European Rare Diseases
Research Alliance

Resource on the State of the Art of Rare Disease Activities
2026 report for Poland

National Mirror Group

Poland is currently in the process of setting up a National Mirror Group (NMG). A data contributing committee has overseen the collection of data from Denmark. Contributors are listed at the end of this report.

Definition of a Rare Disease

Poland 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?

Poland has a National Plan for Rare Disease that is described as being live and time bound. The legal framework for the National Plan for Rare Disease is implemented by a resolution of the Council of Ministers, in accordance with the statutory legislative process. The current plan is for 2026 and there have been two previous editions of the National plan for rare disease in Poland, covering the periods 2021-2023 and for 2024-2025. The Rare Diseases Plan for 2026, adopted by the resolution of the Council of Ministers of 23 December 2025, continues the activities of 2024–2025, focusing on the full implementation of expert centres (OECR), the launch of the Polish Rare Diseases Register and the introduction of the Patient Card.

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Elaboration and Adoption/How was the original plan elaborated?

In Poland, the coordination and supervision of the National Plan is entrusted to the minister responsible for health matters. In order to properly implement the tasks listed in the Rare Diseases Plan, the MoH established a Council for Rare Diseases. In cooperation with the minister responsible for health, they initiate and monitor the implementation of individual stages of the plan in accordance with the given schedule, including, among others, appointing appropriate expert teams, announcing competitions for obtaining OECR status, coordinating cooperation between OECRs, and fulfilling other tasks arising from its competences. The Council for Rare Diseases includes the following stakeholder types:

- Patients/people with a lived experience of a rare condition
- Representative from Poland'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
- Research funders
- Researchers/clinicians from rare disease centres
- European Reference Network (ERN) coordinators or representatives

How is it Implemented and/or Monitored or Evaluated?

The same group that was responsible for drafting Poland's National Plan for Rare Disease is responsible for implementing and overseeing the National Plan. This group was set-up specifically for drafting and implementing Poland's National Plan for Rare Disease. This group meets regularly.

Formal and comprehensive evaluation of previous iterations of the National Plan for Rare Disease took place. Monitoring of the National Plan is carried out by the Rare Disease Council (RDC) and expert opinions are sought, where required. Follow up recommendations are made to the MoH by the RDC, based on RDC meeting reports and national data on rare diseases.

A dedicated budget is in place for Poland's National Plan for Rare Disease. For the year 2026, public funds totalling 6,350,000 PLN have been allocated to the plan.

Select achievements

Below are five select achievements, as a result of Poland's National Plan for Rare Disease:

- Establishment of a Rare Diseases Council
- Centres of expertise are being established and 44 units are members of ERN groups in 23 hospitals
- Neonatal screening has been significantly expanded
- The number of drug programs has significantly increased
- There is close cooperation with patient organisations. A Polish Rare Diseases Register (including Orpha coding) will soon be launched and a Patient Card will be introduced

Additionally, Poland has collaborated with Germany to implement a cross-border screening programme.

Rare Disease Research Programmes and Funding

Poland's National Plan for Rare Disease addresses rare disease research in the following ways:

- Development of genetic and laboratory diagnostics
- Funding for improving public reimbursement/access to clinically indicated diagnostics for rare disease patients
- Scientific research funding (approx. 100,000,000 pln) for non-commercial clinical trials
- Quality control system for laboratories performing large-scale genomic research to ensure the reliability and clinical utility of results

The national plan includes specific mentions of and promotes alignment with the European Network for Health Technology Assessment (EunetHTA), EMA, HTA agencies and drug assessment agencies and IRDiRC. The plan also includes recommendations for international cooperation.

The following areas relating to rare disease research are included within the national plan:

- National RD research investment, opportunities and funding calls
- Registries or registry catalogues for rare diseases
- Biobanks/biosample catalogues for rare diseases
- Ontologies, codification and/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

The national plan acts as a framework. Dedicated funding for rare disease research is provided though regular funding calls by both private and public bodies. Funding is provided by:

- The medical research agency Agencja Badań Medycznych (ABM)
- The National Science Centre Narodowe Centrum Nauki (NCN)
- The National Centre for Research and Development Narodowe Centrum Badań i Rozwoju (NCBR)
- Academic programmes including the Polish Academy of Science Polska Akademia Nauk (PAN)

Rare Disease Registration and Biobanking

In Poland, there is a national registry for rare disease. Whilst a fully comprehensive pan rare disease registry is not yet operational, a national registry framework is under development. Further information is available here.

