RARE DISEASES FEDERATION

Rare Lives and Rare Diseases Workshop

25 March 2026 Bilkent Hotel - Ankara / TÜRKİYE

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WORKSHOP REPORT

FOREWORD

Deniz ATAKAY
President, Rare Diseases Federation

Awareness, Solidarity, and Shared Responsibility in Rare Diseases

We are gathered here today in this closed meeting to discuss rare lives and rare diseases. This issue creates a common ground that brings us together. With this in mind, we have always sought to establish an open, sincere, and safe environment for communication, much like that of a family. Through this workshop, we also wanted to prepare for the major Summit that will take place tomorrow.

We wanted to discuss together what kind of approach we should adopt when it comes to rare diseases: Should we turn away, or should we reach out a helping hand? What can we do to help? As the Rare Diseases Federation, we believe it is important to evaluate all of these questions with you in a sincere and constructive manner.

I would like to thank each and every one of you for being here today, recognizing that many of the challenges we face in life are, in fact, shared challenges, and for valuing this process and approaching it with the intention of developing solutions.

We may also regard today as the beginning of a long and meaningful journey. Because diseases can at times be unsettling and distressing, they may lead some people to distance themselves. There are those who are reluctant to engage because they mistakenly believe such conditions may be contagious, and others who prefer to avoid the difficulties associated with the process. As a result, individuals living with rare diseases can sometimes find themselves excluded from everyday life.

Our primary objective, however, is to provide a healthier future for the children of our country, increase public awareness, and establish a strong foundation for healthier generations. We are only at the beginning of this journey, which is why your presence here is extremely valuable to us.

Today's workshop will be moderated by Gülnur Gökmen on behalf of the Federation and by Prof. Dr. Zafer Çalışkan, Faculty Member at Hacettepe University, representing the academic community.

On this occasion, I would also ask for your understanding regarding my excitement. It is, however, a very appropriate excitement, because together we are undertaking a great responsibility. While it may be possible, in one sense, to cope with our own disease stories, the responsibility we assume takes on a far greater meaning when it comes to benefiting others and helping illuminate their path.

Representatives of the relevant ministries, non-governmental organizations, and service institutions have come together around the same table, and important observations have been made. Once again, I would like to thank all of you for your participation and valuable contributions, and I hope that the outcomes of this workshop will contribute to a productive and meaningful process.

Prof. Dr. Zafer ÇALIŞKAN
Faculty Member, Hacettepe University

Rare Diseases: A Challenge for Healthcare Systems and a Multidimensional Reality

Ms. Deniz began her opening remarks quite modestly by describing this gathering as "the beginning of a journey." However, when we look at the history of the Association and the Rare Diseases Federation, it would be more accurate to describe this not as a beginning, but as an important milestone. Significant progress has already been made. Bringing together different stakeholders and filling a hall such as this requires considerable effort, strong relationships, and a high level of dedication. For this reason, the point we have reached today should be regarded not as a beginning, but as a valuable stage of progress.

I would also like to briefly explain why I am here today. We will be following a moderation format that differs from the conventional approach. This format may be unfamiliar to some of you, but your participation throughout the process will be extremely valuable. In this regard, we plan to proceed through a set of specific questions.

For many years, I have worked in the fields of health economics and pharmaceutical economics. I have undertaken various roles in universities, public institutions, the Social Security Institution, the Ministry of Health, and several non-governmental organizations. More recently, rather than focusing solely on financing and expenditure, I have been involved in initiatives aimed at approaching healthcare services from a more holistic perspective. For this reason, I am pleased to be here today.

To provide a brief framework, I would like to touch on both the situation in Türkiye and developments around the world. At first glance, the term "rare diseases" may suggest a limited population. However, despite their low prevalence, when their different dimensions and particularly their often unseen impact on healthcare systems are taken into consideration, it becomes clear that their sphere of influence is much broader.

Today, for many countries, rare diseases represent a test of healthcare systems in terms of inclusiveness, access to treatment, financing, and equity. Therefore, this issue should not be viewed merely as a matter affecting a small number of individuals. While these diseases are rare when considered individually, collectively they affect a substantial population. In this respect, the term "rare" can be misleading when considering their cumulative impact.

Türkiye, in particular, faces certain distinct dynamics. Lifestyle factors and the relatively high rate of consanguineous marriages contribute to a higher prevalence of genetically based rare diseases. This increases Türkiye's responsibility in this field. I do not use the term "burden" in a negative sense, because although rare diseases are often discussed in terms of access challenges, financing difficulties, or shortcomings, the issue extends far beyond these dimensions.

When rare diseases are considered together with patients, their families, and caregivers, they cease to be solely a matter of treatment access or cost. We are dealing with a multilayered issue that has social, economic, and psychological dimensions. For example, delays in diagnosis are very common. Research shows that reaching an accurate diagnosis can take six to seven years. During this period, serious consequences such as loss of function, cognitive decline, and reduced treatment effectiveness may occur.

This demonstrates that the issue is not confined to the healthcare system alone; it also encompasses education, social services, and caregiving processes. Rare diseases are often life-threatening conditions that significantly reduce quality of life and, in some cases, result in death. Consequently, caregivers and families become an integral part of the process, and the overall impact expands to a much broader societal level.

The fact that many rare diseases emerge during childhood can lead to educational setbacks, difficulties in participating in economic life, and social isolation. For this reason, rare diseases cannot be considered solely under the headings of treatment and cost. They must be addressed as a multidimensional issue involving all stakeholders. Today's event, as well as the panels and presentations that will take place during tomorrow's Summit, are among the most tangible demonstrations of this approach.

One of the areas on which I focus particularly is access to treatment and financing. Following diagnosis, the diversity of treatment options, drug development processes, product launches, pricing, and reimbursement mechanisms create a highly complex structure. Therefore, even when other conditions are met, various challenges in accessing treatment may still arise. At this point, accurately identifying needs is of critical importance.

At the same time, the societal, psychological, and social impacts must not be overlooked. If time permits, we may have the opportunity to discuss these issues in greater detail with specific reference to Türkiye. However, in general terms, it should be emphasized that, rather than focusing solely on the challenges in the field of rare diseases, it is possible to develop solutions through new approaches and alternative methods.

Just as major and comprehensive transformations are important, advancing through small yet effective steps is equally valuable. I believe that everyone in this room can contribute to this process. This is because the issue cannot be resolved solely within the boundaries of the healthcare system; it must be addressed together with its social and economic dimensions.

This multidimensional structure is also clearly evident in Türkiye. In light of all these considerations, it is possible to say that considerable progress has been made. The level of participation here today is one of the strongest indicators of that progress.

On this occasion, I would once again like to thank everyone who has contributed to this effort.

Gülnur GÖKMEN
Rare Diseases Federation

Our Future Depends on What We Do Today

Today, through the journey we have embarked upon together with the Federation, we are actually realizing an important dream. We have carried out numerous initiatives to date and have knocked on the doors of all relevant institutions, particularly the Ministry of Health, the Ministry of National Education, and the Ministry of Family and Social Services. However, our consistent aspiration has been to establish a structure in which all stakeholders can come together around the same table and develop a shared understanding. The fact that such an environment has been created today is extremely valuable and gratifying for us. In this respect, I can say that an important dream has become a reality.

Although our Federation was established approximately two years ago, active efforts had been underway long before that. To the question of whether such a structure is needed in Türkiye, we can answer with a clear "yes." When considered within a framework that also includes individuals living with rare and ultra-rare diseases, it is evident that an umbrella organization is needed to advocate for their rights and strengthen their access to treatment.

The support provided by the Ministry of Health throughout this process has been extremely valuable. Within the scope of efforts carried out in the field of rare diseases, needs assessments and accurate information gathered from the field have been shared with the Ministry, and many processes—particularly the National Action Plan—have been advanced through close collaboration. Today, an important step has been taken to further strengthen this cooperation. In the coming period, we aim to advance this process further through collaboration with the Ministry of Family and Social Services and the Ministry of National Education.

Rare diseases cannot be addressed solely through the concept of "patients." These individuals are people engaged in a lifelong struggle, and together with their families they form an inseparable whole. Accordingly, this field extends beyond health policies alone and encompasses a multidimensional structure that includes social services, education, and all other relevant stakeholders. These individuals are part of everyday life; they have educational journeys, social lives, and, in time, working lives as well.

As a Federation, our approach is not merely to voice problems but to be an active part of the solution process. We believe that significant progress has been made in this regard through our collaboration with the Ministry of Health.

In the coming period, we aim to continue strengthening our cooperation with other ministries as well.

The fundamental principle we adopted at the outset has been this: "Our future depends on what we do today." The steps taken may sometimes appear small or insufficient; however, we believe that the step taken here today is extremely valuable. Together, we will shape the future.

In particular, for individuals living with rare diseases that remain largely invisible and insufficiently understood, we carry out activities through national and international collaborations in line with the United Nations Convention on the Rights of the Child and the Convention on the Rights of Persons with Disabilities. Our primary objective is to undertake initiatives that serve the best interests of these individuals. Our ultimate goal is to contribute to building a future in which healthier generations are raised despite the reality of rare diseases that exists today. Ensuring an equal and fair right to life in all areas of society constitutes our fundamental vision.

Across Türkiye, we conduct efforts to collect accurate data from the field and organize training programs, meetings, and various analyses. Within this scope, a study we previously conducted examined out-of-pocket healthcare expenditures incurred by individuals with rare diseases and their families. The findings revealed that the healthcare expenses of these families are considerably higher than those of other families.

At this point, I would particularly like to emphasize the following: attention is generally focused on families' income levels, while their expenditure levels are not given sufficient consideration. However, when it comes to rare diseases, families face a substantial economic burden. These findings are shared with the relevant institutions, and solution processes are carried out collaboratively. As of today, we aim to strengthen these partnerships even further.

Our areas of work are quite extensive. Our primary focus areas include genetic screening, newborn screening and early diagnosis, diagnostic processes, access to treatment and medicines, organ donation, invisible disabilities, education, discrimination, social participation, and dietary management. We have broad and active working groups dedicated to each of these topics.

In brief, this is how I would summarize the Federation. Building on the work we have carried out to date and the contributions that have been made, we will continue moving forward by contributing more strongly to policy development processes from this point onward.

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SURVEY RESULTS AND DISCUSSIONS

Moderator - Prof. Dr. Zafer ÇALIŞKAN

Faculty Member, Hacettepe University

As in many other fields, particularly in the social and healthcare sectors, the role of civil society organizations is no longer limited to patient or stakeholder advocacy. Today, these organizations have become important actors that identify real needs and demands, facilitate effective communication among stakeholders, and ensure that data gathered from the field is reflected in decision-making processes. The study findings shared a short while ago are a concrete example of this transformation.

In this context, it is also important to emphasize the point that the Rare Diseases Federation has reached and the progress it has made.

In today's workshop, a more interactive approach will be adopted, moving beyond the traditional presentation format. Within this framework, participants' views will be gathered through a pre-prepared questionnaire consisting of eight questions, and an interactive environment for discussion and evaluation will be created based on the results obtained.

The objective is to develop a shared understanding through the contributions of different stakeholders and to bring a multidimensional perspective to policy development processes in the field of rare diseases.

RARE DISEASES: COORDINATION AND AWARENESS ARE PERCEIVED AS THE PRIMARY CHALLENGES

A total of 50% of participants stated that the most critical challenges within the rare disease ecosystem are the lack of coordination (25%) and the lack of awareness (25%). This equal distribution demonstrates that there is an equally strong need both for the integration of operational processes in rare disease management and for increasing levels of social and academic awareness.

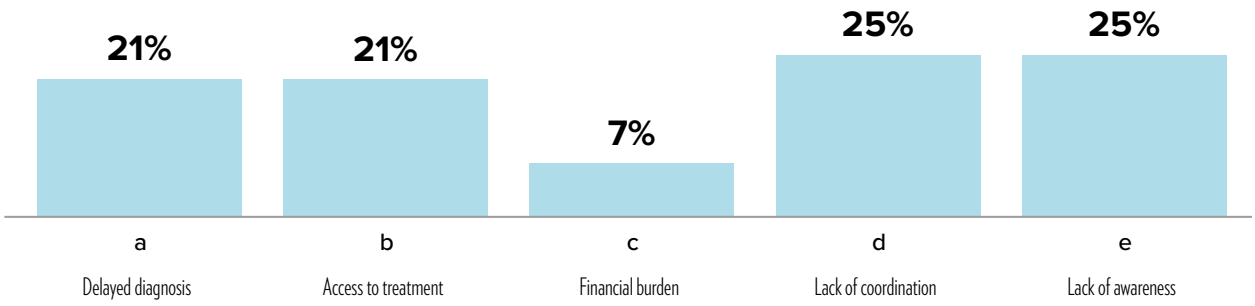
The 42% of participants who identified diagnostic and treatment processes as the main barriers distributed their responses equally between delayed diagnosis (21%) and access to treatment (21%). This picture indicates that structural improvements are needed at both stages of the patient journey, from entering the system through diagnosis to achieving outcomes through treatment.

The 7% who considered the financial burden to be the most critical issue suggests that, alongside other operational and clinical bottlenecks, cost-related concerns are perceived by participants as a secondary priority.

When the overall distribution is evaluated, it becomes evident that the main challenges in the field of rare diseases are concentrated not directly in medical processes, but rather in broader systemic areas such as the functioning of the system (coordination) and its visibility (awareness).

1. In your opinion, what is the most critical challenge in rare diseases?

a. Delayed diagnosis
b. Access to treatment
c. Financial burden
d. Lack of coordination
e. Lack of awareness



Key Problem Areas in Rare Diseases and the Need for Coordination

Moderator - Gülnur GÖKMEN

Rare Diseases Federation

Conceptual Approach and Terminology Shift

It was stated that the phenomenon of rare diseases should be viewed not as a "problem" but rather as a "field of work" that requires a strategic approach. Although the issue was reflected in this manner due to the format of the question, the importance of evaluating the subject as a structural process by all stakeholders was emphasized.

Participant Perspectives and Analysis of the Current Situation

In an environment where all stakeholders were represented, the survey results showed that "lack of coordination" and "lack of awareness" emerged as the most prominent issues, receiving the highest response rates. It was noted that issues such as financial burden, access to treatment, and delayed diagnosis are topics whose causes and origins are already known and widely recognized within the sector; therefore, they received relatively lower response rates.

The Hidden Part of the Iceberg: Strategic Priorities

It was stated that beyond visible issues such as financing and access to treatment, the rare disease ecosystem encompasses a broader dimension that represents the hidden part of the iceberg. In particular, the high response rates attributed to the lack of coordination and lack of awareness were identified as critical indicators for the steps to be taken in Türkiye.

