

Resource on the State of the Art of Rare Disease Activities

2025 Report for Cyprus

National Mirror Group

Cyprus has a National Mirror Group (NMG), which was launched on 25th July 2024. The NMG is coordinated by the Thalassaemia International Federation. This NMG has overseen the collection of data from Cyprus via a data contributing committee. Contributors are listed at the end of this report.

Definition of a Rare Disease

Cyprus 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 Disease

What is the status quo?

Cyprus has a National Strategy for Rare Disease that can be described as open-ended and live. The current National Strategy was adopted in 2013 and was always open-ended. However, it is currently undergoing revision in order to make it more relevant to the present landscape, needs and challenges in Cyprus (for instance, a new national health system was adopted in 2019). A multi-stakeholder workshop will be organised for the purpose of reviewing the strategy and thereafter, the strategy document will be updated and will include timeframes and involved stakeholders.

Elaboration and Adoption/How was the original strategy elaborated?

There was a dedicated group in charge of overseeing the drafting and adoption of Cyprus' National Strategy for Rare Disease. This group included the following stakeholders:

- Patients/people with lived experience of rare condition
- 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

The National Committee for Rare Disorders is now mandated to update the national strategy. In addition to the stakeholders listed above, this body includes the Cyprus Institute of Neurology and Genetics, Cyprus Medical Association (specifically in the fields of rheumatology and oncology), Cyprus Patient Federation, Cyprus Alliance for Rare Disorders, Metabolic Syndromes Clinic, and the Research and Innovation Foundation.

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

There is no dedicated funding for Cyprus' National Strategy for Rare Disease itself, nor for any of the actions outlined within the strategy.

The same group responsible for overseeing the drafting and adoption of Cyprus' National Strategy for Rare Disease is responsible for overseeing the implementation of the strategy. The group meets occasionally but irregularly.

No formal monitoring or evaluation has taken place for Cyprus' National Strategy for Rare Disease.

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

Cyprus' National Strategy for Rare Disease does address research. The strategy includes a specific section on Research. In Cyprus, research has focused on rare conditions that affect subgroups of the local population such as Thalassaemia, Friedreich's ataxia (which is limited to communities in the Paphos district), Sandhoff disease (confined to the Maronite ethnic minority), gangliosidosis GM1 (confined mainly to a single village) and others.

There is a budget allocation for core translational research activities for rare disease at CING (The Cyprus Institute of Neurology & Genetics) for diagnostic and therapy development. The Research Innovation Foundation provides funding for rare disease research via ERDF (European Regional Development Fund), Horizon Europe etc. but there is no set budget for rare disease research in the National Strategy for Rare Disease. There are several research teams working in the following centres: Cyprus Institute of Neurology and Genetics (CING), the University of Cyprus, the Karaïskakio Institute, and the Makarios Hospital. These institutes are partially supported by government funds so there is a contribution, including from the Research and Innovation Foundation (RIF) through which grants from European and local sources are accessed. There is an annual budget allocation for rare disease research for CING departments dedicated to specific disease groups.

Rare Disease Research Programmes and Funding

Rare disease specific research funding programmes, offered by both public and private bodies, are available in Cyprus. These funding calls are offered sporadically. Funding programmes are provided by the following:

Public:

1. Research Innovation Foundation (RIF) through ERDF (Operational Programme ΘΑΛΕΙΑ 2021-2027) including ERDERA JTC calls for Proposals, RESTART 2016 – 2020, Horizon 2020 etc.
2. Cyprus Institute of Neurology & Genetics (government funded)
3. University of Cyprus (government funded)

Private bodies:

1. George and Kaity David Foundation
2. Bank of Cyprus

Prior to the 2024 ERDERA Call for Proposals, there were no specific call for proposals for rare disease, and therefore any successful proposals and subsequent funded projects that focused on one or more rare disease are encompassed in the list of funded projects of the Research and Innovation Foundation [here](#).

Rare Disease is included as one of the thematic priorities in Cyprus's Smart Specialisation Strategy 2023-2030. Cyprus participates in the Horizon Europe Partnership on Rare Diseases with a national contribution of €3 million, providing opportunities to strengthen research capacity and collaboration in this field.

