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Rare Diseases Worldwide and in Georgia: Screening, Diagnosis and Care Challenges

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Abstract

Rare diseases (RDs) affect fewer than one in 2,000 individuals but collectively impact over 300 million people worldwide. Despite their diversity, these conditions share common challenges, including delayed diagnosis, limited treatment options, and psychosocial burden. Ultra-rare diseases - those affecting fewer than one in 50,000 individuals - pose additional difficulties due to their extreme rarity and limited clinical expertise. Current estimates suggest that 3.5–5.9% of the global population lives with RDs, which positions these conditions as a major public health issue rather than isolated clinical rarities.

Newborn screening programs in several countries have shown that early detection of conditions such as severe combined immunodeficiency (SCID) can enable timely, life-saving interventions. Adenosine deaminase (ADA) deficiency, a genetic cause of SCID, illustrates how survival depends considerably on prompt diagnosis. While challenges remain, including gaps in screening, long delays in diagnosis, or even undiagnosed conditions, limited access to care and lack of effective treatments.

Georgia has contributed to this global effort through studies on Alport syndrome, artificial intelligence in rare bone disease diagnostics, and case reports of ultra-rare disorders. these achievements demonstrate the potential of local research to strengthen both national and international rare disease initiatives.

Keywords: rare diseases; ultra-rare diseases; newborn screening; ADA deficiency; Alport syndrome; founder mutation; novel gene; artificial intelligence diagnostics; ultrarare case; Georgia

Introduction

Rare diseases (RDs) are medical conditions that each affect a very small number of people, yet together they represent a major global health concern. The World Health Organization (WHO, 2025) defines a rare disease (RD) as a condition that typically affects fewer than one in 2,000 individuals. Current estimates suggest there are over 7,000 distinct rare diseases identified worldwide, 71.9% of which are genetic in origin and 69.9% have their onset during pediatric age group. Collectively, these disorders affect over 300 million people globally, which corresponds to 3.5–5.9% of the world's population. This prevalence indicates that while individual rare diseases are uncommon, as a group they are far from rare. Many lead to chronic disability or even early death if not recognized and treated. For families, reaching a diagnosis is often a very slow process. Studies have shown that it usually takes five to seven years from the first symptoms until the correct diagnosis is made (Nguengang Wakap et al., 2020). During this time, patients may see multiple doctors, undergo unnecessary tests, or receive incorrect treatments. These delays are harmful since they limit access to appropriate care, leaving families with uncertainty, stress and financial pressure (WHO, 2025).

Within the field of RDs, a smaller group is known as ultra-rare diseases, which affect fewer than one in 50,000 people (Harari & Humbert, 2020). Such conditions are especially difficult to diagnose and manage. Given their rarity, most physicians may never encounter affected patients in their practice. Consequently, clinical expertise is scarce, research remains limited, and effective treatments are often unavailable.

For a long time, RDs received little attention from health systems. They were seen as individual tragedies rather than as a public health issue. However, it is now understood that up to 6% of the global population may live with a rare disease. This realization has changed the way rare diseases are viewed: they are now recognized as a significant global challenge that requires organized responses from health systems (Nguengang Wakap et al., 2020; WHO, 2025).

International initiatives have helped to raise awareness. One important resource is Orphanet, created in France in 1997, which is now the world's largest database on rare diseases. It provides information on prevalence, clinical features, and available treatments, and supports both professionals and families. Policy measures have also been important. In the United States, the Orphan Drug Act of 1983 provided incentives such as tax benefits and market exclusivity to encourage pharmaceutical companies to develop treatments for rare conditions. Since then, hundreds of so-called "orphan drugs" have been developed. Similar legislation in the European Union has also supported research and drug development for rare diseases (Rath et al., 2012; Hassan et al., 2012). Even with these advances, only about 5–10% of rare diseases currently have an effective treatment. Most patients rely on supportive care, rehabilitation, or palliative services. This highlights a major gap between needs and available therapies (WHO, 2025).