The national registry for rare disease embeds FAIR (FAIR stands for Findable, Accessible, Interoperable, Reusable) data principles. Access to the rare disease registry will be regulated by law and is currently in the final stage of the legislative process. The registry is established with regards to the principles of interoperability with public healthcare databases and integration with hospital data systems, in order to minimise physician involvement in data entry. To ensure the reliability of data and the possibility of linking with other public databases, the registry uses classifications, coding systems and reporting systems including ICD9, ICD10, ORDO and OMIM. The registry collects the common data elements for rare disease registries, as recommended by the European Commission through the European Platform on Rare Disease Registration (EU RD Platform).

There are also registries for specific rare diseases within Poland.

In Poland, there are national and institutional biobanks for the collection of rare disease biosamples. Of the 65 biobanks in Poland, 12 are specifically dedicated to rare disease biosamples. The biobanks are part of BBMRI-ERIC (Biobanking and BioMolecular resources Research Infrastructure) catalogue.

Organisation of Rare Disease Care

Centres of Expertise

In Poland there is an official policy for designating Centres of Expertise (CoE) for rare disease. Poland has adopted both a national and a regional approach to designating CoE for rare disease. There are 44 centres that function as CoE at disease-group level, many of which are ERN-affiliated, operating in 23 hospitals. Polish healthcare providers participate in European Reference Networks, which effectively meet the CoE criteria for specific rare disease groups.

Poland uses a national designation process that is closely aligned with the criteria used for designation of Healthcare Providers (HCPs) within European Reference Networks (ERNs). In practice:

- ERN HCP designation criteria are used as the main benchmark
- National designation is not fully synonymous with ERN membership, but criteria are largely equivalent
- Centres are typically designated or recognised by the Ministry of Health and national consultants

Poland's criteria for designating centres of expertise for rare disease include the following requirements (based on the 2011 EUCERD Criteria for Centres of Expertise):

- Contribute to state-of-the-art research: Centres are expected to participate in clinical research, international studies, and ERN collaboration.
- Bring together, or coordinate, or liaise with social services or social workers, in order to serve the specific medical, rehabilitation and palliative needs of rare diseases patients: Centres are expected to cooperate with rehabilitation services and, where relevant, social support structures, particularly for paediatric and complex rare conditions.
- Collaborate with patient organisations to bring in the patients' perspective: Patient organisations are formally involved in rare disease policy discussions and centre development, under the National Plan for rare disease.
- Provide education and training to healthcare professionals from all disciplines, including paramedical specialists and non-healthcare professionals (such as schoolteachers, personal/homecare facilitators) whenever possible: Centres are expected to provide training for healthcare professionals and contribute to professional education.
- Contribute to and provide accessible information adapted to the specific needs of patients and their families, of health and social professionals: National rare disease information portals and specialist clinics provide disease-specific information and guidance.
- Participate in data collection for clinical research and public health purposes: Centres contribute to registries, clinical databases, and public health data collection (including through ERNs and national systems).

ERN participation

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

Newborn Screening

As of 2026, the Polish national newborn screening programme covers 30 rare conditions. The full and officially updated list is maintained by the National Newborn Screening (NBS) Programme, coordinated by the Institute of Mother and Child (Instytut Matki i Dziecka), under the authority of the Ministry of Health.

The Ministry of Health is responsible for setting the official panel and approving expansions as part of the government's health policy programme. Proposed changes to the NBS screening panel (e.g. adding new disease tests) are evaluated by the Agencja Oceny Technologii Medycznych i Taryfikacji (AOTMiT), the Polish Agency for Health Technology Assessment and Tariff System, which provides a formal opinion on draft policy changes. The AOTMiT's opinions are typically based on evidence of clinical effectiveness, health impact, technical feasibility, and cost implications of adding new conditions. The process also involves review by AOTMiT's Rada Przejrzystości (Transparency Council), which issues separate opinions on the policy draft.