The Search for a Model for Türkiye and the Starting Point

It was stated that the objective is to discuss a new model for rare diseases in Türkiye and to examine all existing shortcomings within this process. Despite the complexity and difficulty of the issue, it was concluded that the fundamental starting point of any strategic model to be developed should be coordination.

Holistic Well-Being and Multisectoral Coordination in Rare Diseases

Specialist Dr. Hüseyin ÖRÜN */ Association of Public Health Specialists*

The Scope and Continuity of Health

It was emphasized that the concept of health represents not only a physical condition but also a state of complete psychological and social well-being. It was noted that this state of well-being should not be confined to the hospital setting but should be sustained across all areas of life, including school, work, and social environments. It was further stated that the holistic well-being of caregivers, as well as that of patients, should be protected.

Quality of Life and the Limits of Healthcare Services

It was stated that, particularly in the context of rare diseases, the provision of healthcare services alone is not sufficient to achieve a high quality of life. In this regard, addressing the lack of coordination was identified as one of the most critical requirements for ensuring a high quality of life.

Development Plans and Institutional Strategies

It was recalled that national development plans are prepared under the coordination of the Presidency and that each ministry develops action plans aligned with these overarching strategies. It was emphasized that rare diseases constitute an interdisciplinary issue that falls within the shared responsibility of multiple ministries within this planning framework.

Strategic Collaboration and Future Outlook

It was concluded that the current working group and the meeting itself represent an important and productive starting point for ensuring coordination in the field of rare diseases, owing to their multi-stakeholder structure.

Diagnostic Uncertainty and the Need for Multidisciplinary Coordination in the Management of Rare Diseases

Assoc. Prof. Dr. Merve Deniz PAK GÜRE */ Department of Social Work, Başkent University*

Chronic Diagnostic Uncertainty and Life Challenges

It was highlighted that, despite many years of academic work in the field of rare diseases, there are cases in which the diagnostic process has remained unresolved for as long as 35 years at the individual level. It was stated that this state of uncertainty, characterized by constantly changing diagnoses, repeated genetic screenings, and trials of different medications, makes disease management extraordinarily difficult.

Disruptions in Follow-up Processes Due to Physician Mobility

It was stated that access to healthcare services is adversely affected when experienced healthcare professionals leave the public sector and move into private practice, resulting in significant disruptions in patient follow-up. It was emphasized that when a physician who has followed a patient for years leaves the system, the process often has to start over from the beginning, disrupting continuity of care and creating a substantial financial burden for patients.

Lack of Central Coordination and Guidance in Service Delivery

It was noted that the absence of a single coordinating authority or a "case manager" within the healthcare system leaves patients without adequate guidance as they navigate between different specialties. Within this fragmented structure, where one specialist refers the patient to another, it was observed that the patient ultimately becomes the only actor attempting to manage and solve the process.

Sectoral Integration and Regional Inequalities

It was stated that effective coordination does not exist between healthcare services, social services, and mental health services. It was further noted that even individuals who benefit from academic resources and the advantages of living in major metropolitan areas often struggle to find their way through the system, while regional inequalities further exacerbate the problem. It was concluded that resolving this complex situation requires the entire process to be managed through a single coordination center based on a holistic approach.

The Need for Institutional Leadership and Centralized Structures in the Management of Rare Diseases

Moderator

The "Conductor" Model in Process Management

It was stated that, given the complex nature of rare diseases, there is an absolute need for an institutional structure or governing body capable of managing the process with the precision of a "conductor," ensuring coordination and harmony among all stakeholders. It was emphasized that, under current circumstances, civil society organizations and federations undertake critical responsibilities in this area; however, carrying such a significant burden solely through voluntary organizations or individual efforts is not sustainable.

Data-Driven Management and Registration and Coding Systems

It was stated that deficiencies in data management lie at the core of many of the challenges encountered in diagnosis and follow-up processes. It was further noted that the establishment of an effective registration and coding system would enable patients to be directed to the appropriate centers, prevent duplicate applications, and minimize the time spent navigating the healthcare system.

Financial Sustainability and Public Responsibility

Attention was drawn to the fact that the substantial costs associated with rare diseases are beyond the capacity of individuals to bear on their own. It was stated that, in order to manage the financial burden arising from both treatment and follow-up processes, the issue must be addressed through stronger commitment and a centralized public policy approach.

Systemic Solutions and a Centralized Approach

It was observed that fragmented service delivery leaves patients isolated throughout the process, and it was concluded that a solution can only be achieved through a holistic system coordinated by a central authority.

Care Models, Health Migration, and the Need for Integrated Management in Rare Diseases

Assoc. Prof. Dr. Merve Deniz PAK GÜRE */ Department of Social Work, Başkent University*

Accumulated Literature and Persistent Chronic Challenges

It was stated that, since the publication in 2019 of one of the first academic studies in Türkiye analyzing different care models and existing challenges, the fundamental problems have continued to persist. Despite the years that have passed, patients continue to face the same structural difficulties within similar cycles, and the pace of improvement has remained below expectations.

Service Integration and the "Flagship" Model

It was emphasized that the full integration of healthcare and social services is essential for the effective management of rare diseases. The need for a physician-led model capable of assuming responsibility and centrally managing both financing and treatment processes, much like a "flagship," was highlighted. It was suggested that such a model could help minimize delays and bureaucratic obstacles, particularly in consultation processes.

Regional Inequalities and Health Migration

The phenomenon of "health migration," characterized by the movement of patients from rural areas and smaller provinces to major cities as a result of inequalities in access to healthcare services, was identified as a significant challenge. It was concluded that this migration creates socio-economic pressures on patients and their families while also increasing the workload of major healthcare centers, and therefore should be specifically addressed within strategic planning processes.

The Importance of Process Management and Strategic Coordination in Rare Diseases

Moderator

Holistic Evaluation and a Methodological Approach

It was emphasized that the phenomenon of rare diseases requires a holistic evaluation and systematic process management rather than fragmented solutions. This approach was identified as a fundamental requirement for addressing disruptions throughout the patient journey.

The Critical Role of Coordination

It was emphasized that the discussions held and the views expressed once again confirmed that "coordination" is the most critical success factor in this field. It was further noted that initiatives undertaken without the establishment of an effective coordination mechanism are unlikely to achieve operational efficiency.

The Economics of Early Diagnosis in Rare Diseases and the Need for Strategic Referral Mechanisms

Prof. Dr. Fatih ÖZALTIN */ Director, Hacettepe University Center for Genome Sciences and Rare Diseases Research and Application – Faculty Member, Division of Pediatric Nephrology, Hacettepe University*

Clinical and Irreversible Consequences of Delayed Diagnosis

It was stated that delays in diagnosis, particularly in the field of pediatric nephrology within the spectrum of rare diseases, have devastating effects on patients. It was emphasized that, in this non-static group of diseases, every delay in diagnosis leads to a worsening of the clinical condition and causes the process to progress to irreversible stages. It was noted that once this stage is reached, interventions become increasingly difficult to manage, both in terms of patient health and healthcare system capacity.

Health Economics and Cost-Effectiveness Analysis

It was stated that discussions surrounding the use of high-cost medicines should be evaluated together with the total costs arising from disease progression. It was demonstrated that early diagnosis and treatment processes, even when medication costs are included, are far more economical than the costs associated with severe conditions such as end-stage renal failure, including dialysis and transplantation. In this context, it was assessed that a holistic cost analysis offers a more sustainable model for the national economy.

The Scope of Coordination in Service Delivery

It was emphasized that the concept of coordination should be regarded not merely as a medical process, but as a multidimensional structure encompassing referral to treatment following diagnosis, participation in social life, education, and employment processes. It was concluded that coordinating all of these stages through a single center is the key to addressing systemic deficiencies.

The Role of Research and Application Centers and Field Realities

It was stated that although the primary purpose of university-based research and application centers is to generate policy, they have become the focal point for direct patient applications due to the lack of referral mechanisms in the field. It was noted that patients seeking assistance through e-mails or personal applications provides a concrete illustration of the gaps in the system's guidance and support mechanisms.

Referral Mechanisms and a Structural Solution Proposal

The need for a "Coordination Center" structure that would channel patients to the appropriate specialist centers and provide holistic solutions to their problems was clearly defined. It was stated that the operational details and functional model of such a structure should occupy a central place in future strategic planning efforts.

Priority Preferences in Rare Diseases and the Need for Coordination on a Global Scale

Participant

Priority Analysis and Perceptions of Financial Burden

It was stated that the divergence of opinions among participants regarding the options presented in the survey stemmed from the multidimensional nature of the issue. The fact that the financial burden option ranked lower than the other categories was interpreted not as an indication that the issue is considered unimportant, but rather as a reflection of the growing discussion of alternative solution approaches and an unwillingness to accept this factor as a moral or ethical "barrier."

The Relationship Between Awareness and Coordination

It was observed that views initially focused on the "lack of awareness" gradually evolved toward a focus on the "lack of coordination" as a result of the discussions. It was emphasized that the inability to establish coordination among processes lies at the root of many of the structural challenges encountered in diagnosis and treatment pathways.

Data Management and the Follow-up of Complex Diseases

It was stated that, by their very nature, rare diseases are highly complex and that receiving a diagnosis alone is not sufficient for effective process management. The need was expressed for systems capable of bringing together the many variables that emerge throughout the course of a disease, generating high-quality data, ensuring follow-up, and standardizing management processes.

Global Perspective and a Systemic Starting Point

It was recalled that the issue of coordination is not merely a local challenge but is also recognized globally as one of the most fundamental issues in the management of rare diseases. In this context, it was concluded that the current workshop and stakeholder gathering constitute a strategic starting point for the development of a solution model aligned with international standards.

Financial Uncertainty from an Economic Perspective and the Priority of Awareness

Moderator

The Unpredictability of Financial Burden and Its Multidimensional Nature

It was explained why the financial burden option was not identified as the primary challenge despite expertise in the field of economics. It was stated that, without a comprehensive analysis of the multidimensional structure of the rare disease ecosystem and a thorough examination of all relevant variables, it is not possible to accurately estimate the scale of the financial burden. It was assessed that financial projections made before the structural framework is clearly defined may be misleading.

Delayed Diagnosis and Variable Cost Analysis

It was stated that delays in diagnosis and the prolongation of the process have a direct and variable impact on overall costs. It was emphasized that as the number of parameters involved in the process increases, costs remain in a constant state of fluctuation, making it difficult to speak of a fixed financial burden.

The Social Dimension and an Awareness-Oriented Approach

It was stated that addressing the issue solely through numerical data or financial burden would overlook its social dimensions and therefore provide an incomplete projection. In this context, it was concluded that the issue of "lack of awareness" should be regarded as a strategic priority, as it constitutes one of the fundamental building blocks of structural solutions.

Financial Realities and Strategic Coordination Models from the Perspective of Action Plans

***Prof. Dr. Onur Burak DURSUN** / Editor of the Türkiye Rare Diseases Action Plan*

Field Experience and the Priority of Financial Burden

From the perspective of an individual who has served as both coordinator and editor of the Rare Diseases, Autism, and Addiction Action Plans, and who also works as a clinician in the fields of Fragile X Syndrome and autism, it was emphasized that the financial burden constitutes the most critical challenge. It was stated that, particularly when directly observing the demands of patients and their relatives presented to decision-making authorities, financial needs emerge as the most tangible and urgent issue requiring resolution. As demonstrated in the case of SMA, it was noted that financial accessibility lies at the core of processes that have significant societal implications.

Coordination Models in Public Administration and Implementation Challenges

The existing coordination mechanisms in Türkiye were analyzed through three main models:

- Inter-Ministerial Horizontal Model: It was stated that, in models where one ministry is designated as the responsible authority but lacks hierarchical authority over other stakeholder ministries, significant challenges arise in field implementation and coordination with local administrations.
- Single-Institution Model: It was noted that models in which a single institution (generally the Ministry of Health) assumes the central role help accelerate operational processes by clarifying responsibilities; however, they are insufficient on their own to address the multidimensional nature of rare diseases.
- High-Level Centralized Model: It was stated that models coordinated by the former Prime Ministry or, currently, by the Presidency have produced the strongest results. This high-level structure was identified as the most effective approach.

Financial Infrastructure and Stakeholder Participation

It was emphasized that the clarification of budgetary resources is essential for the implementation of a comprehensive action plan. In this regard, it was stated that the active participation of representatives of financial authorities, such as the Ministry of Treasury and Finance, is of strategic importance. It was observed that the strong institutional infrastructure required for such a centralized structure has not yet been fully established.

A Health-Focused Initial Strategy

While it was stated that the ideal solution would be coordination at the level of the Presidency, it was concluded that, given the current circumstances and the fact that approximately 80% of the workload is concentrated within the health sector, establishing a health-focused model at the initial stage would be a critical and priority step in clarifying responsibilities.

In-Depth Analysis of Coordination Models and the Strategic Framework of the Panel

Moderator

The Thematic Focus of the Panel and the Basis for Discussion

It was stated that coordination models would be addressed in full detail during the panel titled Financing and Reimbursement Practices in Rare Diseases at the Rare Lives and Rare Diseases Summit. It was expressed that the structural proposals and modeling options raised by participants would be discussed within the scope of this panel on a more comprehensive and technical basis.

Question Design and Expansion of Scope

It was stated that similar methodological dilemmas were experienced during the preparation phase of the workshop questions, but that the discussion reached the expected level of depth from the very first question. It was emphasized that the rapid expansion of the topic once again confirmed how critical the selected heading is for the rare diseases ecosystem.

Diversity of Perspectives and the Search for Common Ground

It is anticipated that, as the discussion process progresses, perspectives and solution proposals from different areas of expertise will become clearer. Despite the existence of different approaches, it was concluded that the process would contribute to the formation of a sound model by clarifying both common ground and points of divergence.

The content text I prepared in line with the data you provided for your second question is presented below:

"CONDITIONAL REIMBURSEMENT" IS ADOPTED AS THE PRIMARY APPROACH FOR TREATMENTS WITH LIMITED EVIDENCE

A large majority of participants (69%) stated that the most appropriate approach for treatments with limited but promising evidence is the implementation of conditional reimbursement. This high percentage demonstrates that, rather than completely denying access to innovative treatments characterized by uncertainty, stakeholders favor flexible models based on risk-sharing and ongoing data collection processes.