As noted earlier Genetics is a key determinant in rare diseases. Advances in molecular testing, such as next-generation sequencing, have improved the ability to identify genetic causes. These tools give answers to families after years of uncertainty and sometimes allow earlier treatment. However, access to these technologies is unequal. High-income countries are increasingly able to use genetic testing in routine care, while low- and middle-income countries often lack such facilities, widening the gap in care. RDs also have important psychological and social impacts. Parents of affected children often experience guilt or blame themselves for genetic conditions. Families may feel isolated or stigmatized. Children can face challenges in school due to illness or physical differences, while adults may have difficulties in employment or independence (WHO, 2025). These challenges extend beyond medicine and require psychological, educational, and social support.

In recent years, international organizations have recognized RDs as an equity and human rights issue. Patients with rare diseases are a small population, but collectively they represent millions of people whose needs are often ignored in health planning. In 2021, the United Nations adopted a resolution emphasizing the importance of access to diagnosis, treatment, and social support for people living with rare diseases (Resolutions and Decisions Adopted by the General Assembly During Its Seventy-fifth Session: Volume I,2021). WHO has also stressed that the right to health applies equally to people with rare diseases (WHO, 2025).

One specific concept in RD genetics is founder mutations. A founder mutation is a genetic change that originated in a single ancestor and has been passed down in a particular community, often becoming more frequent if the population is small or if marriages between relatives (consanguinity) are common (Evans, 2015). This explains why some rare conditions appear at relatively high frequencies in certain regions. Studying these mutations helps researchers understand population genetics and also guides targeted screening in communities at risk.

In summary, RDs are no longer seen only as isolated medical anomalies. They represent a large and important public health challenge. Although progress has been made through databases like Orphanet, orphan drug legislation, and advances in genetics, many gaps remain. Most rare diseases still lack effective treatments, many families face long delays in diagnosis, or even remain undiagnosed and access to care continue to be unequal between countries. Recognizing RDs as both a health and equity issue is essential for creating fairer and more effective health systems. With this global context in mind, it becomes important to examine international strategies such as newborn screening and to consider how countries like Georgia can contribute to improving rare disease care and research.

Global Screening Policies and Case Example

One of the most effective ways to improve outcomes in rare diseases is through systematic newborn screening. Screening programs allow early identification of serious but treatable conditions before clinical symptoms appear, making it possible to intervene at the earliest stage of disease. This approach is particularly valuable for genetic and metabolic disorders, many of which are not visible at birth but

can cause irreversible damage if untreated. Early detection prevents severe complications, improves survival, and reduces the long-term burden on families and health systems (WHO, 2025).

Development of Newborn Screening

The concept of newborn screening began in the 1960s, when a blood test for phenylketonuria (PKU) was first introduced. PKU is a metabolic disorder that, if left untreated, results in severe intellectual disability. However, when identified early, dietary management allows normal growth and development. The success of PKU screening was a landmark in public health, showing that population-based testing for rare diseases could change the natural history of an otherwise severe genetic disorder (Screening for phenylketonuria US Preventive Services Task Force, 2008). Over the following decades, many countries adopted newborn screening for PKU and gradually expanded their screening panels to include other genetic and metabolic disorders.

Examples from High-Income Countries

The United States provides one of the best-documented examples of how newborn screening can transform care. A national framework now ensures that all states conduct newborn screening, although panels vary slightly. A landmark study across 11 U.S. programs demonstrated that severe combined immunodeficiency (SCID) can be reliably detected at birth using T-cell receptor excision circle (TREC) assays. Early identification of SCID allowed infants to receive curative therapies such as hematopoietic stem cell transplantation (HSCT) before life-threatening infections developed (Kwan et al., 2014; Hassan et al,. 2012).

In Sweden, a three-year pilot project tested the feasibility of screening for primary immunodeficiencies using a triplex assay measuring T-cell receptor excision circles (TREC), kappa-deleting recombination excision circles (KREC), and the beta-actin gene (ACTB). This approach enabled early recognition of infants at risk for severe immune deficiencies within days of birth. The pilot not only confirmed the accuracy of the method but also demonstrated that molecular techniques could be successfully integrated into a national health system. These experiences illustrate the effectiveness of newborn screening in preventing mortality and disability. They also highlight the cost-effectiveness of early detection: although screening requires upfront investment, it reduces the need for intensive hospital care, long-term disability support, and expensive emergency interventions (Zetterström et al., 2017).