To add a new condition, a formal project or draft change to the national NBS programme is prepared (often by the Ministry of Health or specialty clinical bodies) proposing new conditions. This draft is submitted to AOTMiT for evaluation under public health assessment procedures (linked to the public health benefits and system costs). After receiving opinions from AOTMiT (and sometimes related expert bodies), the Ministry may revise the programme and then formally adopt the updated panel within the health policy programme. New expansions can be introduced initially as pilot screening projects before full national roll-out (e.g. pilot for severe combined immunodeficiency and other rare disorders which started in 2025).

Whilst Poland does not publish a single consolidated 'screening criteria' checklist, the evidence and opinions around expansions reflect implicit assessment criteria:

- Health benefit
- Ability of early detection to significantly improve outcomes, prevent disability, or reduce mortality
- Availability of effective treatment or follow-up care once diagnosed
- Scientific and clinical evidence supporting screening accuracy, benefit, and feasibility.
- Technical feasibility and laboratory capability to reliably test the condition on dried blood spots.

Evidence from other countries is routinely used when expanding Poland's NBS panel. The AOTMiT assesses proposals using international clinical studies, foreign pilot data, and international HTA methods. The Instytut Matki i Dziecka participates in EU-funded and cross-border research projects, including regional pilots (e.g. with Germany), and incorporates findings into national practice. Poland collaborates within European rare-disease networks (e.g. ERNs), sharing expertise and aligning diagnostic and follow-up standards for conditions such as SMA. Poland is not a core pilot country in Screen4Care but policymakers and experts follow and use outputs from major EU initiatives when shaping national pilots and programme updates.

Diagnostics

In Poland, diagnostic tests are provided by specialised and certified laboratories. Genetic testing for rare diseases is performed mainly by university hospitals, specialised clinical genetics centres and accredited molecular diagnostic laboratories. Lists of laboratories exist within administrative and reimbursement frameworks (e.g. National Health Fund contracts, professional accreditation systems). However, there is no single publicly available national registry of reference laboratories specifically dedicated to rare disease genetic testing. ERN affiliated centres function as de facto reference laboratories for specific disease groups.

Genetic testing is partly reimbursed through the public health system (National Health Fund) when clinically justified and ordered by authorised specialists. Selected genetic tests are publicly funded in defined clinical pathways, whilst the National Plan refers to ongoing work on expanding the list of guaranteed healthcare services to include additional advanced genetic tests, such as targeted Next Generation Screening (NGS) panels and Whole Exome Sequencing (WES). Access and scope depends on the clinical indication patient pathway and regional and institutional capacity. There is no universal or automatic reimbursement for all genetic tests, reimbursement is conditional.

Poland collaborates with other countries, specifically Germany, on cross-border genetic testing. Cross-border genetic testing takes place on a case-by-case basis through ERNs under the EU Cross-Border Healthcare Directive.

Poland does not have a single, formal, nationwide 'Undiagnosed Diseases Programme' but several mechanisms exist. The ERN network is often used to solve undiagnosed cases through ERN expert panels.

There is a clinical practice and health system structure to provide genetic counselling for patients with a suspected or confirmed rare disease. Genetic counselling is recognised as part of standard care for patients with suspected or confirmed genetic rare diseases. Counselling is provided mainly by clinical geneticists in specialised centres. Although a policy framework exists, access may be limited by workforce capacity and regional availability,

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National Alliances of Rare Disease Patient Organisations

KRAJOWE FORUM ORFAN is Poland's National Alliance of rare disease patient organisations. For further information, click here. Poland's National Alliance of rare disease patient organisations:

- Is involved in setting strategic direction for rare disease research
- Provides webinars and other forms of learning and capacity-building on patient engagement in rare disease research
- Conducts surveys amongst its members on patient engagement in rare disease research.

Information Resources for Rare Disease

National Orphanet Engagement

In Poland there is a dedicated Orphanet team, hosted by CENTRUM ZDROWIA DZIECKA. The Orphanet team receives 80% of funding from the EU funded grant Orphanet Data for Rare Diseases (OD4RD 2) and 20% of funding is provided by Poland's Ministry of Health. The Orphanet team encourages and ensures that national resources e.g. registries are conveyed to the central Orphanet database, to be included in the full directory. They do this through training and having a representative of the national Orphanet team included as a member of the Council for Rare Diseases at the Ministry of Health.