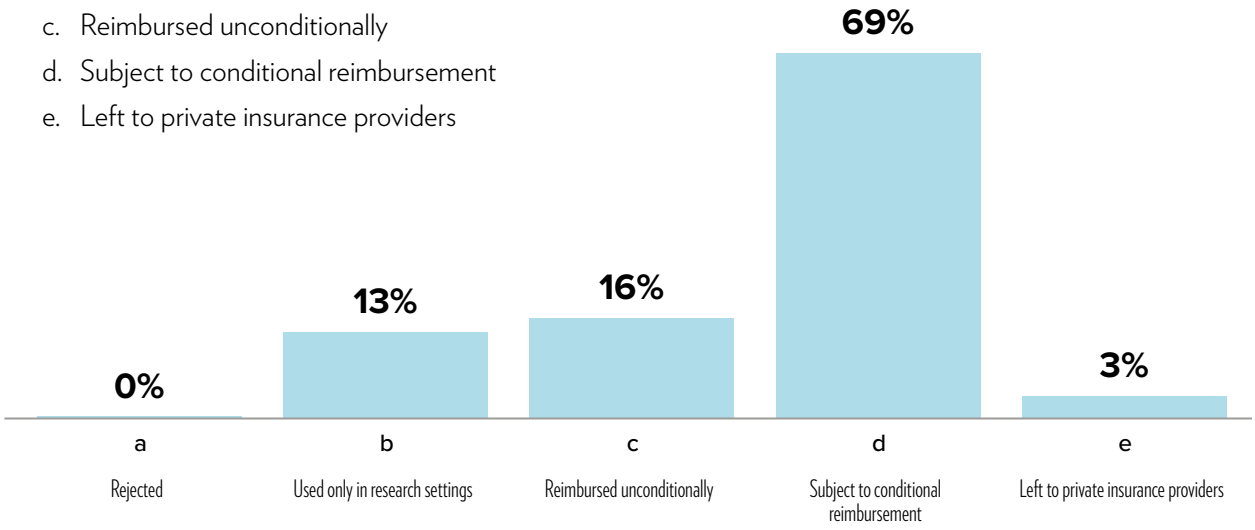
The close distribution between the 13% who argued that such treatments should be limited solely to research settings and the 16% who supported unconditional reimbursement is noteworthy. This distribution indicates the existence of different yet balanced perspectives within the sector regarding the positioning of low-evidence treatments at the intersection of clinical practice and academic research.

The fact that the option of completely rejecting such treatments received 0% support clearly demonstrates that participants reject an entirely restrictive approach toward promising therapies. Meanwhile, the 3% who suggested leaving financial responsibility to private insurance indicates a broad consensus that such critical treatments should remain under the umbrella of public financing.

When the overall distribution is evaluated, it becomes evident that participants tend to balance "evidence" and "access" in relation to innovative treatments through dynamic reimbursement models subject to oversight and evaluation.

2. How should treatments with limited but promising evidence be approached?

a. Rejected
b. Used only in research settings
c. Reimbursed unconditionally
d. Subject to conditional reimbursement
e. Left to private insurance providers



Insufficient Evidence in Rare Diseases and Conditional Reimbursement Models

Moderator

Limited Clinical Evidence and the Management of Uncertainty

It was stated that one of the fundamental challenges in rare diseases is the insufficiency of clinical evidence relating to treatments. Due to the limited size of patient populations, it was noted that clinical studies cannot be conducted on standard scales, making it particularly difficult to predict the long-term effects of drug therapies. It was emphasized that the limited level of evidence, both in terms of health outcomes and cost-effectiveness analyses, creates structural uncertainties for decision-making authorities.

Preferences Regarding Reimbursement Models

According to the survey results, it was stated that the option of "conditional reimbursement" emerged as the most prominent choice by a considerable margin. The fact that the option of "rejection" was not selected by any participant confirmed the solution-oriented approach adopted toward promising treatments. It was assessed that the approach of "leaving the matter to private insurance" received limited support within the framework of seeking an alternative structure capable of assuming the associated risks.

Conditional Reimbursement from a Health Economics Perspective

The conditional reimbursement model was defined as a process of collecting data and generating evidence simultaneously with the use of a medical technology or pharmaceutical product. It was stated that this approach, particularly implemented in the field of oncology and in countries such as Italy where market access agreements are widely used, enables the strengthening of scientific evidence while treatment is being provided. It was further noted that this model offers a rational solution in the field of rare diseases, where uncertainties are particularly high.

Management of Limited Resources and Strategic Planning

It was emphasized that even if success is achieved in diagnosis, screening, and prevention programs, healthcare resources remain limited and therefore must be optimized. It was concluded that the conditional reimbursement system constitutes a strategic tool for the effective management of limited resources and for allocating budgets to treatments whose effectiveness has not yet been fully proven but which demonstrate promising potential.

Conditional Reimbursement and a Data-Driven Approach Through the Lens of the Right to Life in Rare Diseases

Prof. Dr. Tuba EMİNOĞLU */ Division of Pediatric Metabolism, Ankara University / Rare Diseases Research and Application Center*

The Urgency of Access to Life-Saving Treatments

It was emphasized that treatments developed for rare diseases that have reached a certain level of reliability and possess life-saving potential should be made available to patients without delay, particularly when the condition of early diagnosis is met. It was stated that accelerated procedures are already in place for medicines within this scope; however, the success of treatment largely depends on early diagnosis mechanisms.

The Relationship Between Treatment Effectiveness and Early Diagnosis

It was stated that the clinical effectiveness of high-cost medicines decreases significantly in patients who are not diagnosed early. It was noted that this situation poses a risk both to patient health and to the efficient use of public resources, highlighting the importance of implementing treatment access strategies in coordination with diagnostic capacity.

Continuity of Data Collection and Functional Models

Attention was drawn to the difficulties of collecting comprehensive long-term data due to the limited number of patients. It was stated that, in a process where side effects and long-term outcomes become clearer over time, the "conditional reimbursement" model provides a functional solution by enabling a continuous flow of data alongside the delivery of treatment. It was assessed that this model, which has been successfully implemented in many countries, represents the most balanced approach for managing scientific uncertainty.

Ethical Responsibility and Sustainability

It was emphasized that completely excluding promising treatments from reimbursement schemes or restricting them solely to research settings is not an appropriate approach from the perspective of the "right to life." Despite the complexity of financing discussions, it was concluded that there is a necessity to develop models that do not disadvantage patients, protect ethical values, and remain economically sustainable.

Alternative Financing Models and a Dedicated Funding Strategy in Rare Diseases

Participant

Strategy Documents and the Agreed Reimbursement Model

Recalling the previous work carried out during the preparation of the Rare Diseases Strategy Document and Action Plan, it was confirmed that the "conditional reimbursement" model had also been adopted as the primary solution approach within these processes. It was stated that the functionality of this model had been agreed upon through extensive work conducted with academic and bureaucratic stakeholders.

Dedicated Funding and Presidential Coordination

It was emphasized that, beyond the existing options, a "dedicated funding" mechanism represents a strategic alternative for managing the process. In particular, it was stated that the financing of promising treatments should be provided through a specific fund established under the coordination of the Presidency. It was assessed that such a structure would alleviate the bureaucratic and financial burden associated with traditional reimbursement systems.

National Resource Planning and Parliamentary Studies

It was stated that, as a result of the work carried out over the past five years within the strategy commissions of the Grand National Assembly of Türkiye (TBMM), the country possesses sufficient resources that could be allocated to this field. It was further noted that funds specifically designed for rare diseases would directly facilitate access to innovative treatments by removing financial barriers.

Systemic Relief and a Patient-Centered Solution

It is anticipated that the implementation of a dedicated funding model would both protect the general health insurance budget and increase the speed of patient access to treatment. It was concluded that this approach is of critical importance for establishing a sustainable and results-oriented financial architecture within the rare diseases ecosystem.

International Dedicated Funding Practices and Strategic Planning

Moderator

Global Model Reviews and Life-Threatening Disease-Oriented Funding

It was stated that, when different country experiences around the world are examined, "dedicated funding" mechanisms developed for life-threatening diseases can be observed, particularly in countries such as New Zealand and Australia. It was noted that such structures serve as functional models that facilitate financial access for rare and critical disease groups.

Workshop Methodology and the Design of Future Sessions

It was stated that the absence of a dedicated funding option among the survey choices stemmed from a strategic session-planning approach. It was emphasized that this topic is intended to be addressed in greater detail and depth during the later stages of the workshop and was therefore not prioritized within the current voting process.

Support for the Model and Participant Consensus

It was observed that dedicated financing models for life-threatening diseases are supported and that this approach is consistent with the general tendency of the participants. It was concluded that discussions will continue regarding the adaptability of this model to the Turkish rare diseases ecosystem as a strategic solution approach.

BUDGETARY PRESSURE AND LACK OF INSTITUTIONAL OWNERSHIP ARE THE MAIN BARRIERS TO RARE DISEASE POLICIES

A total of 38% of participants identified budgetary pressure as the primary reason why rare diseases do not receive sufficient attention on the policy agenda. This finding indicates that financial constraints within the healthcare economy directly hinder policy development processes in high-cost and specialized fields such as rare diseases.

The 31% of participants who drew attention to the managerial dimension of the issue stated that institutional ownership is weak. These findings suggest that, even where financial resources are available, the absence of a leadership structure capable of assuming responsibility for the issue and ensuring inter-institutional coordination is perceived as a significant barrier to policy development.

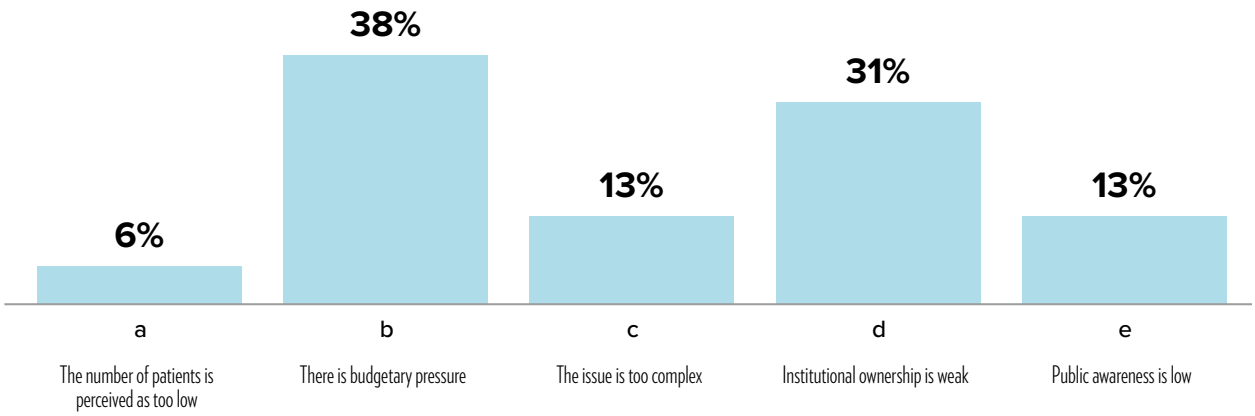
The proportion of participants who cited the complexity of the issue and low levels of public awareness remained equal at 13%. This indicates that both the technical challenges associated with rare diseases and the lack of public support constitute secondary barriers of similar weight for policymakers.

As a noteworthy finding, the option suggesting that the number of patients is perceived as too low accounted for only 6% of responses. This result demonstrates that the rarity of rare diseases or the relatively small patient population is no longer viewed as a sufficient reason for exclusion; rather, the core issue lies in financial and administrative management capacity.

When the overall distribution is evaluated, it becomes clear that the principal reasons why rare diseases fail to secure a stronger place on the policy agenda are not demographic factors, but rather economic concerns and a lack of institutional ownership.

3. Why do rare diseases not receive sufficient attention on the policy agenda?

a. The number of patients is perceived as too low
b. There is budgetary pressure
c. The issue is too complex
d. Institutional ownership is weak
e. Public awareness is low



The Place of Rare Diseases on the Policy Agenda and the Perception of Budgetary Pressure

Moderator

Policy Prioritization and Institutional Ownership

When examining the reasons why rare diseases do not receive sufficient attention on the policy agenda, it was stated that, according to the survey results, the issues of "budgetary pressure" and "weak institutional ownership" emerged as the leading factors. It was emphasized that, despite increasing awareness and significant progress in Türkiye in recent years, the building blocks of the process must be put in place and institutional ownership must be strengthened in order to accelerate progress.

Terminological Clarification: A "High-Priced" Rather Than "Expensive" Approach

From the perspective of health economics, it was stated that describing rare disease treatments as "expensive" is conceptually inaccurate. Instead, it was noted that the term "high-priced" should be preferred. It was further stated that healthcare expenditures should be viewed not as a cost item but as an investment instrument, as empirical evidence demonstrates that every unit of expenditure on programs such as vaccination and screening generates positive returns to society and the economy that exceed tenfold in the long term.

The Source and Focus of Budgetary Pressure

It was observed that discussions regarding budgetary pressure are generally reduced solely to treatment and pharmaceutical costs. However, it was stated that the balancing effect of early diagnosis, screening programs, and preventive approaches on budgets is not discussed sufficiently. It was further noted that approaching the rare diseases ecosystem from a broader perspective, rather than focusing exclusively on treatment, would enable a more accurate interpretation of budgetary pressure.

Stakeholder Representation and Differences in Perspective

It was stated that perceptions of budgetary pressure vary according to stakeholders' positions. It was concluded that the absence of financial authorities (such as the Ministry of Treasury and Finance) from the process may cause budget-focused discussions to remain one-sided, and that all decision-making authorities must come together on common ground in order to establish a comprehensive policy agenda.

Access to Orphan Drugs and Structural Delays in the Diagnostic Journey

Participant

Global Approval Data and the Current Situation in Türkiye

It was stated that, despite the existence of 66 treatment options approved by the European Medicines Agency (EMA) for orphan diseases, the number of accessible medicines in Türkiye remains extremely limited, at only one. It was further noted that, across rare diseases in general, identified treatment options still remain at a low level of approximately 5%, although clinical trial data relating to targeted therapies continue to increase.

Operational Time Losses in Reimbursement Processes

Attention was drawn to observations that, in Türkiye, reimbursement processes can at times take longer than the marketing authorization process itself when evaluating patient access. It was emphasized that this situation results in a significant loss of time in delivering treatments to patients. While conditional reimbursement models theoretically have the potential to facilitate access, it was stated that, in practice, additional criteria introduced within these models can make the process more complex.

The "Critical Threshold" in the Diagnostic Process and the Patient Journey

It was shared that, in Türkiye, the average time required for an individual with a rare disease to receive an accurate diagnosis is seven years and that more than eight different specialist opinions are sought during this process. It was stated that the length and complexity of the patient journey directly and negatively affect treatment outcomes. Attention was drawn to the risk that procedures added to conditional reimbursement mechanisms may further prolong an already lengthy diagnostic process.

The Determining Role of Budgetary Pressure on Access

Reference was made to the correlation between the limited presence of rare diseases on the policy agenda and budgetary pressure. It was concluded that financial constraints and weaknesses in access opportunities are the primary drivers of structural problems throughout the entire continuum from diagnosis to treatment, and that minimizing this pressure is a prerequisite for solution-oriented approaches.