Challenges in Low- and Middle-Income Countries

In contrast, many low- and middle-income countries (LMICs) face major barriers to implementing newborn screening. Health systems often lack laboratory infrastructure, trained personnel, and financial resources. Competing health priorities, such as infectious diseases, maternal health, and basic primary care, may overshadow investment in rare disease screening. In many cases, only a few conditions are screened, and in some countries no systematic screening exists at all. The absence of newborn screening contributes to the “diagnostic gap” that characterizes rare diseases worldwide.

Families may spend years visiting multiple doctors, undergoing repeated tests, and seeking answers without success. In the meantime, irreversible damage may occur, and opportunities for early intervention are lost. For metabolic disorders or immunodeficiencies, the critical window for treatment may pass before diagnosis, with tragic consequences for patients and families **(WHO, 2025).**

Adenosine Deaminase Deficiency as a Case Example

Adenosine deaminase (ADA) deficiency is a genetic cause of SCID and provides a clear example of why newborn screening matters. Infants with ADA deficiency are born without functional immune systems and are extremely vulnerable to infections. Without treatment, most die in early childhood. However, when the condition is detected early, outcomes are markedly improved. Research shows that infants diagnosed at birth and treated with HSCT have significantly higher survival rates compared with those diagnosed after clinical symptoms appear. Additional treatment options such as enzyme replacement therapy and gene therapy have also become available, but these are most effective when started in infancy (Hassan et al., 2012; Hershfield, 2024). ADA deficiency thus demonstrates how newborn screening can turn a fatal disease into a manageable condition, saving lives and reducing costs for health systems.

Policy Lessons

The experiences of PKU, SCID, and ADA deficiency provide several important lessons for policymakers worldwide:

1. **Equity in health care:** All children should have the opportunity to be screened for serious but treatable conditions, regardless of where they are born (WHO, 2025).
2. **Cost-effectiveness:** Although newborn screening requires investment, it prevents disability and long-term complications, reducing healthcare expenditures (Kwan et al., 2014).
3. **Integration with genetics:** As genomic technologies become more affordable, screening panels can expand to cover more conditions, improving early detection and treatment opportunities (Hershfield, 2024).
4. **Global cooperation:** sharing data and successful strategies, helps countries with limited screening programs to implement them more quickly and effectively, which reduces differences in access to care (Nguengang Wakap et al., 2020).

Lessons for Georgia

For countries like Georgia, where systematic newborn screening is not yet established, the international evidence offers clear guidance. Even introducing a small panel of conditions could prevent severe outcomes and demonstrate the value of early detection. Georgia has shown strong participation in research on rare diseases, yet integrating this knowledge into routine healthcare

practice remains challenging. Building laboratory infrastructure, training specialists, and piloting screening programs are realistic first steps. In the long term, Georgia could also benefit from regional cooperation with neighboring countries, sharing data and expertise to establish cost-effective screening systems across the Caucasus.

Rare Disease Research in Georgia

Although Georgia is a small country with limited health system resources, its scientists have contributed important findings to the international literature on RDs. These contributions are valuable because data from the Caucasus region remain scarce, and documenting population-specific genetic variants provides insights both for local patients and for the global medical community. Recent studies highlight Georgia's role in describing founder mutations, participating in international collaborations, and reporting ultra-rare conditions.

Alport Syndrome and a Founder Mutation

Alport syndrome is a hereditary kidney disease caused by pathogenic variants in type IV collagen genes, mainly *COL4A3, COL4A4,* and *COL4A5*. It is characterized by persistent hematuria, progressive kidney dysfunction, hearing loss, and ocular abnormalities. The clinical course is highly variable: in some patients, symptoms remain mild and limited to hematuria, while in others the disease progresses to end-stage renal failure during adolescence or early adulthood (Adone & Anjankar, 2023). Due to its variable clinical presentation and the need for genetic testing for confirmation, Alport syndrome is often underdiagnosed in regions with limited diagnostic resources (Nguengang Wakap et al., 2020).

In 2023, Tkemaladze and colleagues reported on three ethnically Azerbaijani, consanguineous families from Algeti village in the Marneuli region of Georgia. Genetic analysis revealed a *COL4A3 c.765G>A* variant in affected individuals (Tkemaladze et al., 2023). The authors suggested that this represents a founder mutation - a genetic change that originated in a common ancestor and became more frequent in the community because of limited genetic mixing (Evans, 2015).

