European Rare Diseases
Research Alliance

Resource on the State of the Art of Rare Disease Activities

2025 Report for Bulgaria

National Mirror Group

Bulgaria has a National Mirror Group (NMG), which was launched on 25th September 2025. The NMG is coordinated by Rare Diseases Bulgaria. This NMG has overseen the collection of data from Bulgaria via a data contributing committee. Contributors are listed at the end of this report.

Definition of a Rare Disease

Bulgaria 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.). Bulgaria's most recent National Plan, now expired, espoused this definition.

Status Quo of any National Plan or Strategy for Rare Disease

What is the status quo?

Bulgaria does not currently have a national plan or strategy that can be described as live and in-date. Between 2009-2013, Bulgaria did have a National Plan/Strategy named the National Programme for Rare Diseases by Bulgaria's Ministry of Health, within the framework of the Europlan project. Bulgaria was therefore one of the first EU countries to adopt a national plan/strategy for rare disease.

There are no plans to renew or a adopt a new national plan or strategy for rare disease. The Ministry of Health (MoH) considers Bulgaria's Ordinance 16 for Rare Diseases, adopted in 2014, as a successor of the previous programme. Ordinance 16 is legislation that includes terms and conditions for registration of rare diseases and for the expert centres and reference networks for rare diseases.

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Elaboration and Adoption/How was the original plan elaborated?

Bulgaria's previous National Programme for rare diseases had a dedicated group responsible for overseeing the drafting/adoption of the Programme. The following stakeholder types were included in this group:

- Patients/people with lived experience of rare condition
- Bulgaria's National Alliance of Patient Organisations
- Health Ministry/Competent National Authority in charge of Health or Care
- Researchers/clinicians from rare disease centres

Was there funding for the Plan? How was it Implemented and/or Monitored or Evaluated?

A different group was set-up to implement and oversee the National Programme for rare diseases for Bulgaria. The following stakeholder types were included in this group:

- Patients/people with lived experience of rare condition
- Bulgaria's National Alliance of Patient Organisations
- Health Ministry/Competent National Authority in charge of Health or Care
- Researchers/clinicians from rare disease centres

This implementation/oversight group was specifically set-up for this purpose. The group met regularly when the Programme was in date. This group also carried out formal monitoring and evaluation of the Programme.

Bulgaria's National Programme for rare diseases had a dedicated budget of €1 million annually.

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

Bulgaria's National Programme for rare diseases has now expired but it did address rare disease research.

Rare Disease Research Programmes and Funding

Ther are no specific programmes or funding calls reserved for rare disease in Bulgaria, but rare disease projects are funded within general research programmes.

Rare Disease Registration and Biobanking

Bulgaria does not have a national nor regional rare disease registry. There are no current plans to create a national or regional registry for rare disease due to the lack of political support and resources. However, disease specific registries for rare disease exist in Bulgaria. These registries are based at

national expert centres, within European Reference Networks (ERNs), and at hospital clinics on a voluntary basis.

Bulgaria has biobanks but these are not specific for the collection of rare disease biosamples. The Molecular Medicine Center (MMC), Medical University Sofia (MU-Sofia) run the Bulgarian national node (BBMRI.bg) of the BBMRI-ERIC (the European Biobanking Research Infrastructure). The metadata for biobanks in Bulgaria are included within the BBMRI catalogue.

Organisation of Rare Disease Care

Centres of Expertise

Bulgaria has a national policy for designating Centres of Expertise for rare disease. Bulgaria has centres designated for groups of rare diseases, as well as specific conditions. The national criteria for designating Centres of Expertise is detailed within Bulgaria's Ordinance 16/2014. As of October 2025, there are 40 designated Centres of Expertise in Bulgaria. The centres are listed here.

Bulgaria'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
- Bring together, or coordinate, multidisciplinary specialised medical and paramedical expertise such as physiotherapists, speech therapists, dieticians, genetic counsellors, etc.
- Collaborate with patient organisations to bring in the patients' perspective
- 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
- Contribute to and provide accessible information adapted to the specific needs of patients and their families, of health and social professionals
- Participate in data collection for clinical research and public health purposes

ERN participation

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

Newborn Screening

Currently, three conditions are screened for in Bulgaria's Newborn Screening programme. They are Phenylketonuria (PKU), Congenital Adrenal Hyperplasia (CAH) and Congenital Hypothyroidism (CH). A further three diseases will be screened for in Bulgaria in 2026. Those diseases are Spinal Muscular Atrophy (SMA), Cystic fibrosis (CF) and Severe Combined Immunodeficiencies (SCID). A policy (Ordinance No.26/2007) exists for Newborn Screening in Bulgaria.

There are collaborations between Bulgaria and other countries around newborn screening decisions. The Bulgarian Association for Promotion of Education and Science (BAPES) and the Bulgarian Association for Personalised Medicine (BAPEMED) are involved in Screen4Care.

Diagnostics

A current list of laboratories which carry out genetic testing is included in Bulgaria's Ordinance 26/2007. These laboratories receive funding for reagents and consumables only for genetic tests. Genetic tests for children are reimbursed in Bulgaria.

Bulgaria is involved in initiatives to address undiagnosed patients/people with currently undiagnosable conditions. Bulgaria participates in Undiagnosed Diseases Network International (UDNI) and is a partner in ERDERA's Clinical Research Network. There is a policy in place to ensure the provision of genetic counselling for patients with a suspected or confirmed rare disease. In Bulgaria's current National Medical Genetics guidelines, genetic counselling is a mandatory component of genetic testing.

National Alliances of Rare Disease Patient Organisations

Rare Diseases Bulgaria Association is Bulgaria's National Alliance of Rare Disease Patient Organisations. Bulgaria's National Alliance is involved in setting strategic direction for rare disease research. Rare Diseases Bulgaria is a partner in ERDERA and leads Bulgaria's National Mirror Group. They are involved in webinars and capacity-building programmes through EURORDIS and ERNs. Surveys on patient engagement in rare disease research are conducted amongst their members through the EURORDIS Rare barometer initiative.

Bulgaria's National Alliance has identified a lack of budget for patient advocacy organisations as a barrier to patient engagement in rare disease research.

Information Resources for Rare Disease

National Orphanet Engagement

Bulgaria has an operational, national, Orphanet team, which is hosted by The Institute of Rare Diseases. The team receives funding from an international grant (OD4RD).

Helplines

Bulgaria has a national helpline/portal in place that is dedicated to rare disease. This is run by the Institute for Rare Diseases, Bulgarian Huntington Association, Rare Diseases Bulgaria, and Bulgaria's Ministry of Health. Bulgaria's national helpline/portal receives a mix of public and private funding. It is only available for patients with a rare disease.

Training and Education

In Bulgaria, there are rare disease training activities offered by ERN expert Centres, for free, to researchers and the training is delivered in Bulgarian. Topics included in these training activities are diagnostics, awareness of rare disease and clinical research.

Orphan Medicinal Products (OMPs)

41 OMPs are included within Bulgaria's Positive Drug List (PDL). Further information can be found here. Bulgaria does have Early Access Programmes/Expanded Access programmes. Compassionate treatment in Bulgaria is regulated by changes to the Law on Medicinal Products in Human Medicine and changes to Ordinance No 10, adopted in October 2018, and funded by pharmaceutical companies. Bulgaria is not involved in any formal efforts or initiatives with other countries to support access to medicines and therapies for rare diseases.

ERDERA European Rare Diseases Research Alliance

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The Data Contributing Committee of Bulgaria, 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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