TÜSEB TAÇESE
TÜRKİYE MATERNITY, CHILD AND ADOLESCENT HEALTH INSTITUTE

Childhood Rare Muscle Diseases Workshop

WORKSHOP REPORT

October 20, 2025

Online

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President's Message

Health Institutes of Türkiye (TÜSEB) leads innovative studies systematically conducted in the field of health sciences and technologies, providing scientific, technical, and financial support to practitioners and researchers. In this context, it acts with the mission of a domestic and national Türkiye in the healthcare sector and the vision of being an international leader organization focused on R&D. With the motto **"Science and Technology for Human Health,"** it aims to bring new momentum and energy to the healthcare sector. It supports all kinds of scientific and technological studies that have the potential to add value to our country and humanity in order to increase and sustain Türkiye's competitiveness.

Türkiye Maternity, Child and Adolescent Health Institute (TAÇESE) is one of the nine institutes established under **TÜSEB**. With the motto **"Healthy Mother, Healthy Child, Healthy Generations,"** our Institute's objectives include "to ensure the production, use, and benefit of techniques and methods for determining the causes, prevention, diagnosis, and treatment of diseases related to maternal, child, and adolescent health; to make collaborations and partnerships with relevant institutions and organizations in the private, foundation, or public sector for this purpose; and to support entrepreneurship in these areas."

Childhood and adolescence periods, which fall within our field of activity, are known as the periods when individuals' physical, mental, and social development is fastest. Adequate healthcare services and supportive environments provided at early ages help children and youth maximize their developmental potential. In this context, to achieve a healthy society, it is essential to develop and implement appropriate health policies for these specific life periods.

The **"Childhood Rare Muscle Diseases Workshop"** was held with the aim of discussing current problems and developing solution proposals regarding **Duchenne Muscular Dystrophy (DMD)** and **Spinal Muscular Atrophy (SMA)**, which are rare childhood diseases. The workshop is a continuation of the **"DMD Awareness Meeting"** held on September 17, 2025 with the participation of DMD patients and their families. At the event, the outputs obtained from DMD families were evaluated in terms of being transformed into projects, and current developments and planned research on SMA were discussed. The results of the meeting, held with the participation of the relevant department presidency of the **Ministry of Health** and specialist academicians, have been systematically reported along with views and suggestions.

Hoping the outcome will be beneficial to our country...

Prof. Dr. Şirin Güven

President of the Türkiye Maternity, Child and Adolescent Health Institute

Executive Summary

The **Childhood Rare Muscle Diseases Workshop**, organized online by the **Türkiye Maternity, Child and Adolescent Health Institute (TAÇESE)** under **TÜSEB** on October 20, 2025, was held to evaluate the current situation and develop solution-oriented policies in the field of **Duchenne Muscular Dystrophy (DMD)** and **Spinal Muscular Atrophy (SMA)**. This workshop, which serves as a continuation of the **DMD Awareness Meeting** on September 17, 2025, was carried out with the participation of **Ministry of Health** representatives, specialist academicians, and researchers.

The main topics that stood out as a result of the workshop are as follows:

- **Screening and Preventive Services:** The success of the premarital (more than 2.1 million people) and newborn (3 million babies) screening programs conducted in the field of SMA is evident, and through this, a decrease in the incidence of the disease is observed. For DMD, since there is no definitive cure yet, priority should be given to carrier screening, mandatory genetic counseling, and free PGT (Preimplantation Genetic Testing) practices instead of newborn screening.
- **Service Delivery and Follow-up:** There is a need to increase the functionality of the 24 neuromuscular disease units across the country, to create a national follow-up algorithm for DMD, and to establish an uninterrupted chain of care that will enable the transition of patients from childhood to adult units after the age of 18.
- **Data Management:** Increasing data entries and data quality in the Rare Diseases Data System (NHVS) established by the Ministry of Health in 2022 will support evidence-based decisions.
- **Domestic R&D and Innovation:** It is critical to incentivize projects aimed at the domestic production of genetic diagnostic kits, mRNA-based drug studies, and medical devices, especially cough assist devices which experience difficulties in reimbursement coverage.
- **Ethical and Social Approaches:** Society's health literacy should be increased against "peddling of false hope" and unsupervised charity campaigns directed at gene therapies lacking sufficient scientific evidence or used off-label, and collaborative work should be done with other institutions regarding legal processes.
- **Project Collaborations:** Under the coordination of TÜSEB and the Ministry of Health, the initiation of multicenter clinical trials such as the post-gene therapy follow-up of SMA and the analysis of newborn screening results was evaluated.

In conclusion, this workshop presents a roadmap centering on preventive health services, standardized care processes, and domestic production in the management of childhood rare muscle diseases. In the context of the topics addressed, it has established a scientific and service-based foundation that will contribute to Türkiye's vision of a productive healthcare model.