Identifying a founder mutation has both scientific and public health significance. Scientifically, it adds to the global classification of *COL4A3* variants, contributing to knowledge of genotype–phenotype relationships in Alport syndrome. From a public health perspective, it shows that certain communities may be at higher risk and would benefit from targeted genetic counseling and testing. The study recommended offering genetic testing to residents of Algeti with persistent hematuria, which could allow earlier diagnosis, preventive monitoring, and family counseling. Beyond its immediate findings, this study demonstrates how careful genetic research in Georgia can uncover population-specific patterns that would otherwise remain hidden. For a small country, such contributions are important not only for local healthcare but also for the global literature on hereditary kidney disease.

Artificial Intelligence in Rare Bone Disease Diagnostics

Georgian scientists are also engaging in international collaborations that explore innovative technologies for RD care. In 2025, Javanmardi et al. conducted one of the first global surveys on the use of artificial intelligence (AI) in rare bone disease (RBD) diagnostics. The study included 103 participants from 27 countries, with Georgian researchers contributing as co-authors. Most respondents were physicians (89%), primarily from academic medical centers (81%). The main specialties represented were medical genetics, pediatrics, and endocrinology, together accounting for 69% of participants. The survey revealed strong recognition of the importance of imaging: 91% of respondents rated imaging as "very" or "extremely important" in diagnosing rare bone diseases, and 81% identified X-rays as the most critical tool. When asked about integrating AI, 81% of participants reported being somewhat or extremely likely to adopt image-recognition software into their clinical practice. This reflects optimism about the ability of AI to support clinicians by improving accuracy and reducing diagnostic delays. However, respondents also highlighted barriers, including limited datasets for training AI, lack of transparency in algorithms, and challenges integrating AI tools into routine clinical workflows (Javanmardi et al., 2025).

For Georgia, participation in this global survey is highly significant. It demonstrates that Georgian researchers are not isolated from international innovation but are actively contributing to debates about the future of medicine. This involvement shows that even countries with constrained resources can participate in shaping the development of cutting-edge technologies, ensuring that local perspectives are represented in global research agendas.

Complex Glycerol Kinase Deficiency Case Report

Another contribution from Georgia is the documentation of ultra-rare disorders through clinical case reports. In 2025, Bregvadze and colleagues described two male siblings with complex glycerol kinase deficiency (CGKD), an extremely rare X-linked metabolic disorder. CGKD is caused by deletions in the Xp21 chromosomal region that often extend across several genes, leading to a broad and variable clinical phenotype (Bregvadze et al., 2025). The affected brothers presented with nonspecific features, including developmental delay and metabolic disturbances, which could easily have been misinterpreted as more common conditions. Only genetic testing clarified the diagnosis, confirming CGKD. The report highlighted that in ultra-rare conditions, molecular diagnostics are essential for reaching a correct diagnosis, since clinical signs alone are not sufficient.

Although based on only two patients, this case report contributes important information to the very limited global literature on CGKD. For ultra-rare conditions where only a handful of cases are known worldwide, each report adds knowledge that can guide future diagnosis and management. Such

publications also demonstrate the value of clinicians in Georgia systematically documenting and publishing their experiences, thereby contributing to international databases and medical education.

Together, these three studies illustrate the diverse ways in which Georgian researchers are advancing RD knowledge. These contributions highlight that despite limited resources, Georgian researchers are actively engaging with both local and global aspects of RD research. They provide data relevant for international science while also showing the challenges and opportunities faced in a lower-middle–income setting.

Discussion

RDs collectively affect millions of people worldwide and represent a growing challenge for health systems. While each condition is individually uncommon, their combined prevalence is significant, and they present unique challenges in diagnosis, treatment, and social support (WHO, 2025; Nguengang Wakap et al., 2020). International experiences show that progress is possible, but gaps remain, particularly in LMICs. This section discusses global lessons, ethical and equity considerations, and the specific situation in Georgia, before outlining opportunities for improvement and regional cooperation.

