bilateral agreements with individual countries concerning genetic testing. The Austrian National Action Plan for Rare Diseases (NAP.se) does emphasise the importance of coordinating clinical care through the establishment of specialised centres for rare diseases, known as centres of expertise, and their networking within Austria as well as with the relevant European Reference Networks (ERNs). This networking facilitates the exchange of knowledge and experience at the international level and promotes the use of international expertise in the diagnosis and treatment of rare diseases.

Austria has developed a comprehensive strategy to address rare and undiagnosed diseases through its National Plan for Rare Disease (NAP.se). However, in Austria, data on rare diseases are not collected centrally but are documented through locally maintained registries or via reporting to international registries by centres of expertise and specialized centres. Austria actively participates in the Undiagnosed Diseases Network International (UDNI). However, there is no national plan or strategy for the follow-up and re-evaluation of undiagnosed cases.

National Alliances of Rare Disease Patient Organisations

Pro Rare Austria is Austria's National Alliance of rare disease patient organisations. Pro Rare Austria is not currently a member of Austria's NMG. Pro Rare Austria is actively engaged in political and strategic work to improve the framework conditions for research on rare diseases. Examples include actively participating in health-policy consultations, collaborating with the Federal Ministry of Social Affairs, Health, Care and Consumer Protection, and contributing patient perspectives to national strategies, such as those on research funding and the implementation of the European Action Plan for Rare Diseases. However, they currently have no direct involvement in national research committees.

As an umbrella organization, Pro Rare Austria does not currently offer its own training programs or webinars specifically on patient involvement in research. However, it does support and share information about training and exchanges opportunities provided by its member organisations.

Several structural and legal barriers have been identified by Pro Rare Austria that impede or limit patient involvement. These include, among others, the lack of a legal status for patient organisations within the healthcare system, as outlined in the legal opinion commissioned by Pro Rare Austria, as well as insufficient official recognition and structural integration of patient representatives in research processes and political decision-making structures.

Information Resources for Rare Disease

National Orphanet Engagement

There is currently an operational national Orphanet team in Austria, hosted by Medical University of Vienna. The Orphanet team is partly funded by an international grant - the EU4Health: OD4RD2 grant (EU4H-2022-DGA-IO1-IBA, Project 101110100). Austria's national Orphanet team encourages/ensures that national resources such as registries, expert centres etc. are conveyed to the central Orphanet database, to be included in the full directory. The inclusion criteria and procedure can be found on the national Orphanet page here.

Helplines

Austria has a National Helpline dedicated to rare disease. The helpline has been available since the foundation of the 'Forum Seltene Krankheiten' in 2011. The inquiries are screened by Prof. DDr.med. Johannes Zschocke and referred regionally or are discussed at the monthly Rare Disease board meetings and then distributed. The helpline is available for medical doctors and patients.

Training and Education

There are specific rare disease training activities available in Austria. Austrian Medical Association (Österr. Ärztekammer), ProRare (Patient Organization), EURORDIS, EU Projects (e.g. EJPRD) provide these trainings. They are aimed at medical staff, patients/families and students. These trainings are

partially free and are offered in English and German. Some Austrian higher education institutions offer courses about rare disease. However, there are no dedicated bachelor or master programmes focused on rare diseases, as these topics are typically integrated into general medical education. Instead, courses on rare diseases are more commonly found in specialised continuing education programs for physicians or within research centres at medical faculties, such as those at the Johannes Kepler University Linz, during medical conferences and in the context of EU Projects that provide certificates. For the general public and/or patients, training activities are organised by patient advocacy groups.

Here some examples of rare disease training activities offered in Austria:

- Zentrum Seltene Genetische Erkrankungen Salzburg offers, among other things, the promotion of education, advanced training, and continuing education in the field of rare diseases for students, nursing staff, and physicians at the university hospital, as well as for representatives of the regional healthcare system.
- Wiener Selbsthilfegruppen offers training activities on topics such as role distribution in self-help groups, digital communication, interview and media training, and the social and healthcare system in Austria.
- EURORDIS offers several online courses in the field of rare diseases, some of which are now also available in German. These courses were developed in collaboration with subject matter experts such as researchers and patient representatives and provide access to valuable resources and specialised training. The free courses cover a range of key topics and promote flexible and intuitive learning.
- Certificates awarded to medical staff in the context of Symposia and Conferences, such as DACH SE Tagung wurde für den Erwerb des Fortbildungsdiploms der Österreichischen Ärztekammer mit 11 Punkten approbiert (ID1012639).

There is a need for a comprehensive overview of ongoing activities in Austria. Alongside research centres, medical societies, patient advocacy, and EU projects etc., professional conferences are essential drivers in this area to share results from research that can be implemented in clinical care and can raise awareness. The national action plan (NAP.se) has been updated and most recently a publication was published putting forward recommendations for training activities for general practitioners[1].

Orphan Medicinal Products (OMPs)

No information was available in 2025, on the number of OMPs available in Austria.

[1] Eisenmann, Alexander; Kanitz, Elisabeth (2025): Vorschläge zur Umsetzung von Fort- und Weiterbildungsmaßnahmen zu seltenen Erkrankungen für Ärztinnen und Ärzte im niedergelassenen Bereich (Maßnahme 56 des NAP.se). Konzept. Gesundheit Österreich, Wien

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The Data Contributing Committee of Austria, 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 in Europe, is composed of the following individuals:

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