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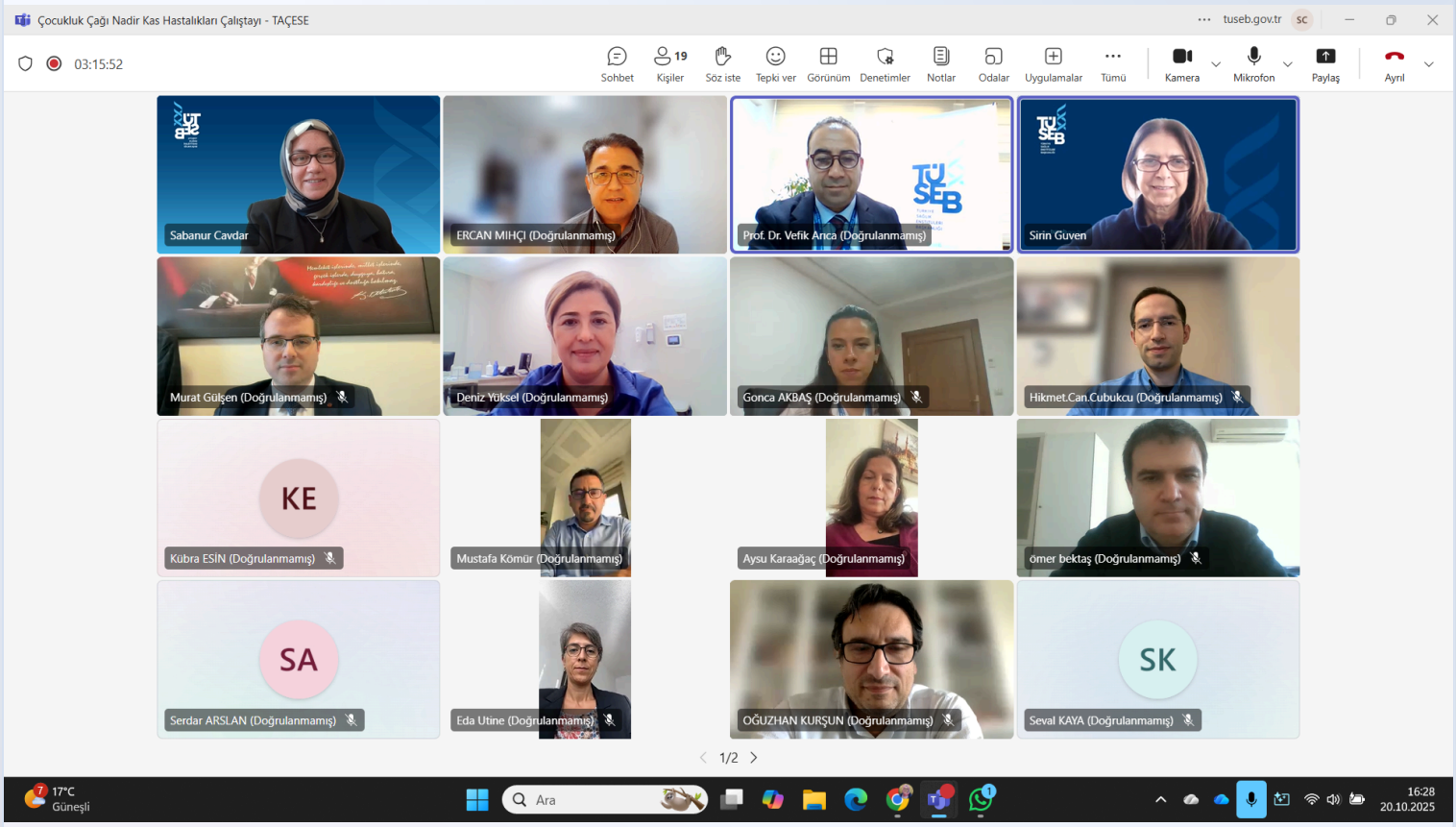

Childhood Rare Muscle Diseases Workshop

Online

OCTOBER 20, 2025 • ONLINE

1. Childhood Rare Muscle Diseases Workshop

1.1 Objective and Scope

Organized by the **Türkiye Maternity, Child and Adolescent Health Institute (TAÇESE)** affiliated with the **Health Institutes of Türkiye**, the **"Childhood Rare Muscle Diseases Workshop"** was held to evaluate the current situation, address the problems encountered in service delivery, and develop solution proposals in the fields of **Duchenne Muscular Dystrophy (DMD)** and **Spinal Muscular Atrophy (SMA)**, which are rare neuromuscular diseases seen during childhood.