Global Lessons

Experiences from high-income countries demonstrate that systematic approaches can transform outcomes in rare diseases. The introduction of newborn screening for phenylketonuria (PKU) in the 1960s provided the first example of how early detection could prevent severe disability (European Respiratory Review, 2022). Since then, newborn screening has expanded to include many conditions, particularly genetic and metabolic disorders. The United States has shown that population-based screening for severe combined immunodeficiency (SCID) can identify affected infants at birth, allowing timely interventions such as hematopoietic stem cell transplantation (HSCT) before infections occur (Kwan et al., 2014). Similarly, the Swedish pilot project demonstrated that molecular techniques, including triplex assays for immune deficiencies, can be successfully integrated into national health systems (Zetterström et al., 2017).

The case of adenosine deaminase (ADA) deficiency, a genetic cause of SCID, highlights the importance of early detection. Studies have shown that survival improves significantly when infants are diagnosed through screening and treated promptly (Hassan et al., 2012; Hershfield, 2024). These examples demonstrate that early intervention not only saves lives but also reduces the long-term costs of care.

Ethical and Equity Considerations

Rare diseases are increasingly recognized as an equity and human rights issue. Equity in health means that all individuals should have fair access to timely diagnosis and appropriate care, regardless of how common or rare their condition is. However, patients with rare diseases often experience inequities: long diagnostic delays, lack of specialized care, and limited availability of effective therapies (WHO, 2025). From a human rights perspective, the right to health applies equally to people with rare conditions. The United Nations resolution on rare diseases, adopted in 2021, emphasized the importance of access to diagnosis, treatment, and social support for all affected individuals ("Resolutions and decisions adopted by the General Assembly during its seventy-fifth session: Volume I," 2021). This framing highlights that neglecting rare disease patients is not only a medical issue but also a violation of fairness and justice.

Cost and affordability also raise ethical questions. Many orphan drugs are extremely expensive, sometimes costing hundreds of thousands of dollars per year. Governments must decide whether to fund these treatments, balancing individual patient needs with the allocation of limited healthcare resources. This dilemma illustrates how rare diseases intersect with broader debates about distributive justice in health policy (Nguengang Wakap et al., 2020).

Lessons for Georgia

Against this global background, Georgia illustrates both the opportunities and the challenges of addressing rare diseases in LMICs. The country currently lacks a national newborn screening program, meaning that treatable conditions such as ADA-SCID may go undiagnosed until symptoms appear (Kwan et al., 2014). Genetic testing facilities are limited and often unaffordable for families, creating barriers to early detection and management. Financial hardship is a recurrent theme, as parents may need to travel abroad for testing or treatment, and therapies may be interrupted when costs cannot be sustained (WHO, 2025).

At the same time, Georgia has produced valuable scientific contributions. The identification of a founder *COL4A3* mutation in Alport syndrome highlights how local research can uncover population-specific risks and inform targeted interventions. Participation in international collaborations on artificial intelligence in rare bone disease diagnostics shows that Georgian researchers are integrated into global scientific networks. Case reports such as the documentation of complex glycerol kinase deficiency add unique data to the global literature.

Opportunities for Improvement

Several steps could strengthen Georgia's approach to RDs: National registry, Pilot newborn screening programs, Expanded access to genetic testing, Financial support for families, Public and professional awareness.

Because Georgia is a small country, regional collaboration offers an efficient way to strengthen rare disease care. Neighboring countries such as Armenia and Azerbaijan face similar challenges, and shared initiatives could reduce costs and expand expertise. Joint registries, training programs, and laboratory networks would provide economies of scale. The European Reference Networks provide a successful model, showing how countries can pool resources for rare conditions. A Caucasus rare disease network could help bridge gaps and connect the region to broader European and global initiatives.

Conclusion

RDs may be individually rare, but Collectively they represent a major global health concern. International experience demonstrates that newborn screening, genetic testing, and orphan drug policies can significantly improve outcomes, yet inequities remain, particularly in LMICs. Recognizing rare diseases as an equity and human rights issue underscores the need for fair access to diagnosis, treatment, and support for all patients (WHO, 2025).

Georgia illustrates both the promise and the challenge of RD research and care. Its researchers have made notable contributions to international knowledge, identifying founder mutations, engaging in global AI collaborations, and documenting ultra-rare conditions. Yet systemic barriers - the absence of newborn screening, limited access to genetic testing, and financial hardship - mean that many patients still lack timely care.