Decision-Making Processes Through the Lens of Budget Management and Public Sector Representation

Moderator

Institutional Participation and the Need for Direct Responses

It was emphasized that the representation of decision-making authorities such as the Social Security Institution (SGK), the General Directorate of General Health Insurance, the Pharmaceuticals Department, and the Ministry of Treasury and Finance is of critical importance in order to facilitate more informed discussions on budget management and financial sustainability within the rare diseases ecosystem. It was stated that the participation of these institutions, which are directly responsible for financing-related matters, would enable more direct and operational responses from a budgetary perspective.

The Current Sector Landscape and Pharmaceutical Policies

It was stated that the current situation regarding pharmaceutical policies and reimbursement lists in Türkiye is a reality well known to all stakeholders. It was concluded that models developed specifically for rare diseases cannot be considered independently of the country's broader pharmaceutical ecosystem and that solution proposals in this field must be designed in coordination with the realities of public finance.

LACK OF INTER-INSTITUTIONAL COORDINATION IS SEEN AS THE PRIMARY POLICY BOTTLENECK IN TÜRKİYE

More than half of the participants (56%) stated that the most fundamental policy challenge in the field of rare diseases in Türkiye is the lack of inter-institutional coordination. This dominant result reveals that the inability of institutions to work in a synchronized manner throughout policy development and implementation processes is regarded as the greatest obstacle within the system and that an urgent governance reform is needed in this area.

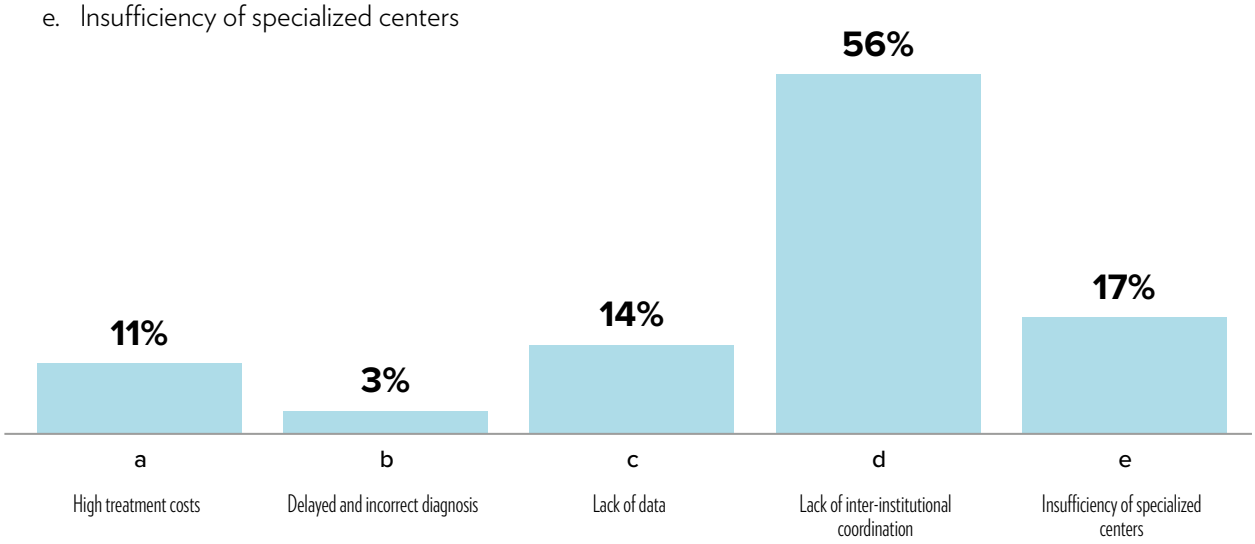
The 17% of participants who identified the insufficiency of specialized centers as the primary challenge, together with the 14% who drew attention to the lack of data, emphasized the need to strengthen the operational infrastructure. This distribution indicates that, following the issue of coordination, both tangible service delivery points (specialized centers) and the information infrastructure (data) required to support decision-making mechanisms are viewed as critical deficiencies.

While 11% of participants identified high treatment costs as the primary challenge, only 3% selected delayed and incorrect diagnosis. This finding demonstrates that participants' focus has shifted away from medical or economic outcomes themselves and toward the structural and administrative processes that generate these outcomes, namely coordination and centralization.

When the overall distribution is evaluated, it becomes evident that, before financial resources or technical diagnostic processes, the establishment of a multi-stakeholder collaboration model and institutional integration is required for the advancement of rare disease policy in Türkiye.

4. In your opinion, what is the most fundamental policy challenge in Türkiye?

a. High treatment costs
b. Delayed and incorrect diagnosis
c. Lack of data
d. Lack of inter-institutional coordination
e. Insufficiency of specialized centers



Policy Deadlocks and the Central Role of Strategic Coordination

Moderator

Multidimensional Problem Analysis and Diversity of Scope

It was determined that the fundamental policy challenges in the field of rare diseases should be evaluated across a broad spectrum, ranging from insufficient data and the lack of specialized centers to inequities in geographical distribution and high treatment costs. It was emphasized that this complex structure must be addressed not only from a medical perspective but also through the design of inclusive policies.

Lack of Inter-Institutional Coordination and the Governance Crisis

It was stated that the strikingly high rate at which the lack of inter-institutional coordination was identified as the primary challenge demonstrates how strongly the main theme of the workshop resonates in practice. In light of the findings obtained, it was acknowledged that rare disease policy is not merely a healthcare issue but also a chronic "governance problem." It was determined that all strategic pathways ultimately point to the need for strong coordination among public institutions and stakeholders.

Structural Deficiencies and the Policy Agenda

It was stated that structural gaps, such as the lack of data and the concentration of specialized centers in certain regions, are both consequences and drivers of inadequate coordination. It was assessed that establishing strategic governance becomes more difficult in a system where information and services are not distributed evenly.

The Governance Gap and the Need for Guidance

It was stated that the current discussions provide strategic guidance on how to address the governance gap within the system. It was concluded that the management of rare diseases should be redesigned through a holistic architecture in which administrative, financial, and technical processes are integrated and operate in coordination with one another.

Coordination-Oriented Policy Design

It was emphasized that future policies should be shaped around a "central coordination structure" capable of addressing communication gaps between institutions and ensuring the efficient use of resources. It was stated that this structural transformation constitutes the cornerstone for resolving all chronic challenges within the rare diseases ecosystem.

THE ESTABLISHMENT OF A NATIONAL FUND IS THE TOP PRIORITY FOR TÜRKİYE

More than half of the participants (56%) stated that the first step to be taken in the field of rare diseases in Türkiye should be the establishment of a national fund. This high preference rate reveals a strong consensus that existing financial models are insufficient to meet the high-cost and sustainable treatment needs associated with rare diseases.

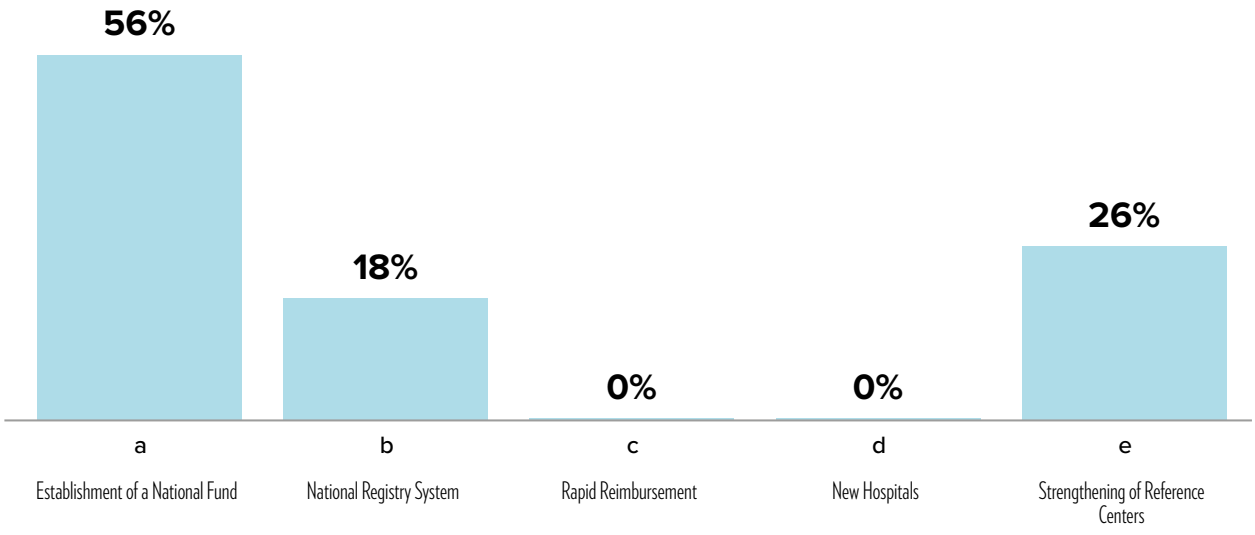
The 26% of participants who prioritized systemic and structural development identified the strengthening of reference centers as the most critical step. Meanwhile, the 18% who regarded a national registry system as the first priority point to a data-driven approach, emphasizing that the effectiveness of other interventions will remain limited without accurate data and patient tracking.

As a noteworthy finding, the options of rapid reimbursement and new hospitals received 0% support and were not selected by any participants. This result demonstrates that, in their search for solutions, participants are focusing not on deficiencies in physical infrastructure (hospital buildings), but rather on the quality of financial mechanisms (a national fund) and existing specialist structures (reference centers). Furthermore, the fact that rapid reimbursement was not selected at all reinforces the perception that the issue is not merely about the speed of processes, but also about the management and sustainability of resources.

When the overall distribution is evaluated, it becomes clear that financial security (a National Fund) and qualified service capacity (Reference Centers) have been identified as the primary strategic priorities for the future of rare disease policy.

5. What should be the first priority in Türkiye?

a. Establishment of a National Fund
b. National Registry System
c. Rapid Reimbursement
d. New Hospitals
e. Strengthening of Reference Centers



The Need for a National Fund and Sustainability in Resource Management

Moderator

Financial Resource Management and Sustainability

It was stated that a strong consensus has emerged among stakeholders regarding the establishment of a national fund. However, it was emphasized that, beyond the creation of a financial pool, the sustainability of such a structure represents the most critical threshold. It was noted that gathering resources at the initial stage is a manageable process, but that it is necessary to clearly define the criteria according to which these resources will be used, as well as where and how they will be allocated.

The Dilemma of Strategic Choice and Prioritization

It was stated that determining whether the proposed fund should prioritize high-cost treatments, early diagnosis and screening programs, or preventive healthcare approaches constitutes a comprehensive area for discussion. It was assessed that the fund should be designed not merely as a payment mechanism, but as a strategic instrument capable of ensuring the holistic management of the system.

The Priority of Legal Definitions and Regulatory Infrastructure

Participant

Legislation as the Cornerstone

It was emphasized that, in order to implement responses such as a "national fund" or "reference centers" to the question, "What should be done first in Türkiye?", priority must be given to the legislative framework that will provide the legal basis for these structures. It was stated that legislation is not merely a regulatory instrument but also a process of "definition" that determines how the entire system will function.

The Need for Definition and Standardization

It was stated that, in order to ensure the continuity of initiatives within the rare diseases ecosystem, clear definitions must first be established at the legislative level. It was noted that technical issues such as the management of the fund, the authorization of reference centers, and data-sharing protocols cannot be sustainable unless they are built upon a solid legal foundation.

The Legal Dimension of Governance

It was assessed that discussions concerning coordination and fund management must be supported by a "legal framework"; otherwise, institutional ownership is likely to remain weak. In this context, it was emphasized that one of the highest-priority components of the Rare Diseases Action Plan should be a comprehensive legislative initiative that clearly defines the roles and responsibilities of all stakeholders.

The Strategic Importance of Legislative Infrastructure and Prevalence Definitions

Moderator

The Structural Impact of the Lack of Definitions

It was stated that the absence of fundamental definitions, regarded as the "A, B, C" stages of legislation within the rare diseases ecosystem, creates a constraining effect on the system as a whole. It was noted that the fact that the widely accepted prevalence threshold of 1 in 2,000 is not formally incorporated into legislation in Türkiye creates uncertainty regarding the legal basis of planned initiatives. It was further stated that, unless this fundamental issue is resolved, it becomes difficult for the administrative and financial mechanisms built upon it to operate at full capacity.

The Relationship Between Fund Size and Disease Definitions

It was emphasized that clarifying the prevalence threshold (such as 1 in 2,000 or 1 in 10,000) is not merely a statistical matter, but a financial parameter that directly determines the size and scope of a national fund. It was stated that, once the definition is clarified, it will become possible to determine which diseases will be included within the funding pool, thereby forming the basis of budget planning. It was assessed that establishing a sustainable financial management model becomes more difficult in a system where such boundaries have not been clearly defined.

Reference Centers and Geographic Planning Criteria

Attention was also drawn to the determining role of legislation in the establishment of reference centers. It was stated that the criteria according to which centers will be authorized and the manner in which their geographic distribution will be standardized are directly linked to the scope of the rare disease definition. In this regard, it was noted that legislation should be viewed not only as a set of rules but also as an operational roadmap.

An Implementation-Oriented Approach and the Role of Public Authorities

Although the workshop primarily focused on implementation-oriented topics, it was acknowledged that the underlying issue of "definition and scope" constitutes an undeniable priority. It was recalled that the decision-making public authority (the Ministry) plays the determining role in clarifying which diseases fall within this strategic scope and where the legal boundaries should be drawn.

National Registry Systems and Structural Barriers to Data Entry

Specialist Dr. Murat GÜLŞEN */ Head of the Department of Autism, Intellectual Disabilities, and Rare Diseases*

Specific Data Generation and the Limitations of ICD Coding

It was stated that one of the fundamental challenges within the rare diseases ecosystem is the inability to establish specific diagnostic groups due to the limitations of ICD coding. To address this deficiency, it was recalled that the "Rare Diseases Data System" was established in 2022. However, it was noted that data entry has not reached the desired level within the current data infrastructure due to physicians' workload and time constraints.

Mandatory Registration Mechanisms and System Integration

It was stated that one of the main shortcomings of existing data systems stems from the fact that rare disease registrations are not currently included within a mandatory entry framework integrated into reimbursement

systems. While records linked to reimbursement processes are maintained regularly, it was noted that the absence of an incentivizing or mandatory mechanism for data entry specific to rare diseases contributes to the persistence of data deficiencies. In this regard, it was stated that efforts to appoint "data officers" and further develop registry systems continue as a strategic priority.