Helplines

In Poland there are national rare disease information portals, coordinated under the Ministry of Health framework, that are available for patients and healthcare professionals to use. The portals are:

- Dedicated to rare diseases as a whole
- Operated by professionals (medical experts, researchers, institutional staff)
- Supplemented by input from patient organisations

- Some resources allow email or contact-form enquiries, rather than a continuous phone helpline
- Information is available mainly in Polish, with selected resources also available in English
- Content includes:
    - Disease information
    - Diagnostic pathways
    - Expert centres
    - Links to patient organisations
    - References to European resources such as ERN or Orphanet

The informational portals provide information and guidance, rather than case-by-case medical advice. The portals receive public funding only.

Poland does not operate a single, nationwide, 24/7 telephone helpline covering all rare diseases.

Training and Education

Within Poland, there are dedicated rare disease training activities. Topics of rare disease training activities include:

- Diagnostics: Training for clinicians and laboratory specialists in rare disease diagnostics (including genetic testing, NBS expansion, and genomic interpretation) is provided through medical universities, clinical genetics departments, and ERN-linked centres.
- Rare disease awareness raising: Educational initiatives for healthcare professionals are organised under the National Plan and through academic centres and patient organisations to improve recognition of rare diseases.
- Data management and data quality: Clinical genetics specialisation training and laboratory accreditation processes include quality standards for genomic testing, aligned with EU and ERN standards.
- Registries: Training related to rare disease registries and data reporting occurs within ERN participation and national registry development initiatives.
- Clinical research: Centres of Expertise and university hospitals provide training in rare-disease-focused clinical research, including participation in international trials and ERN studies
- Standards and quality of genetics/genomics data in clinical practice and laboratories

Whilst training activities are provided in Poland, they are fragmented, project-based and decentralised. There is no single, national, coordinated training programme for rare diseases as a whole. Activities are delivered by universities, expert centres, research projects, ERN affiliated centres and patient organisations. Additionally, the Medical University Gdansk is part of ERDERA and is involved in a task to create an academic programme for Rare Disease.

Rare disease training in Poland is mainly free of charge (particularly webinars, project-based trainings and patient-focused activities), however some postgraduate or specialist courses are fee-based. Training is primarily provided in Polish but some materials and trainings are available in English (particularly those developed in EU projects and ERNs).

Orphan Medicinal Products (OMPs)

Poland reimburses and makes available approximately 90–110 OMPs. This figure refers to OMPs that are authorised by the European Medicines Agency (EMA) and are priced, reimbursed or provided through national drug programmes, within the Polish public health system (National Health Fund). It does not include all EMA authorised OMPs, only those with positive reimbursement decisions in Poland. It is important to note that the exact number fluctuates annually due to new EMA approvals, reimbursement decisions, withdrawals and renegotiations. Poland does not publish a single static official list explicitly labelled 'reimbursed OMPs'. Based on this, figures are derived from Ministry of Health reimbursement lists, National drug programmes and Orphan designation status at EMA.

In Poland, pricing and reimbursement decisions are centralised and taken by the Ministry of Health. Access to OMPs is often provided through dedicated drug programmes, particularly for ultra-rare and high- cost therapies. Reimbursement negotiations consider clinical effectiveness, budget impact and unmet medical needs. Access has improved significantly in recent years but delays between EMA authorisation and national reimbursement still occur, particularly for very high-cost therapies.

Poland has formal mechanisms that allow access to medicinal products for rare diseases before full reimbursement including:

- Compassionate use procedures
- Individual patient access under specific legal and medical criteria
- Temporary access mechanisms linked to ongoing reimbursement processes

These mechanisms are regulated nationally and applied on a case-by-case basis.

Poland participates in structured European cooperation, relating to access to medicines, including:

- EU-level cooperation on Health Technology Assessment (HTA)
- Joint clinical assessments under the EU HTA Regulation
- Informal and technical collaboration with other Member States on pricing, evidence generation and access pathways

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The Data Contributing Committee of Poland, which provided this 2026 data (correct as of the end of February 2026) 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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- Maja Bartoszewicz-Moritz - coordinator of Patient COUNCIL at Rare Disease Centre of GUMed,Patien Advocate