Rare Disease Registration and Biobanking

Cyprus does not currently have an operational cross-disease national rare disease registry; several disease-specific registries exist or are under development. Within Cyprus' current National Strategy for rare disease, a proposal for a national registry was included. This registry has not materialised but the Cyprus National Mirror Group has proposed developing a national registry that will include all people living with a rare disease. To do this, there is a need to align with the National Electronic Health Authority (NEHA), which is developing the National Health File. This alignment may delay the final product. A major barrier in doing this is the current use of paper records for patient data and the amount of time required to develop a digital patient record, from which registry data could be derived automatically. The creation of registries is therefore dependant on disease specific efforts of digitisation (and FAIRification) of existing patient data and on putting a system in place for registry maintenance.

Rare disease specific registries developed by interested scientific and clinical groups of rare diseases exist either in separate departments that deal with patients with rare diseases or with patients' groups. Examples of registries include the registry for neurological disorders and myopathies of the Cyprus

Institute of Neurology and Genetics (CING), the Archive of Congenital Heart disease, the registry of haematological disorders at the Nicosia General Hospital, and the registry of the Cyprus Thalassaemia Centre. The hemoglobinopathy national registry is currently being compiled by the collection of data from the national thalassemia clinics. To date, 409 patients have been consented, including data entry for Nicosia and partial data entry for Limassol; Larnaca and Paphos are pending. Additionally, disease specific registries have been developed by patient groups.

There is a national biobank within Cyprus, but it is not specifically or exclusively for the collection of rare disease biosamples. The [Biobank.cy](https://biobank.cy) is managed by The Molecular Medicine Research Centre (MMRC) of the University of Cyprus. The metadata for biobanks in Cyprus is available within [Biobank.cy](https://biobank.cy) and the [Biobanking and BioMolecular resources Research Infrastructure \(BBMRI\)](#).

Organisation of Rare Disease Care

Centres of Expertise

There is a national policy in place in Cyprus to designate Centres of Expertise in Rare Disease. Cyprus has Centres of Expertise for groups of rare disease.

EUCERD criteria were used to designate Centres of Expertise, together with the ERN member criteria. Basically, there is an open accreditation process for National Centres of Excellence & Reference Centres for departments across Cyprus to be certified within the European Reference Centre Network. There is also an ongoing procedure in order to evaluate other departments as national centres for rare diseases. However, there is no comprehensive policy/strategy/informative campaign to address the patients in these centres when necessary. This leads to a lack of special attention and the opportunity to centralise management, that could help accelerate expertise.

The following requirements are included within the criteria:

- 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
- 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
- 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

Centres of Expertise have been designated to participate in the following European Reference Networks (ERNs): ERN-RND (Rare Neurological Diseases); EpiCARE (Rare and Complex Epilepsies);

ERN-ITHACA (Rare Malformations); ERN-EuroBloodNet (mainly through the Thalassaemia centre); ERN EURO-NMD (neuromuscular diseases); and ERN-LUNG (Primary Ciliary Dyskinesia).

ERNs

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

Newborn Screening

Two conditions are screened for in Cyprus' newborn screening programme. The conditions are Phenylketonuria (PKU) and Congenital Hypothyroidism (CHT). However, a new neonatal screening program has been put in place, that will screen for an additional eight metabolic disorders. The Neonatal Screening Program in Cyprus is run by an NGO (the Center for Preventive Paediatrics "Americos Argyriou" - CPP). It is mainly but not wholly financed by Cyprus' government. In parallel the group has a Neonatal Hearing Screening Program. The paediatric Society of Cyprus (a scientific body) is represented, and along with the Ministry of Health, can suggest additions to the newborn screening programme. The small population of Cyprus makes other rare metabolic conditions detectable only every few years so only the two more common conditions have remained on the list. The new and expanded newborn screening programme will be implemented by a coalition between CPP, the Cyprus Institute of Neurology & Genetics (CING) and the Archbishop Makarios III Hospital (Ministry of Health).

Diagnostics

Diagnostic testing is carried out by the following centres in Cyprus:

- Thalassaemia Centre at the Makarios Hospital, for population screening of haemoglobin disorders
- The Cyprus Institute of Neurology and Genetics for most molecular tests for a wide range of genetic diseases
- The Karaïskakio Foundation laboratories for haematological malignancies and HLA (Human Leukocyte Antigens) typing
- The Center for Preventive Paediatrics "Americos Argyriou" for neonatal screening
- [Medicover Genetics Cyprus](#) is a private laboratory that covers a wide range of genetic conditions including rare diseases

There is provision for the reimbursement of tests through the National Health System (NHS). Additionally, there is a procedure where the ministry of health (Government's Subsidized Patients sector) can cover the financial cost of a genetic test, not provided in laboratories within Cyprus, under revision of a special committee.