The Definition Problem in Legislation and the Impact of Prevalence Thresholds

It was emphasized that rare diseases do not yet have a clearly defined legal definition within the legislative framework and that the prevalence threshold of 1 in 2,000, which is widely accepted under European standards, has been adopted within the scope of the Action Plan. Although the Action Plan constitutes the first official document in this field, it was stated that uncertainties remain regarding how this threshold will be reflected in regulations and binding legislation.

Debates on Inclusiveness and Ethical Boundaries

It was cautioned that determining a prevalence threshold (for example, 1 in 2,000 or 1 in 50,000) should not be used solely as a tool for reducing costs. It was stated that there is a significant risk that patient groups falling outside any rigidly defined threshold may encounter barriers to accessing services. Therefore, it was emphasized that the question of where legal boundaries should be drawn must be evaluated carefully from both the perspectives of financial sustainability and patient rights.

The Methodological Necessity of National Prevalence Studies

It was stated that data collected only from selected pilot centers or a limited number of provinces may be insufficient to represent Türkiye as a whole. It was concluded that, in order to avoid creating inaccurate projections during the transition to a legislative framework, comprehensive prevalence studies covering the entire country and taking regional differences into account are required.

Centers of Excellence and a Tiered Management Strategy

Prof. Dr. Fatih ÖZALTIN / *Director, Hacettepe University Center for Genome Sciences and Rare Diseases Research and Application – Faculty Member, Division of Pediatric Nephrology, Hacettepe University*

Prioritizing Reference Centers

It was stated that, given the specific and complex nature of rare diseases, the most strategic answer to the question, "What should be the first priority in Türkiye?" is the strengthening of reference centers. It was emphasized that, even if a national fund is established, the effective use of resources will remain uncertain unless a specialized "center of excellence" structure is created. In this regard, it was stated that patient-oriented, authorized centers specializing in specific disease groups should be defined.

Reducing Diagnostic Delays and Accreditation

It was stated that the diagnostic journey, which currently takes an average of 6–7 years in Türkiye, can only be shortened through specialized centers. It was proposed that these structures should be accredited by a high-level institution such as TÜSEB and that coordination among centers should be ensured through a central coordination unit. It was assessed that this model would also serve as the primary source of high-quality data feeding the national registry system.

Financing Approaches and the Time Cost of Reimbursement Processes

It was stated that the lack of data encountered in rare diseases should not be used as a justification for delaying reimbursement processes. Referring to examples from Europe (Italy, France, and the United Kingdom), it was

recalled that the discussion is focused not on the question of "Should we pay?" but rather on "How should we pay?" It was emphasized that Türkiye should also move toward a solution-oriented reimbursement vision capable of managing uncertainty.

Multi-Layered Processes and the Loss of Treatment Effectiveness

It was stated that the current reimbursement system consists of a lengthy and highly layered structure. It was noted that, due to delays in licensing and reimbursement stages, diseases continue to progress and that, in some cases, patients may have passed the stage at which treatment can provide meaningful clinical benefit even when financing is eventually secured. It was stated that this situation represents both a human and economic loss, underscoring the critical importance of accelerating processes.

Risk Management: Prevention at the Source and Screening Programs

It was stated that the most fundamental and effective policy is to prevent disease before it develops, while it is still at the risk stage. It was further noted that expanding screening programs and strengthening early diagnosis mechanisms represent the most rational strategy for reducing the burden of disease on the healthcare system. It was concluded that preventing risk at its source is indispensable for a sustainable healthcare policy.

The Classification of Rare Diseases in Light of Epidemiological Data

Prof. Dr. Uğur ÖZBEK */ Principal Investigator, İzmir Biomedicine and Genome Center*

Prevalence Figures and an Inclusive Strategy

It was emphasized that threshold values used in the definition of rare diseases, such as 1 in 2,000 or 1 in 10,000, while scientifically valid parameters, should not become restrictive factors in policy development processes. Referring to Orphanet data, it was stated that only 10–15% of the approximately 6,500 identified diseases fall within this prevalence range, whereas the remaining 85% consist of much rarer conditions occurring at frequencies of "one in one hundred thousand" or "one in one million."

Ultra-Rare Diseases and a Holistic Approach

It was cautioned that narrowly defined thresholds may result in ultra-rare disease groups, which constitute a significant portion of the affected population, being excluded from the system. It was stated that, while a technical distinction between rare and ultra-rare diseases may be made, such a distinction should not create barriers to access. It was further noted that including ultra-rare cases within the scope of the system from the outset would not create an unmanageable burden and is, in fact, necessary for the establishment of an equitable model.

Needs-Based Rather Than Disease-Based Policy

It was stated that the primary issue is not the prevalence of individual diseases, but rather the accurate identification of the common needs shared across these disease groups, including diagnostic infrastructure, access to specialist centers, and sustainable financing. It was assessed that solution models should be designed through a holistic architecture encompassing all rare and ultra-rare disease groups, rather than through fragmented structures.

Systemic Integration and Resource Efficiency

Support was expressed for the model centered on centers of excellence and coordination. It was emphasized that a solution can only be achieved through an inclusive strategy that takes into account the needs of all stakeholders and all disease groups.

Functional Classification and the Need for a Türkiye-Specific Coding System

Participant

Categorical Approaches in Rare Diseases

It was stated that, rather than evaluating all cases on the same scale, a functional classification model should be adopted for the definition and monitoring of rare diseases. In light of the limitations of existing ICD codes, it was proposed that a Türkiye-specific "Rare Diseases ICD System" should be established. Within this framework, it was suggested that diseases be categorized as preventable, treatable, preventable-treatable, and undiagnosed patient groups, thereby enabling more efficient management processes.

Prioritization and Resource Management

It was stated that attempting to manage a disease with a prevalence of 1 in 2,000 and a condition occurring once in a million under the same operational framework places a significant burden on the system. While all groups share common social and economic needs, it was emphasized that prioritizing treatable conditions is a strategic necessity for policymakers and regulators. It was further stated that limited public resources should be allocated rationally to areas where the greatest clinical benefit can be achieved.

Preventive Healthcare Services and Early Diagnosis Strategies

It was emphasized that expanding screening programs, particularly for diseases within the "preventable" category, is critically important in reducing the disease burden at its source. It was assessed that identifying diseases that can be prevented through early intervention would minimize the burden on both individual health outcomes and public finances.

Public Awareness and the Reality of Consanguineous Marriage

Taking into account Türkiye's demographic realities and the prevalence of consanguineous marriages, it was stated that specific screening and educational programs addressing this issue should be expanded. It was concluded that increasing public awareness represents the most effective long-term strategy for the prevention of rare diseases.

Registry Systems and Holistic Management

It was stated that registry systems should be developed in a manner that encompasses not only known diseases but also the "undiagnosed patient group." Based on the recognition that each disease group has distinct clinical and economic needs, it was emphasized that registry systems, early diagnosis, and treatment processes should be coordinated as an integrated whole, aligned with Türkiye's budgetary capacity and strategic objectives.

Resource Constraints and Ethical Dilemmas from a Health Economics Perspective

Moderator

Resource Constraints and the Risk of Neglect

It was stated that, due to the limited nature of healthcare resources, certain patient groups may face the risk of

being excluded from services or neglected. It was emphasized that this situation raises profound ethical and moral debates and that such risks must be managed carefully when designing financing models.

Alternative Financing Models and Equity Oversight

It was stated that some financing models implemented globally are ethically questionable. In particular, approaches that seek to place a greater share of financial responsibility on specific risk groups or impose additional obligations on individuals based on demographic realities such as consanguineous marriage were noted to be inconsistent with the principles of social justice.

The Principle of Equality and Citizenship Rights

It was stated that, within a system in which individuals fulfill their tax obligations, restricting access to healthcare services on the basis of personal choices or risk factors is contrary to the principle of "equality." It was assessed that such approaches do not rest on a fair or ethical foundation and are not considered applicable within Türkiye's current resource structure and societal expectations.

The Strategic Balance Between Equity and Efficiency

It was stated that health authorities must confront the reality of limited resources while simultaneously safeguarding the principles of equality and justice. It was further noted that ensuring financial sustainability without compromising human and ethical values represents one of the most sensitive aspects of rare disease policy.

Future Perspectives and Financial Architecture

It was concluded that the financing models to be discussed in greater detail during the relevant sessions of the Summit should encompass not only mathematical budget management but also a societal "equity oversight" dimension. It was emphasized that maintaining this delicate balance constitutes a cornerstone of the sustainability of the Rare Diseases Action Plan.

A Holistic Approach: Transitioning from a Medical Focus to Social and Strategic Governance

Deniz ATAKAY */ President of the Rare Diseases Federation*

System Sustainability and Resource Planning

It was stated that focusing solely on existing patients is not sufficient for the management of rare diseases, as the process requires systemic continuity. Emphasizing the need to reduce the number of affected births, it was noted that, if such births continue, the continuous generation of resources for areas such as the employment of healthcare professionals, the establishment of specialized centers, and ongoing training programs becomes a necessity. Under such circumstances, it was stated that the system must evolve beyond merely providing treatment and develop into a structure that also builds capacity.

Multi-Stakeholder Governance and Inter-Ministerial Coordination

It was stated that efforts in the field of rare diseases should not remain solely the responsibility of the Ministry of Health. It was proposed that the process should be expanded to encompass all stages of an individual's life through the active involvement of institutions such as the Ministry of Family and Social Services and the Ministry

of National Education. The importance of managing social services and special education needs, which arise after the stages of diagnosis and treatment, through a holistic strategy was emphasized.

Managing Risk at Its Source and Raising Awareness

It was stated that structural challenges cannot be resolved without increasing public awareness of rare diseases and establishing appropriate educational models. It was cautioned that, if preventive measures (such as screening programs and genetic counseling) remain insufficient, both the number of individuals requiring long-term care and the rate of births affected by rare diseases will continue to rise. It was assessed that such an increase would create an unsustainable burden on both the social fabric and the public economy.

Expanding the National Strategic Action Plan

It was stated that the healthcare system has achieved an important milestone by preparing its current strategic plans, but that these plans must be integrated with the operational processes of other ministries. It was concluded that, rather than adopting an exclusively treatment-oriented approach, there is a need for a national coordination structure that is preventive, inclusive, and encompasses the entire life cycle of the individual.

Future Outlook: Socio-Economic Balance

It was stated that success in the management of rare diseases depends on synchronizing medical interventions with social support mechanisms. It was emphasized that the key factor in preventing the growth of the social and economic burden is the prevention of risk at its source and the development of education and care models that enable individuals to participate fully in society.

A "HOLISTIC ASSESSMENT" APPROACH DOMINATES REIMBURSEMENT DECISION-MAKING

A large majority of participants (69%) argued that reimbursement decisions in rare diseases should be based on the assessment of all relevant parameters rather than focusing on a single criterion. This high percentage clearly demonstrates the need for multi-criteria decision analysis (MCDA) models that incorporate clinical, economic, and ethical considerations.

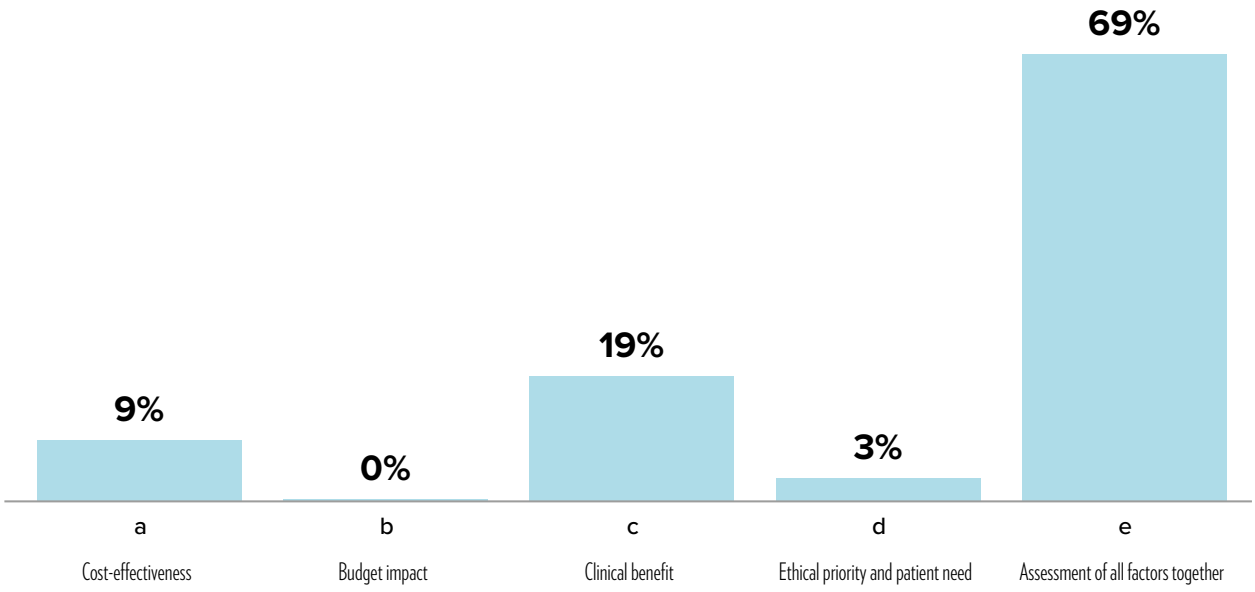
The 19% of participants who identified clinical benefit as the most important criterion emphasized that treatment effectiveness should take precedence over all other considerations. While 9% prioritized cost-effectiveness analyses, only 3% identified ethical priority and patient need as the primary criterion.

Notably, the criterion of budget impact received 0% support. This finding demonstrates that participants oppose reimbursement decisions being driven solely by short-term financial obligations or budgetary constraints. It also indicates a sector-wide consensus that the overall value of treatment—including the balance between benefit, cost, and patient need—should be placed at the center of decision-making rather than budgetary limitations alone.

When the overall distribution is evaluated, it becomes evident that, in the formulation of reimbursement policies, stakeholders expect a transparent and holistic assessment framework that integrates clinical evidence, economic sustainability, and patient needs, rather than relying on narrow or single-dimensional approaches.

6. What should be the most important criterion in reimbursement decisions?

a. Cost-effectiveness
b. Budget impact
c. Clinical benefit
d. Ethical priority and patient need
e. Assessment of all factors together



A Multidimensional Assessment Approach in Reimbursement Decision-Making

Moderator

Pharmacoeconomic Parameters and Decision-Making Mechanisms

It was stated that factors such as cost-effectiveness, clinical outcomes, and alternative treatment options constitute the foundation of decisions regarding the inclusion of rare disease treatments within reimbursement schemes. It was noted that the prominence of the option "assessment of all factors together" in the survey results stems from the complementary nature of these elements. It was emphasized that, in order to ensure that decision-making processes rest on a rational foundation, it is essential to analyze a comprehensive set of data rather than relying on a single variable.

