Resource on the State of the Art of Rare Disease Activities

2025 Report for Türkiye

National Mirror Group

Türkiye has a National Mirror Group (NMG), which was launched on 10th February 2025. The NMG is coordinated by Izmir Biomedicine and Genome Centre (IBG). This NMG has overseen the collection of data from Türkiye via a data contributing committee. Contributors are listed at the end of this report.

Definition of a Rare Disease

Türkiye 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 Strategy espouses the definition that a rare disease affects 1 in 2,000 people.

Status Quo of any National Plan or Strategy for Rare Disease

What is the status quo?

Türkiye has a National Strategy that can be described as live, in-date and timebound. Türkiye's Rare Diseases Health Strategy Document and Action Plan covers the period of 2023-2027.

Elaboration and Adoption/How was the original strategy elaborated?

Türkiye's Rare Diseases Health Strategy Document and Action Plan was developed according to the EU Commission's Council Recommendation No. 2009/c 151/02 on Action in the Field of Rare Diseases, within the framework of the Europlan project.

A dedicated group was responsible for overseeing the drafting and adoption of the national strategy. The group included the following stakeholders:

- Directorate General of Health Services
- Directorate General of Public Health

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- Health Institutes of Türkiye (TÜSEB)
- Turkish Medicines and Medical Devices Agency
- Ministry of Health Press Counsel
- Ministry of Public and Social Services
- Directorate General of Health Information Systems
- Directorate General of Public Hospitals
- Directorate General of Health Development
- Council of Graduate Education
- Scientific and Technological Research Council of Türkiye (Tübitak)

The same group is responsible for implementing and overseeing Türkiye's Rare Diseases Health Strategy Document and Action Plan. This group meets occasionally but irregularly.

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

No funding is committed to Türkiye's national strategy itself but there are funds dedicated to deliver actions within the action plan.

Formal and comprehensive evaluation takes place of Türkiye's national strategy. The overseeing body within the Ministry of Health follows the national strategy and its progress through predetermined, yearly, progress, indicators.

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

Türkiye's national strategy addresses rare disease research. Research related activities within the strategy are financially supported through TUBITAK and TUSEB. The strategy includes a specific call section for Rare and Undiagnosed Diseases. The strategy promotes alignment with Orphanet and specifically aims to raise awareness with primary care practitioners, relevant specialists, and healthcare providers involved in the diagnosis and follow-up process. The national strategy addresses the following topics related to rare disease research:

- National RD research investment, opportunities and funding calls
- International/transnational RD research investment, opportunities and funding calls
- Registries or registry catalogues for rare diseases
- Biobanks/biosample catalogues for rare diseases
- Ontologies, codification 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 strategy also includes plans to create an inventory for researchers working in rare diseases, develop infrastructure to produce local diagnostic and screening tests, and treatment methods.

Rare Disease Research Programmes and Funding

There are no programmes or funding calls reserved specifically for rare disease projects in Türkiye, but rare disease projects are funded from general research programmes. A policy decision has been made to allocate a portion of the national research budget specifically to rare disease research.

Rare Disease Registration and Biobanking

Türkiye has a National Rare Disease Registry System, which integrates more than 8,000 diseases translated into Turkish from Orphanet terminology. Specialists in medical genetics, paediatric genetics, paediatric metabolism, paediatric neurology, and clinical genetics are encouraged to enter data into the registry, but the system allows other specialties to also enter data. Integration with hospital and laboratory IT systems is ongoing but not yet complete, and data entry remains voluntary. The national registry uses ICD10 and Orphanet coding systems.

There are some disease-specific registries run by clinical/research groups in Türkiye e.g. neuromuscular disease registries. Many disease-specific registries are led by university hospitals, research centres or professional societies and are not always centrally coordinated.

Türkiye has four major biobanks: IBG Biobank (Izmir), Acıbadem University Biobank (Istanbul), Hacettepe University Biobank (Ankara), and Istanbul University Biobank (Istanbul). The biobanks are all BBMRI (Biobanking and BioMolecular resources Research Infrastructure) members. The biobanks are regulated by the Ministry of Health’s Tissue and Organ Transplantation Department. The Hacettepe Biobank specifically collects rare disease biosamples. The metadata for the IBG biobank is available in BBMRI catalogues and the metadata for the Hacettepe University biobank is available in EuroBioBank.

Organisation of Rare Disease Care

Centres of Expertise

In Türkiye, a national policy is currently under development for designating centres of expertise for rare disease.

Newborn Screening

Six conditions are included in Türkiye’s Newborn Screening Program: Phenylketonuria (PKU), Biotinidase Deficiency, Cystic Fibrosis (CF), Congenital Adrenal Hyperplasia (CAH), Spinal Muscular Atrophy (SMA) and Congenital Hypothyroidism. Policy decisions about which conditions to add to the newborn screening programme are made by the Ministry of Health General Directorate of Public Health. A National Committee for newborn screening is currently working on expanding the programme.