There is not a standardised policy for cross-border testing. There are only individualised cases where the team addressing the patient is collaborating with laboratories outside of Cyprus, in order to meet the patient's needs. There are agreements by individual laboratories but on a payment basis, unless approved by the Government's Subsidized Patients sector (for those services which are not available in Cyprus).

There is no national approach to address undiagnosed patients/people with currently undiagnosable conditions in Cyprus. However, there are physicians in Cyprus who are part of the Undiagnosed Disease Network International (UDNI) and actively participate in the diagnostic working group and Hackathons.

There is a policy in place in Cyprus to ensure national providers provide genetic counselling for patients with a suspected or confirmed rare disease. Patients with rare and genetic disorders have the option to be referred for genetic counselling either to the CING, the Genetics clinic of the paediatric department of the Makarios Hospital, or the Rare Disease clinic at the Karaiskakeio Foundation, or in the case of haemoglobin disorders to the Cyprus Thalassaemia Centre. There are no defined policies about the professionals that request genetic testing. This creates a difficult situation with over testing, or testing with incorrect phenotyping, that leads to confusing results creating patients' anxiety. In a lot of cases, there is no pre-test genetic counselling.

National Alliances of Rare Disease Patient Organisations

Cyprus Alliance for Rare Disorders (CARD) is Cyprus' National Alliance of Rare Disease Patient Organisations. Further information can be found [here](#). CARD is involved in Cyprus' National Mirror Group and is involved in setting the strategic direction of rare disease research in Cyprus. CARD participates in the National Committee for Rare Diseases, that is tasked with the revision of the current national strategy, as well as other direct actions on an EU level. CARD hosts a range of patient engagement capacity building sessions on many subjects including accessing new pharmaceutical products, access to the state's social services/benefits, and psychosocial empowerment.

CARD identified that lack of critical mass in small countries is the biggest factor for the unavailability of research and is a barrier to patient engagement in rare disease research. Other barriers to patient engagement in rare disease research include language used and the geographic isolation of Cyprus.

Information Resources for Rare Disease

National Orphanet Engagement

There is no operational Orphanet team in Cyprus.

Helplines

There is a national helpline in Cyprus dedicated to rare disease available for anyone to use. CARD provides this service and patients can engage with CARD either by telephone or by interview. CARD

can provide support and advice for social issues and administrative issues. For medical issues, CARD refers patients to local experts. The helpline is supported through a mixture of public and private funding. An information portal is available for both patients and professionals. The portal was funded by the Cyprus Research and Innovation Foundation.

Training and Education

Rare disease training activities are available in Cyprus. Topics of these training activities include:

- Diagnostics
- Raising awareness of rare disease
- PPIE (Patient and Public Involvement and Engagement)

Examples of rare disease training activities in Cyprus are postgraduate courses (MSc & PhD) in molecular genetics run by CING and an annual scientific conference hosted by Cyprus' National Alliance of rare disease patient organisations.

Orphan Medicinal Products (OMPs)

The exact number of OMPs available in Cyprus is unknown. It is difficult to estimate due to the different processes and bodies involved. More specifically, OMP integration in the national drugs list following EMA central authorisation is the responsibility of the Pharmaceutical Services of the Ministry of Health. List pricing is also the responsibility of the Pharmaceutical Services. Price negotiation with the marketing authorisation holder, reimbursement and management of the list of reimbursed OMPs via the National Health Service (NHS) is the responsibility of the Health Insurance Organisation (HIO), the only payer in the national health system. Not all OMPs are reimbursed by HIO. If an OMP is required but not reimbursed, a request can be made for case-by-case consideration by the Committee for the Examination of Named Patient Requests for Medicines which falls under the HIO. In addition, OMPs not in the HIO list of reimbursed medicines, may be accessed by patients via a documented request to the Committee for the Examination of Individual Drug Requests under the Ministry of Health's Pharmaceutical Services.

An effort to ascertain how many OMPs authorised by EMA are available (i.e. priced and reimbursed) via the NHS and how many are available via the Committee for the Examination of Named Patient Requests for Medicines was conducted by the CYNMG and the Ministry of Health. It was found that 129 OMPs are available via the NHS and a further 14 via the Committee for the Examination of Named Patient Requests for Medicines, although this does not take into account OMPs that may be accessed via other routes (e.g. Committee for the Examination of Individual Drug Requests).



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The Data Contributing Committee of Cyprus, 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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