Traditional Assessment Criteria: Clinical and Financial Dimensions

Drawing on experience from commissions operating under the General Health Insurance (GSS) system, it was stated that the clinical level of evidence, available alternatives, and the direct impact on the public budget are the primary factors examined when a medicine is brought forward for evaluation. However, it was also noted that this technical assessment process is not always sufficient to address the specific challenges inherent to rare diseases.

The Missing Link in Decision-Making: Social and Societal Impact

It was emphasized that existing legislation and implementation practices focus primarily on the medical and financial dimensions of treatments, while remaining insufficient in assessing their social and societal impacts. It was stated that analyses of societal burden, which have been successfully implemented in other countries, should also be integrated into decision-making mechanisms in Türkiye.

Prioritizing Patient Need and Societal Burden

It was stated that reimbursement decisions should not be determined solely by clinical success or budgetary discipline. It was assessed that, for a fair and equitable model, factors such as the depth of patient need, the caregiving burden placed on families, and the indirect socio-economic costs generated by the disease must also be incorporated into the decision-making process.

A Vision for Holistic Assessment

It was concluded that the reimbursement system should evolve into a flexible structure capable of bringing together scientific evidence, financial realities, and societal conscience within a single framework. It was emphasized that, in the context of rare diseases, the most rational roadmap would be the development of a "Value-Based Reimbursement" approach designed within an expanded framework that also incorporates social benefit.

AN INTEGRATED AND MULTIFUNCTIONAL ROLE IS EXPECTED FROM PRIMARY CARE

An overwhelming majority of participants (75%) stated that primary healthcare services should assume responsibility for early diagnosis, referral, and coordination as an integrated whole in the management of rare diseases. This finding demonstrates that primary care is viewed not merely as a "point of entry" but as a strategic component that should take active responsibility at every stage of the patient journey, from diagnosis through ongoing care.

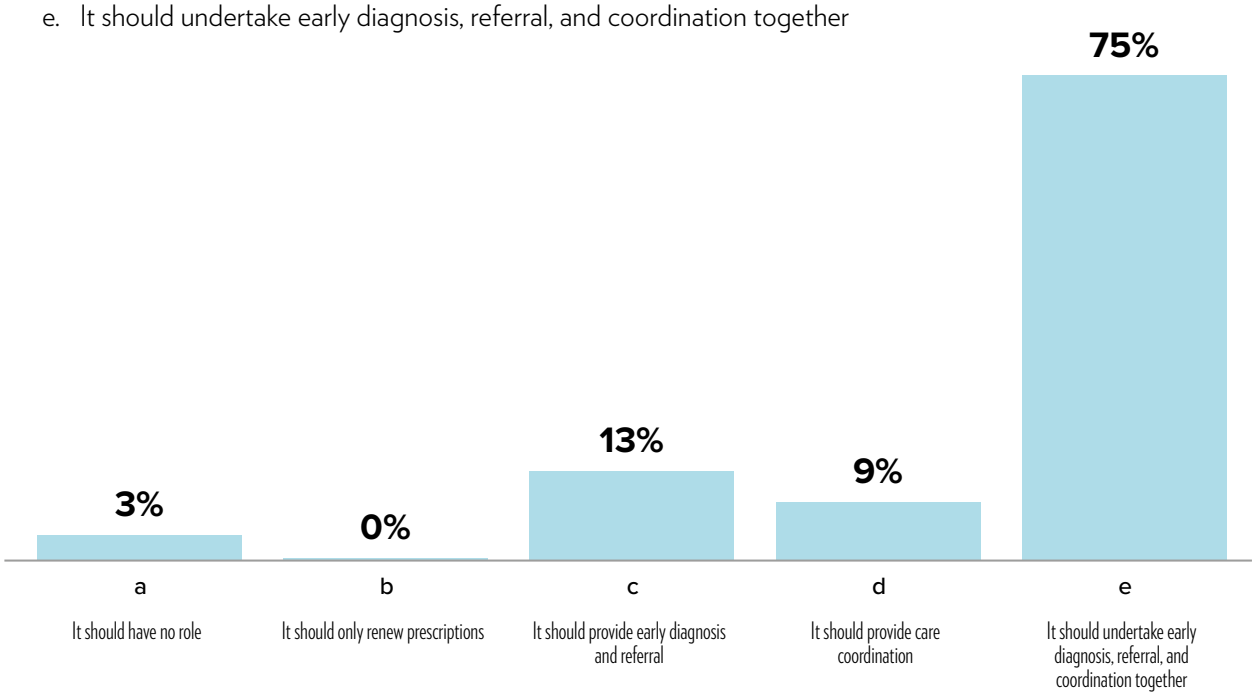
Among participants who favored a more limited and specific role for primary care, 13% stated that the focus should be on early diagnosis and referral, while 9% argued that care coordination should be its primary function. This distribution reinforces the need for a holistic approach that combines the clinical responsibilities of primary care with its operational management functions.

The fact that the option of prescription renewal alone received 0% support reflects a clear opposition to positioning primary care as a passive administrative unit. Furthermore, the fact that only 3% of participants believed primary care should have no role in this process demonstrates that family medicine is regarded as an indispensable stakeholder within the rare diseases ecosystem.

When the overall distribution is evaluated, there is a strong expectation that primary care should be strengthened in rare disease management from both a clinical perspective (diagnosis and referral) and a managerial perspective (coordination), and that it should be integrated into the center of the system.

7. What should be the role of primary care?

a. It should have no role
b. It should only renew prescriptions
c. It should provide early diagnosis and referral
d. It should provide care coordination
e. It should undertake early diagnosis, referral, and coordination together



The Functional Role of Primary Healthcare Services and Diagnostic Complexity

Moderator

Systematic Coding and the Early Suspicion Stage

It was stated that the lack of an established systematic coding infrastructure for the identification of rare diseases within primary healthcare services constitutes a fundamental structural challenge. It was noted that, in family medicine practice, patients generally present with non-specific symptoms such as "fatigue," "tiredness," or "dizziness," which often leads to the possibility of a rare disease being overlooked at the initial stage. It was further stated that the pathway from symptom to diagnosis does not follow a linear trajectory within the current system, creating significant discontinuities in the diagnostic process.

The Priority of Coordination and Referral Mechanisms

It was emphasized that primary care is not expected to provide treatment for rare diseases; however, it should play a central role in early suspicion, appropriate guidance, and effective referral mechanisms. Given that family medicine serves as the first point of contact in the patient's diagnostic journey, it was stated that its strategic importance lies in functioning as a "coordination center" for directing complex cases to specialized units.

The Need for a Digital Ecosystem and Inter-System Integration

It was stated that Türkiye possesses a highly developed and widespread digital health infrastructure, including systems such as e-Nabız, Medula, and various patient follow-up platforms. However, it was noted that these platforms currently operate as separate and disconnected modules, preventing data from being processed through a holistic approach. It was assessed that the fact that each system serves only its own limited purpose results in information loss, particularly in areas such as rare diseases that require multidisciplinary follow-up.

Holistic Data Management and Inter-Institutional Coordination

It was stated that the integration of digital systems is not merely a technical requirement but also a means of establishing an efficient framework for collaboration among stakeholders such as the Ministry of Health, the Ministry of National Education, and the Ministry of Family and Social Services. It was concluded that, through such integration, all aspects of a patient's journey—from education and social support to diagnosis and treatment—should be brought together within a single, holistic digital monitoring mechanism.

Digital Transformation in Governance

It was emphasized that strengthening primary care depends on providing physicians with decision-support systems capable of raising suspicion of rare diseases and enabling digital systems to "communicate" with one another. It was stated that robust digital integration represents the most fundamental tool for addressing the lack of coordination in rare disease management.

Clinical Awareness and the Paradigm of Diagnostic Suspicion in Primary Care

Prof. Dr. Fatih ÖZALTIN / *Director, Hacettepe University Center for Genome Sciences and Rare Diseases Research and Application – Faculty Member, Division of Pediatric Nephrology, Hacettepe University*

Analysis of Early Signs and the Management of Diagnostic Suspicion

It was stated that primary healthcare services serve as a critical "filtering mechanism" for the early detection of rare diseases. The ability to recognize complex conditions underlying common symptoms such as dizziness, fatigue, or developmental delay in children was described as the starting point of the system. Although family physicians are not expected to establish a specific specialist diagnosis, identifying that "something is not progressing as it should" and directing the patient to the appropriate center was considered the most critical threshold in the process.

Medical Education and Academic Awareness Strategies

It was emphasized that medical curricula should be enriched with a rare diseases perspective in order to train future family physicians. In this regard, it was stated that elective courses introduced within Hacettepe University (HÜGEM) and awareness activities encompassing students across all stages of medical education constitute the cornerstones of a long-term solution. The objective is to equip future physicians with the capacity to consider the possibility of rare diseases as a clinical option throughout their professional careers.

Decision-Support Systems and Technological Integration

It was stated that the use of artificial intelligence-based technological systems is essential to enable primary care physicians to manage both their workload and the complexity of symptoms. It was noted that a "smart algorithm"

integrated into Family Health Centers by the Ministry of Health could identify and suggest the possibility of rare diseases based on entered symptoms. This type of digital support was described as one of the most effective tools for minimizing delays in diagnosis (the diagnostic odyssey).

Proposed Structural Model and Coordination Architecture

With regard to Centers of Excellence and Central Governance, a three-tier structural model was proposed for the successful management of rare diseases:

1. **Specialized Centers of Excellence:** Establishment of structures with advanced expertise in specific disease groups.
2. **Strategic Coordination Board:** Creation of a central body responsible for coordinating all centers and managing operational workflows.
3. **Ministerial Oversight and Accreditation:** Operation of this board under the direct authority of the Ministry of Health, with responsibility for overseeing the entire patient journey from end to end.

Transition from Coding Systems to a Data-Oriented Registry System

It was stated that existing ICD codes are inadequate for rare diseases and are generally used by physicians only for mandatory diagnostic test entries. Therefore, it was emphasized that, rather than being constrained by rigid coding systems, a more flexible and comprehensive "Rare Diseases Registry System" should be established. It was concluded that original registry platforms based on real-world data and designed to facilitate patient follow-up—such as the model developed at Hacettepe University—should be expanded to the national level.

International Primary Care Models and Symptom-Based Referral

Moderator

Global Best Practices and Classification Systems

It was emphasized that internationally recognized classification systems exist to support the early identification of rare diseases within primary healthcare services. It was stated that these systems, which include approximately 1,300 specialized diagnostic and symptom codes, have been successfully implemented in countries such as Ireland. These frameworks were noted to enable family physicians to identify complex cases beyond the scope of standard clinical protocols.

Symptom-Based Flagging and Integration with Reference Centers

It was stated that, beyond traditional diagnostic processes, the method of "symptom-based flagging" plays a key role in directing patients to reference centers. It was noted that, without requiring physicians to establish a definitive diagnosis, patients can be referred directly to specialized centers when atypical clinical findings are flagged within the system. This model was assessed as one of the most effective approaches for shortening diagnostic timelines and improving patient safety.

The Position of Primary Care Within the Early Diagnosis Ecosystem

It was stated that such flexible and inclusive coding systems implemented at the primary care level facilitate the identification and inclusion of "missed cases" within the rare diseases ecosystem. It was concluded that systematically

recording findings beyond well-known symptoms not only improves the quality of epidemiological data but also helps maintain the workload of reference centers at a rational level.

System Integration and Adaptation

It was emphasized that the success of international models is rooted in the fact that primary care services are supported by technological infrastructure and operate within a framework of "real-time" data sharing with higher levels of care. It was stated that optimizing Türkiye's existing digital capacity to incorporate such symptom-based referral models constitutes a strategic necessity.

Time Management in the Diagnostic Journey and Cost Optimization

Prof. Dr. Fatih ÖZALTIN / *Director, Hacettepe University Center for Genome Sciences and Rare Diseases Research and Application – Faculty Member, Division of Pediatric Nephrology, Hacettepe University*

Reducing Diagnostic Delays and Enabling Early Intervention

It was stated that designing primary healthcare services as an effective filtering and referral mechanism could significantly reduce the period of "undiagnosed disease" (diagnostic odyssey), which may range from 5 to 10 years in rare diseases. It was noted that involving family physicians in the process would prevent patients from losing time within the system without clear direction and would facilitate access to specialized units at the earliest stage, when the opportunity for effective clinical intervention is greatest.

Preventing Unnecessary Healthcare Expenditures and Diagnostic Burden

It was emphasized that, due to the absence of effective referral mechanisms, even relatively manageable clinical conditions such as anemia can result in repetitive and unnecessary diagnostic investigations across multiple specialties. It was stated that this situation imposes a substantial economic burden on public finances while also reducing the efficiency of the healthcare workforce. It was assessed that managing suspicion of rare diseases through a rational algorithm would help eliminate such unnecessary expenditures within the healthcare system.

Reducing Systemic Burden and Improving Resource Efficiency

It was stated that accurate symptom-based assessment of patients can lead to lower-cost, outcome-oriented solutions. It was concluded that the unnecessary burden placed on the healthcare system can only be reduced through a structured discipline of "screening and referral" beginning at the primary care level.

Clinical Experience and Practical Applications

Based on field experience, it was stated that directing complex cases to inappropriate specialties at the initial stage both delays the diagnostic process and increases outpatient workload in hospitals. Therefore, it was emphasized that the strategic priority in rare disease management should be "coordination at the point of entry," which enables the most efficient use of available resources.

THE DEMAND FOR "INSTITUTIONAL AND CONTINUOUS REPRESENTATION" OF PATIENT ORGANIZATIONS IS COMING TO THE FORE

A total of 67% of participants stated that patient organizations should be represented in a formal and continuous manner within decision-making processes related to rare diseases. This strong majority reflects a clear sector-wide expectation that patient associations should move beyond being organizations whose views are sought externally and instead become a direct and permanent component of decision-making mechanisms.