Diagnostics

There are many governmental and specialist laboratories in Türkiye that can conduct genetic tests. A list of specialist laboratories is available here. Genetic tests are reimbursed by the Social Security Agency of Türkiye. There is no particular policy or agreement in place for cross border genetic testing. Turkish clinical researchers and centres are involved in the Undiagnosed Diseases Network International (UDNI) activities and working groups. There is a policy in place to ensure national providers provide genetic counselling for patients with a suspected or confirmed rare disease.

National Alliances of Rare Disease Patient Organisations

Türkiye has a national alliance of rare disease patient organisations named Rare Disease Federation of Türkiye. Further information can be found here. Türkiye's National Alliance is involved in Türkiye's National Mirror Group and in setting the strategic direction for rare disease research. The National Alliance provides webinars and other forms of learning and capacity-building training on patient engagement in rare disease research. The National Alliance conducts analysis studies, collects data, develops projects based on the results, and hold meetings with authorities.

Türkiye's National Alliance has identified several barriers limiting patient engagement in rare disease research. These barriers include lack of awareness about research opportunities, geographical and financial difficulties, and limited communication between researchers and patient groups. Many patients also face challenges accessing research centres and are rarely involved in study design. The national alliance aims to overcome these barriers by improving communication, promoting patient education, and encouraging collaboration between researchers, institutions, and patient communities.

Information Resources for Rare Disease

National Orphanet Engagement

There is an operational Orphanet team in Türkiye, hosted by TÜSEB (Health Institutes of Türkiye). The team is financially supported by the host organisation.

Helplines

Türkiye does not have a national helpline dedicated to rare disease.

Training and Education

There are rare disease training activities available in Türkiye. The following topics are included in these training activities:

- Diagnostics
- Raising awareness of rare disease
- Standards and quality of genetics/genomics data in clinical practice and laboratories
- Registries
- Biobanks

These training activities are provided by research institutions, graduate schools, rare disease centres, the National Alliance of Rare Disease Patient Organisations, and professional medical societies.

A PhD program in Rare Disease has been developed and included in medical school curricula.

Orphan Medicinal Products (OMPs)

There is no official public registry in Türkiye listing the exact number of reimbursed OMPs. Therefore, estimates rely on the analysis of EMA-designated OMPs and their accessibility via Turkish health system channels. According to the most comprehensive studies available, of 105 orphan medicines authorised by the European Medicines Agency (EMA), as of January 2020:

- 71 were accessible in Türkiye (either licensed locally or via import mechanisms).
  - Of these, 23 were licensed and 48 were unlicensed but accessible via special access routes.
- 34 of these 71 medicines were reimbursed through the national health system:
  - 17 of the licensed OMPs were reimbursed.
  - 17 of the unlicensed OMPs (typically accessed via import) were reimbursed.

This means that approximately 34 EMA-designated OMPs were both accessible and reimbursed in Türkiye as of 2020 (Kockaya et al., 2021).

A separate 2023 industry report (AIFD & IQVIA) indicates a broader figure: 151 of 416 EMA/FDA-designated orphan drugs were accessible in Türkiye (via licensing or import), and 76 of these were locally authorised. However, this source does not confirm how many of these were reimbursed (Ertok et al., 2023).

Türkiye does not currently have a national orphan drug law. There is no domestic legal definition of ‘orphan medicinal product’ nor regulatory incentives like fee waivers, market exclusivity, or accelerated approval pathways that are common in the EU or US.

Pricing is set via international reference pricing, often leading to lower price approvals. Reimbursement decisions are made by the Social Security Institution (SGK) and reviewed by reimbursement commissions. OMPs are often evaluated under standard processes without special pathways, which may delay or hinder access.

Orphan drugs imported via the ‘Medicines Brought From Abroad’ mechanism accounted for over 80% of the total foreign drug budget in some years, despite being a minority in terms of drug volume (Atikeler et al., 2020).

Most accessible/reimbursed OMPs in Turkiye are for oncology, metabolic, and immunological conditions (ATC-L and A codes). Other areas, like neurological and dermatological orphan indications, are underrepresented (Kockaya et al., 2021).

Türkiye has multiple early/expanded access programmes that allow patients with rare conditions to obtain unlicensed or investigational medicinal products. These programmes are:

- **Named Patient Programme (NPP):**
    - Physicians can request access to unlicensed medicines for individual patients when no alternatives exist in Türkiye. These medicines are imported via the Turkish Pharmacists’ Association (TEB) and require Ministry of Health approval. This route is often used for OMPs not yet approved in Türkiye.

- **Compassionate Use Programme (CUP):**
    - This programme grants access to investigational medicines (typically those in Phase II or III clinical trials) for patients with serious or life-threatening conditions. The sponsor must provide the drug free of charge. Regulatory approval is required.

- **Foreign Drug List System:**
    - This list is maintained by TEB and includes select unlicensed medicines that can be routinely imported upon physician request. Reimbursement is sometimes possible for these drugs via the Social Security Institution under special rules.

These programmes are regulated under the ‘Regulation on the Supply of Medicines from Abroad’, last updated in February 2024, which introduced QR-code tracking and stricter controls on import processes (Ozgun et al., 2024).

Türkiye is not involved in any formal international collaborations for orphan drug access such as joint HTA, pricing, or procurement initiatives.

ERDERA
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The Data Contributing Committee of Türkiye, 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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