Moving forward, Georgia could strengthen its RD response by establishing registries, piloting screening programs, expanding access to diagnostics, and collaborating regionally. With these steps, the country has the potential not only to improve the lives of its own patients but also to serve as a model for how smaller nations with limited resources can contribute meaningfully to global RD research and public health equity.

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იშვიათი დაავადებები მსოფლიოში და საქართველოში: სკრინინგი, დიაგნოსტიკა და მართვის გამოწვევები

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რეზიუმე

იშვიათი დაავადება არის მდგომარეობა, რომელიც ყოველი 2 000 ინდივიდიდან 1- ზე ნაკლებს უვლინდება და ჯამურად 300 მილიონზე მეტ ადამიანს აზიანებს მსოფლიოში. განსხვავებულობის მიუხედავად, მათ გააჩნიათ საერთო პრობლემები: დაგვიანებული და გადადებული დიაგნოსტიკა, შეზღუდული წვდომა მკურნალობაზე და მნიშვნელოვანი ფსიქოსოციალური და ფინანსური ტვირთი ოჯახებისთვის. ულტრაიშვიათი დაავადებები, რომლებიც ყოველი 50 000 ინდივიდიდან 1 - ზე ნაკლებში ვლინდება, კიდევ უფრო დიდი გამოწვევაა კლინიკური გამოცდილებისა და დიაგნოსტიკური შესაძლებლობების ნაკლებობის გამო. მსოფლიოს მოსახლეობის 3.5–5.9%-ს იშვიათი დაავადება აქვს, რაც არა მხოლოდ ცალკეული იშვიათი სამედიცინო შემთხვევაა, არამედ მნიშვნელოვან საზოგადოებრივი ჯანმრთელობის პრობლემას წარმოადგენს.

ახალშობილთა სკრინინგი ერთ-ერთი ყველაზე ეფექტური სტრატეგიაა შედეგების გასაუმჯობესებლად. მოწინავე ქვეყნებში ჩატარებულმა სკრინინგმა დროულად გამოავლინა მძიმე კომბინირებული იმუნოდეფიციტი (SCID) და ადენოზინდეზამინაზას (ADA) დეფიციტი, რაც ახალშობილებში სასიცოცხლოდ მნიშვნელოვანი მკურნალობის დაწყების საშუალებას იძლევა, ეს კი ცხადყოფს რომ ადრეული დიაგნოზი განაპირობებს სიცოცხლის გადარჩენას. მიღწევების მიუხედავად კვლავ არსებობს გამოწვევები როგორიცაა როგორც დაგვიანებული, გადადებული დიაგნოსტიკა, ასევე დიაგნოზის გარეშე არსებული დაავადებები, შეზღუდვები სკრინინგში, მკურნალობასა და მართვაში.

ქართველმა მეცნიერებმა მნიშვნელოვანი წვლილი შეიტანეს გლობალურ ძალისხმევაში, ალპორტის სინდრომის, მეტაბოლურ დისფუნქციასთან ასოცირებული ულტრა იშვიათი შემთხვევის კვლევებით და იშვიათი ძვლის დაავადებების დიაგნოსტიკაში ხელოვნური ინტელექტის (AI) გამოყენებასთან დაკავშირებით საერთაშორისო კვლევაში მონაწილეობის მიღებით. მიუხედავად არსებული სიძნელეებისა სკრინინგსა და დიაგნოსტიკაში და მართვაში, მიღწევები ცხადყოფს ადგილობრივი კვლევების პოტენციალს, რომლებიც აძლიერებს როგორც ლოკალურ ასევე საერთაშორისო ინიციატივებს იშვიათი დაავადებების სფეროში.

საკვანძო სიტყვები: იშვიათი დაავადებები; ულტრაიშვიათი დაავადებები; ახალშობილთა სკრინინგი; ადენოზინ დეამინაზას დეფიციტი; ალპორტის სინდრომი; ფაუნდერის მუტაცია; ახალი გენი; ხელოვნური ინტელექტი დიაგნოსტიკაში; ულტრაიშვიათი შემთხვევა; საქართველო