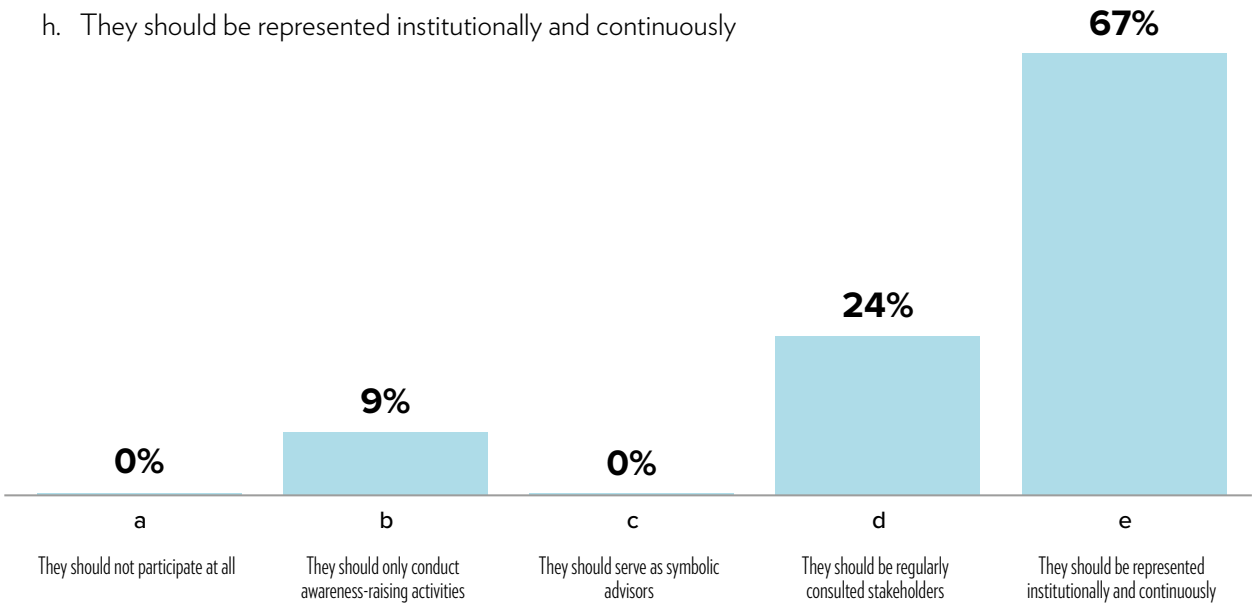
The proportion of participants who believe that patient organizations should serve as regularly consulted stakeholders stands at 24%. This finding demonstrates that a combined 91% of participants agree that patient representation should be established on a systematic and periodic basis.

While 9% of participants viewed the role of these organizations as being limited to awareness-raising activities, the options of symbolic advisory status or complete exclusion from decision-making processes received 0% support. These results demonstrate that patient organizations are not perceived as merely symbolic or "showcase" entities; rather, they are regarded as functional partners that add value to processes and contribute to solutions.

When the overall distribution is evaluated, it becomes clear that, within the rare diseases ecosystem, the "patient-centered" approach is expected to evolve into a "patient representation-based governance" model, with such representation being granted a formal institutional identity.

8. How should patient organizations participate in decision-making processes?

d. They should not participate at all
e. They should only conduct awareness-raising activities
f. They should serve as symbolic advisors
g. They should be regularly consulted stakeholders
h. They should be represented institutionally and continuously



The Strategic Role of Patient Associations in Decision-Making Processes

Moderator

The Level of Participation and Institutional Representation

It was stated that the discussion in rare disease management has moved beyond whether patient associations should participate and has instead focused on the level and manner of their participation. In line with stakeholder preferences, it was emphasized that patient associations should be represented within processes on an institutional and continuous basis. It was noted that this approach supports the objective of transforming associations from organizations limited to awareness-raising activities into active and professional components of the system.

The contributions of patient associations to the system were defined under three main headings:

- **Provision of Real-World Data:** Delivering biopsychosocial data from patients' daily lives to decision-makers, beyond the scope of clinical studies.
- **Field Intelligence:** Communicating implementation challenges and patient needs directly and rapidly to the relevant authorities.
- **Coordination Support:** Strengthening the bridge between patients and the healthcare system throughout the diagnostic and treatment journey.

International Models and Reimbursement Committees

It was observed that patient associations around the world have evolved from the role of "consulted stakeholders" to that of "active participants." It was recalled that, particularly in countries such as the United Kingdom (for example, within NICE-related processes), patient representatives may participate at certain levels in reimbursement committees dealing with ultra-rare diseases. It was stated that such models constitute an important reference point for the coordination structures planned in Türkiye.

A Proposal for Coordination and Integration in Türkiye

It was emphasized that the representation of patient associations within the proposed national coordination structure in Türkiye should be granted formal legal and institutional status. It was stated that such integration would enhance both the transparency of decision-making and the patient-centered nature of the system.

The Structural Evolution of Patient Associations

It was noted that patient associations are increasingly evolving into more professional organizations with greater technical capacity. It was concluded that this transformation has moved associations beyond their traditional role as advocacy groups, positioning them instead as strategic partners involved in the development of healthcare policies. It was stated that, for the sustainability of the system, incorporating this professional expertise into decision-making mechanisms is not merely an option but a necessity.

The Transition from Volunteerism to Institutionalization in Patient Advocacy

Patient Association Representative

The Strategic Value of Experiential Knowledge

It was stated that patient associations have evolved into their current structures through a process that begins at the moment of confronting a disease and is sustained through significant personal dedication and sacrifice. While scientific knowledge is provided by academic and medical authorities, it was emphasized that the realities derived from "lived experience" constitute a complementary source of insight for the system. It was noted that the integration of these two sources of knowledge—scientific and experiential—is the key to developing realistic solutions in rare disease policy.

The Limitations of Awareness Activities and the Need to Expand Scope

It was stated that an approach focused solely on awareness-raising activities is insufficient to address the multidimensional and structural challenges associated with rare diseases. It was noted that associations operating on a voluntary basis cannot realistically shoulder the entire burden of prevention, awareness-building, and public education at the national level. Therefore, it was emphasized that the role of patient associations should extend beyond that of a mere "awareness group" and evolve into that of a professional stakeholder within decision-making mechanisms.

The Need for Inter-Ministerial Integration

It was recalled that rare diseases encompass not only a medical dimension but also educational and social service dimensions. Accordingly, it was proposed that a coordinated working model be established among the Ministry of National Education, the Ministry of Family and Social Services, and the Ministry of Health. It was stated that patient associations should occupy a strategic position within this multidimensional structure by providing input, sharing experience, and actively participating in the process.

A Systematic and Professional Participation Model

It was stated that the inclusion of patient associations in the system should not take the form of transferring the entire burden onto them. Rather, it should be built upon a professional framework of cooperation in which the data and insights brought from the field by associations are heard, understood, and incorporated into decision-making. It was concluded that a more systematic and nationally effective model of awareness and governance can only be achieved when associations become an integral part of an institutional structure.

The Necessity of Institutional Representation

It was stated that positioning patient associations not merely as consulted entities but as genuine solution partners would directly influence the success of the Rare Diseases Action Plan in practice. It was emphasized that being part of a strong, institutional structure with national-level representative capacity is of vital importance in enabling associations to accurately convey field realities to the system.

Economic Sustainability and the Financing Dilemma

***Assoc. Prof. Dr. Mehmet GÜNDÜZ** / Secretary General, Pediatric Nutrition and Metabolism Society*

Recurring Challenges and Approaches to Solutions

It was stated that, despite many years of work in the field of rare diseases, the fundamental challenges and proposed solutions continue to revolve around similar themes. It was noted that discussions with public institutions reveal a comparable picture and that solution-oriented approaches do not always achieve the expected impact due to operational burdens. It was emphasized that every new initiative developed to address existing challenges brings additional financial and administrative demands to the system.

Financial Sustainability and Analysis of the Pharmaceutical Budget

It was stated that the treatment costs associated with rare diseases have reached critical levels in relation to the national healthcare budget and imported pharmaceutical expenditures. It was emphasized that approximately 90% of Türkiye's imported pharmaceutical budget is allocated to this area and that, when compared with concrete economic indicators, the sustainability of this situation remains open to debate. By way of illustration, it was noted that export revenues generated through the efforts of millions of people (using hazelnut exports as an example) are being spent on high-cost genetic therapies for a very limited number of patients, raising questions regarding the alignment of the system with economic and mathematical realities. Particular attention was drawn to the financial and mathematical challenges associated with funding treatments for a very small patient population through resources generated by the broader economic productivity of society.

Domestic Production and Next-Generation Therapeutic Technologies

It was stated that encouraging the domestic production of medicines and genetic therapies represents a strategic necessity as an alternative to treatment models dependent on external sources. It was further noted that local pharmaceutical manufacturing is of strategic importance in overcoming current financial constraints. Strengthening domestic companies in this field is expected to accelerate processes. It was cautioned that fund-based solutions may prove insufficient from the perspective of budgetary discipline in cases such as sickle cell anemia, where treatment costs can reach millions of dollars per patient. In addition, it was stated that genetic therapies and next-generation approaches such as CRISPR are likely to fundamentally reshape current discussions in the near future and that investments should be directed toward these innovative fields.

Primary Healthcare Services and Epidemiological Surveillance

It was suggested that rare diseases should be recognized as a public health issue and that solutions should begin at the primary healthcare level. The importance of family physicians conducting "index case" follow-up, particularly for individuals diagnosed with rare diseases, and extending screening efforts to encompass family members and relatives was emphasized. Within this framework, it was stated that priority should be given to approaches aimed at "eliminating disease at its source" by identifying carriers and establishing mechanisms to prevent disease transmission.

Preventive Medicine and Diagnostic Strategies

It was stated that the focus of discussions should shift from treatment and financing toward preventive medicine practices designed to prevent diseases from occurring in the first place. It was proposed that attention should be directed not only toward treatments that ensure survival with disease but also toward models aimed at achieving complete recovery. It was emphasized that, alongside the importance of early diagnosis, the state's regulatory role should be strengthened in controlling genetic transmission and raising public awareness on these issues.

Centers of Excellence and Digital Health Applications

It was stated that, rather than directly replicating European models, Türkiye should develop its own centers of excellence tailored to national needs. In this context, the establishment of specialized structures capable of ensuring coordination was proposed. It was also noted that the Regulation on Remote Healthcare Services (telemedicine), issued by the Ministry of Health, is expected to facilitate access to specialist physicians for patients in rural areas and support educational processes.

Stakeholder Participation and Institutional Collaboration

It was stated that the representation of physicians and family associations within pharmaceutical reimbursement and evaluation committees should be strengthened. It was emphasized that, in order to balance financing-oriented approaches with realities observed in the field, patient associations should be involved more actively in decision-making processes through realistic, field-based data and insights.

Awareness, Acceptance, and Coordination: A Holistic Approach to Rare Diseases

Fazilet DURMUŞ

Deputy Director General, General Directorate of Special Education and Guidance Services, Ministry of National Education

I would like to begin my remarks as a mother, a woman, and a Deputy Director General serving within the General Directorate of Special Education and Guidance Services of the Ministry of National Education. At the same time, as the spouse of a physician and the mother of a physician, I can say that I have a certain degree of familiarity with the field.

I would like to start with a short story. Several people are having a conversation about awareness while traveling in a vehicle. The discussion becomes quite animated when the vehicle suddenly jolts, yet the conversation continues uninterrupted. After a while, the driver turns around and says, "Friends, do you realize that we just hit a goat?" Through this example, I would like to emphasize the following: even while discussing awareness, there may be situations of which we are not actually aware. For this reason, I believe that the concept of awareness needs to be reconsidered.

There are two fundamental dimensions of awareness: an individual's awareness of themselves and the awareness of others toward that individual. Particularly in studies involving individuals with autism, and based on our experience as educators, one of the most common challenges encountered in the field is that families initially struggle to accept the situation. However, once acceptance takes place, the process moves forward much more rapidly. Therefore, creating early awareness at the societal level is of great importance for ensuring a healthy progression of the process.

Self-awareness forms the foundation of this journey. In this regard, Yunus Emre's saying, "Knowledge means knowing; knowledge means knowing yourself," powerfully captures the essence of awareness. The process begins with an individual knowing themselves.

Awareness is a process that starts with the individual, extends to the household, and then spreads throughout society. Coordination, in turn, is directly related to this level of awareness. Establishing coordination among institutions is not sufficient on its own; the process must also be embraced intellectually and culturally. In this respect, acceptance and awareness emerge as two fundamental concepts that progress hand in hand.

Although budgetary constraints are an important factor in policy development processes, increasing societal awareness generates demand and momentum that can reduce the impact of such constraints. Therefore, awareness has a decisive influence on policymakers' decision-making processes.

The necessity of legislation is evident, as a legal framework is required to ensure sustainable coordination. At the same time, I believe that civil society organizations should play an active role in funding processes. When one looks at the deep-rooted cultural fabric of Anatolia, it becomes clear that civil society has always held an important place.

The phrase "the Century of Compassion," included in the "Century of Türkiye" vision put forward by Mr. Recep Tayyip Erdoğan, also supports this approach. The concept of compassion embodies mercy, love, and respect as a unified and powerful value. Bringing together volunteerism and institutional structures will create a significant transformation in this field.

When we examine healthcare service delivery, it becomes evident that the effectiveness of primary healthcare services is of critical importance. The foundation of the system is built upon individuals first seeking care at the

primary care level and then being referred to higher levels of care as needed. However, in current practice, individuals often present directly to secondary healthcare institutions, creating bottlenecks within the system.

At this point, the role of family physicians is critically important. Conducting preliminary assessments through telemedicine applications when appropriate, followed by proper referrals, could contribute to a more effective healthcare system. Diagnosis, referral, coordination, and access processes must be addressed through a holistic approach.

Similar challenges are also encountered within the appointment system. Although referrals made through family physicians can sometimes result in faster outcomes, public awareness regarding this issue remains insufficient. Therefore, society must also be educated in these areas.

It is well known that preventive healthcare services are more effective and sustainable than treatment-oriented approaches. However, achieving this requires carefully designed transitions between different levels of healthcare services.

As educators, I would particularly like to emphasize that educational settings are the most effective environments for instilling this awareness. Over time, children internalize what they learn and reflect it both in their own lives and in society. I believe that the seeds of awareness planted today will yield far stronger results in the future through the individuals who will become tomorrow's decision-makers.

The relationship between awareness and coordination can be illustrated through a simple example: just as applause initiated by one person grows stronger as others join in, coordination naturally becomes stronger as awareness increases. In this process, the role of civil society organizations is extremely critical.

Indeed, once awareness is established, coordination often develops organically.

Within this framework, I would like to conclude by referring to Mustafa Kemal Atatürk's statement, "Entrust me to Turkish physicians," and by expressing my confidence that Turkish physicians, educators, and all stakeholders in society will successfully carry this process forward.

Analysis of Pediatric Rare Diseases and Chronic Disease Progression

Moderator

The Pediatric Patient Population and Clinical Reality

It was noted that approximately 50% of individuals diagnosed with rare diseases are children, demonstrating that the issue is not only a healthcare matter but also one of education and social development. It was stated that a significant proportion of these children, even after receiving a diagnosis, continue to struggle with metabolic disorders and chronic progressive conditions that cannot be fully treated. It was emphasized that the dynamic and progressive nature of these diseases creates lifelong needs for specialized support.

Structural Challenges in Diagnosis and Access to Treatment

It was stated that the challenges of diagnosis and barriers to treatment access, which are at the center of sectoral discussions, produce much deeper social consequences within the pediatric population. From an educational perspective, it was observed that medical difficulties create direct barriers to a child's educational experience and social integration.

The Categorization Debate: The Relationship Between Rare Diseases and Disability

From the perspectives of the Ministry of National Education and the Ministry of Family and Social Services, the necessity of addressing rare diseases as a distinct category separate from disability was raised as a critical question. It was stated that existing definitions of disability may sometimes be insufficient to encompass the specific metabolic needs, episodic manifestations, and progressive nature associated with rare diseases.

Strategic Positioning Across Ministries

Although evaluating rare diseases under the broader disability framework may provide certain bureaucratic advantages, it was debated whether defining them as a separate policy area would be beneficial due to their unique characteristics, such as specialized dietary requirements, genetic counseling, and continuous medication monitoring. It was considered that such a distinction could be necessary, particularly for revising special education curricula and social support packages in a disease-specific manner.

Holistic Approach and the Human Dimension

It was observed that the inclusion of an educator's perspective brings a human dimension to the discussion that extends beyond medical terminology. Based on the reality that a child with a rare disease is not only a patient but also a student and a social individual, it was concluded that education, family, and healthcare policies should be synchronized at the intersection of these domains.

Access to the Education System and Early Warning Mechanisms

Fazilet DURMUŞ

Deputy Director General, General Directorate of Special Education and Guidance Services, Ministry of National Education

Data Sharing and the Institutional Awareness Challenge

It was determined that the greatest obstacle preventing children with rare diseases from fully benefiting from their educational rights is a lack of awareness. It was emphasized that the system's capacity for intervention remains limited when information regarding diagnosed children is not communicated to educational institutions by their families. It was proposed that an automatic alert system be established between the healthcare level where the diagnosis is made (family medicine or hospitals) and the Ministry of National Education, operating in accordance with the principles of personal data protection and confidentiality.

Balancing Constitutional Rights

It was stated that a delicate balance must be established between the confidentiality of personal data and the child's constitutional right to education. It was considered that a digital alert automatically transmitted to the relevant district directorates of national education upon diagnosis within the healthcare system would enable rapid and effective intervention in the child's educational journey.

Service Capacity and Flexibility of the Ministry of National Education

It was stated that the education system possesses a strong infrastructure for providing services to children with rare diseases and special needs. According to current data:

- Home-Based Education: Access is provided to more than 10,000 students.
- Hospital-Based Education: Continuous educational services are provided to more than 500 students.

These figures demonstrate the operational capacity of the system.

Implementation Flexibility and Educational Hours

It was stated that although regulations prescribe a minimum of 10 hours of education per week, this duration can be increased to as much as 30 hours based on the needs of children with rare diseases, and that no restrictions exist in this regard. It was emphasized that the primary challenge lies not in service provision itself, but in reaching the child who requires these services.

The Process of Accepting a Diagnosis and Parent Collaboration

It was observed that even in more common diagnostic groups, such as specific learning disabilities or autism, parents often require time to accept the diagnosis and engage with the system, and that this process becomes considerably more complex in the context of rare diseases. It was emphasized that, in order to strengthen guidance and counseling services, not only a medical diagnosis but also the family's trust in and cooperation with the system must be established simultaneously.

Strategic Coordination

It was concluded that, in the management of rare diseases, the issue of coordination should be expanded to encompass not only healthcare services but also educational services. Establishing a digital bridge extending from the healthcare system to the education system was identified as being of vital importance for ensuring the sustainable social and academic development of children with rare diseases.

Digital Ecosystems and Inter-Institutional Data Integration

Moderator

Synchronization of the e-Nabız and e-School Systems

It was emphasized that Türkiye's strong digital infrastructure should be optimized to facilitate inter-institutional

collaboration. It was observed that, through the integration of the existing e-School and e-Nabız systems, the health information and special needs of a child with a rare disease could be automatically transferred to the education system during school registration or transfer procedures. It was proposed that this integration be designed as a mechanism that informs educators solely about the supportive measures required, without overwhelming them with detailed medical information.

Psychosocial Burden and Delays in the Diagnostic Process

It was stated that the time required to receive a diagnosis for a rare disease can extend to as much as seven years and that this period of uncertainty constitutes a "nightmare" for families. It was observed that as diagnosis is delayed, the disease progresses and irreversible damage may occur, while families experience significant medical and psychological strain throughout the process.

Social Service Models and a Multidisciplinary Approach

Within the framework of international best practices, particularly examples from Germany, the importance of multidisciplinary models in which patients are not sent directly home following diagnosis was emphasized. In these models:

- **Psychological Support:** Managing the trauma experienced by families following diagnosis.
- **Intervention by Social Service Professionals:** Conducting home visits to assess the family's physical and social needs on site.
- **Development of a Roadmap:** Providing guidance to families unfamiliar with the disease regarding their rights and the services available to them.

Social Service Capacity and Target Population Analysis

It was observed that Türkiye's social services infrastructure possesses the capacity to conduct home visits. However, these services are currently focused primarily on families living below the poverty line and those actively seeking assistance. It was stated that such criteria alone are insufficient in the context of rare disease management.

Identifying "Hidden Poor" Families

It was emphasized that the system must be capable of identifying "hidden poor" families—those whose income appears adequate on paper but who are effectively placed under financial strain by the high costs associated with treatment, care, specialized diets, and medical devices required for rare diseases. It was concluded that, in order to integrate such families into social support mechanisms, a new social service screening model should be developed that focuses not only on income levels but also on expenditures and needs.

Rare Diseases from a Social Policy Perspective: Current Situation and Areas for Development

***Ayşe TÜRKAY AYDEMİR** / Representative of the Ministry of Family and Social Services*

It was stated that rare diseases are addressed as a distinct and separate area by the Ministry. Current efforts are primarily focused on the social needs that arise after the onset of disease, regardless of whether an individual is classified as a patient. Within this framework, the emphasis is placed not directly on the disease itself, but rather on the social needs of individuals and their families, as well as on raising awareness in this area.

It was noted that social assessment processes are generally carried out by specialists working within hospitals. However, there is currently no dedicated unit specifically focused on rare diseases. Nevertheless, various initiatives that touch upon this field are being implemented. For example, awareness training on genetic diseases is provided during premarital preparation programs, and participation in such training is encouraged through support programs offered to young people. It was stated that, although these initiatives have been in place for many years, they have not progressed sufficiently and should be further developed. This meeting has made it even clearer that these efforts need to be strengthened and that closer cooperation with stakeholders is required.

It was emphasized that policy development processes require the joint engagement of public resources, legislation, and civil society organizations. In this context, determining how the Ministry of Family and Social Services should position itself within the field of rare diseases, and identifying the areas in which it can contribute, should be shaped through the collective assessment of all stakeholders.

Within the framework of the Ministry's general approach, it was stated that not only income levels but also expenditure levels are important indicators. However, the systematic identification and evaluation of such expenditures do not fall directly within the Ministry's area of authority. Determining these needs is generally only possible through applications and notifications submitted by individuals. Therefore, it was noted that establishing a comprehensive and systematic identification mechanism does not appear easy under current conditions. Following identification, the allocation of resources would constitute a separate matter to be evaluated within the scope of the Ministry's available capacities.

It was acknowledged that the ideal outcome would be for these families to have access to all the support they require through social services during the application process. However, the Ministry's service portfolio covers a very broad range of areas, including children, persons with disabilities, relatives of martyrs and veterans, and social assistance programs, all of which place a significant burden on the budget.

Within this context, it was suggested that treating rare diseases as a separate policy area, establishing a dedicated unit under the disability framework, and developing disease-specific policies could be considered as a potential approach. It was further stated that any steps taken in this direction should be shaped through the contributions of all relevant stakeholders.

Coordination and Screening Programs in Preventive Healthcare Services

Moderator

Collaboration Beyond Budgetary Considerations and the Awareness Ecosystem

It was emphasized that the fight against rare diseases should not be reduced solely to financial resources and budgetary allocations. It was observed that strategic coordination among the Ministry of Health, the Ministry of National Education, and the Ministry of Family and Social Services could enhance public awareness without generating additional costs. It was stated that, through inter-ministerial synchronization, increasing participation in screening programs and disseminating early diagnosis as a state policy throughout society represent the most effective long-term methods for reducing treatment costs.

Premarital and Newborn Screening Strategies

It was observed that premarital screening programs, implemented with the objective of preventing diseases before they occur (primary prevention), serve as a critical mechanism for identifying at-risk couples. Particular emphasis was placed on expanding these screening programs in regions where consanguineous marriages are prevalent and on maximizing opportunities for early intervention during the newborn period.

The Current Status and Scope of National Screening Programs

Specialist Dr. Rahşan ÇINAR */ Ministry of Health, General Directorate of Public Health, Head of the Public Health Screening Programs Unit*

Analysis of Misinformation and Social Resistance in Screening Programs

It was observed that a significant amount of misinformation has recently emerged regarding Newborn Screening Programs (NSPs), leading to a substantial increase in screening refusals compared to previous years. It was recalled that a similar trend has been observed in vaccine refusals (approximately 200,000 cases), and it was emphasized that this situation has evolved into a public health security concern. It was stated that disruptions to this vital nationwide service due to unfounded claims must be prevented.

Strategic Target Audiences and Communication Channels

It was proposed that awareness-raising activities should extend beyond the medical boundaries of the Ministry of Health and be carried out in collaboration with institutions that have deep reach within society. In this context:

- **The Presidency of Religious Affairs (Diyanet):** Reaching broad segments of society through Friday sermons and religious guidance channels by associating screening programs with the concepts of protecting life and safeguarding entrusted responsibilities.
- **The Ministry of Family and Social Services:** Incorporating the importance of screening programs into premarital education packages and institutionalizing the positive outcomes achieved through these efforts.

Systematic Inter-Institutional Coordination and Visibility

It was stated that the fragmented efforts currently carried out by ministries and civil society organizations (CSOs) should be replaced with a more systematic, visible, and high-impact structure. The importance of establishing coordinated awareness weeks or national campaign periods, during which all institutions simultaneously communicate the same key messages, was emphasized.

Local Governance and the Strategic Role of Neighborhood Headmen (Muhtars)

***Fazilet DURMUŞ** / Deputy General Director, General Directorate of Special Education and Guidance Services, Ministry of National Education*

The Closest Point of Contact to Communities: Muhtars

It was emphasized that involving neighborhood headmen (muhtars)—the administrative units closest to households both physically and socially—is critically important in the management of rare diseases. Due to their close collaboration with Social Assistance and Solidarity Foundations (SYDVs), muhtars were identified as one of the most reliable channels for recognizing family needs, socio-economic conditions, and forms of "hidden poverty" that may not be visible through official records.

Access to Information on Genetic Predisposition and Family History

Drawing on field experiences from provinces such as Bilecik, Sakarya, and Mersin, it was noted that muhtars often possess extensive knowledge of local family structures and histories. It was stated that sensitive information—such as the occurrence of similar symptoms among relatives or the presence of hereditary conditions—can often be identified more quickly and accurately through muhtars, who serve as custodians of local institutional memory.

Facilitating Rapid Access to Social Support Mechanisms

To address challenges in identifying families genuinely in need of social support, it was proposed that the field-level verification capacity of muhtars be utilized more effectively. It was observed that muhtars could function as both verification and guidance centers, enabling families to be assessed not only according to declared income levels but also according to the substantial financial burdens created by rare diseases and their real-life impact on household circumstances.

Workshop Closing Remarks and Future Vision

Moderator - Gülnur GÖKMEN

The Power of Synergy and Collective Participation

All perspectives shared throughout the workshop, the proposed solutions, and the interactions among stakeholders were described as representing the beginning of a new era in the field of rare diseases. It was emphasized that the steps taken during the workshop have created an initial "snowball effect," which, with the continued support and commitment of all stakeholders, has the potential to grow into a comprehensive and lasting transformation.

Strategic Outcome: A Holistic Future

It was stated that the synergy generated during the meeting is expected to translate into tangible outcomes across multiple domains, including health policies, educational models, social services, and local governance. In her closing remarks, the moderator emphasized that the valuable contributions made by participants should not remain merely as ideas, but rather serve as foundational building blocks for the development of a sustainable and institutionalized rare disease ecosystem.

Workshop Process Management and Future Outlook

Moderator - Prof. Dr. Zafer ÇALIŞKAN

Deepening Discussions and the Goal of Consensus

It was observed that the perspectives presented during the workshop would be further enriched at the larger meeting scheduled for the following day through the participation of additional stakeholders, technical presentations, and diverse viewpoints. It was stated that the findings generated during the workshop would be combined with the outcomes of the next day's discussions and subjected to a comprehensive evaluation, identifying both areas of strategic consensus and chronic challenges requiring further attention. The conceptual foundation established during the workshop is intended to evolve into a concrete roadmap through these subsequent discussions.

Shared Responsibility and the Central Role of Public Institutions

From an academic perspective, it was emphasized that the ultimate responsibility and implementation authority for the process rests with public institutions. In managing a complex and multi-layered field such as rare diseases, the roles of civil society and academia were described as being primarily focused on generating evidence, developing ideas, and proposing projects. However, it was noted that the successful implementation of these initiatives ultimately depends on public authorities embracing the issue and creating the necessary policy and operational framework.

Functional Roles of Institutional and Social Stakeholders

For the sustainability of the process, stakeholder responsibilities were defined as follows:

- Public Institutions: Providing active leadership, establishing regulatory frameworks, and creating implementation mechanisms.
- Academia: Supplying scientific evidence and developing evidence-based policy recommendations.
- Civil Society and Patient Organizations: Contributing field experience, representing stakeholder perspectives, and supporting advocacy efforts.

Commitment to Continuous Transformation and Progress

It was concluded that public institutions should assume a more active and transformative role in the period ahead. Looking toward future meetings, it was expressed as a shared aspiration that current challenges will have been addressed and that discussions can focus on more advanced strategic objectives. Serving as a preparatory workshop for the major Rare Lives and Rare Diseases Summit to be held the following day, the event concluded with expressions of appreciation to all participants and a reaffirmation of the importance of collective action and collaboration.

FINAL REMARK

For a healthier future, collective action should be built upon a patient-centered governance model that strengthens public awareness and early diagnosis capacity, establishes a sustainable financial architecture grounded in value-based healthcare, and brings all public institutions together under a centralized coordination mechanism.

Participant Profile

The Rare Lives and Rare Diseases Workshop was conducted in a closed format with the participation of 40 invited stakeholders. Participants included representatives from key public institutions and organizations, notably senior officials from relevant departments of the Ministry of Health, as well as representatives from the Ministry of Family and Social Services, the Ministry of National Education, the Social Security Institution (SSI), industry stakeholders, and civil society organizations active in the field.

RARE DISEASES FEDERATION

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