The workshop serves as a continuation of the **"DMD Awareness Meeting: Experiences and Solution Proposals from the Patient's Perspective"** event held on September 17, 2025, and aimed to reflect the outputs obtained from patient and family experiences into the improvement of healthcare services and research and project development processes.

The event was planned with the participation of the **Ministry of Health General Directorate of Health Services Department of Autism, Mental Special Needs and Rare Diseases**, specialist academicians from the pediatric neurology, genetics, and physical therapy and rehabilitation fields of universities, and the **TÜSEB-TAÇESE** team to evaluate current developments, ongoing studies, and collaboration opportunities in the field of **DMD** and **SMA**.

1.2 Workshop Program

The **Childhood Rare Muscle Diseases Workshop**, held online on October 20, 2025 by the **Health Institutes of Türkiye (TÜSEB) Türkiye Maternity, Child and Adolescent Health Institute (TAÇESE)**, provided a comprehensive evaluation environment for strengthening healthcare service delivery and increasing multidisciplinary collaboration in the field of rare muscle diseases.

At the opening of the workshop, moderated by **Prof. Dr. Vefik Arıca**, following the speeches made by **Prof. Dr. Kürşat Bora Çarman**, **Dr. Murat Gülşen**, and **Prof. Dr. Şirin Güven**, the final report of the "DMD Awareness Meeting" conducted by **TAÇESE** was presented by **Dr. Sabanur Çavdar**. In this session, outputs derived from the views of **DMD** patients and families were evaluated in terms of patient support, care coordination, and project development.

Subsequently, the current efforts and future plans of the **Ministry of Health** regarding rare muscle diseases were conveyed by **Dr. Murat Gülşen, Head of the Department of Autism, Mental Special Needs and Rare Diseases at the Ministry of Health General Directorate of Health Services**; suggestions for strengthening multidisciplinary centers, **TÜSEB-TAÇESE**

project supports, and developing ongoing projects in diagnostic-treatment processes were discussed.

In the workshop, problems such as access to treatment for rare diseases, diagnostic processes, the effectiveness of multidisciplinary centers, and domestic drug/device development projects were addressed, and current developments and ethical issues in **SMA** and **DMD** treatments were emphasized. Consensus points were discussed regarding the initiation of clinical studies in this field and how **TAÇESE** could support these projects.

The workshop outputs offer an institutional roadmap aimed at increasing the quality of service in the field of rare muscle diseases, strengthening patient and family support, and developing research-oriented policies.

Program Flow

TIME	SESSION	SPEAKER / RESPONSIBLE
14.00-14.10	Opening Speeches	Prof. Dr. Kürşat Bora ÇARMAN - *Eskişehir Osmangazi University* *Faculty of Medicine, Department of Pediatric Neurology* Dr. Murat GÜLŞEN - *MoH General Directorate of Health Services* *Head of Department of Autism, Mental Special Needs and Rare Diseases* Prof. Dr. Şirin GÜVEN - *TAÇESE President*
14.10-14.30	Presentation of TAÇESE "DMD Awareness Meeting: Experiences and Solution Proposals from the Patient's Perspective" final report	Dr. Sabanur ÇAVDAR
14.30-15.00	Actions taken and plans by the Ministry of Health in the field of rare muscle diseases	Dr. Murat GÜLŞEN
15.00-15.45	Support proposals for DMD patients and their families, strengthening multidisciplinary centers, TÜSEB TAÇESE project supports	-
15.45-16.30	Evaluation of diagnostic and therapeutic projects in SMA and DMD	-
16.30-17.00	Evaluation and Closing	-

Figure 1. Childhood Rare Muscle Diseases Workshop Program

1.3 Workshop Participants

The **"Childhood Rare Muscle Diseases Workshop"** was held with the participation of the **TAÇESE** team, as well as representatives from the **Ministry of Health General Directorate of Health Services Department of Autism, Mental Special Needs and Rare Diseases**, and specialist academicians from the pediatric neurology and genetics fields of universities.

2. Workshop Notes

2.1 Opening Speeches

The workshop began with opening speeches moderated by **Prof. Dr. Vefik Arıca**.

Prof. Dr. Kürşat Bora Çarman touched upon the scientific collaborations in the field of neuromuscular diseases that have been ongoing for many years. Expressing that this workshop held with **TÜSEB** solidifies the experience of the neuromuscular diseases scientific committee at a different institutional level, Prof. Dr. Çarman stated that studies like this workshop coordinated by **TÜSEB-TAÇESE** would contribute to the development of new projects and innovative approaches.

Dr. Murat Gülşen addressed the participants on behalf of the **Ministry of Health General Directorate of Health Services Department of Autism, Mental Special Needs and Rare Diseases**. Dr. Gülşen expressed great satisfaction with the efforts and collaborations of all stakeholders of the Ministry of Health in the field of rare diseases. Thanking the workshop participants, he shared his wishes for the workshop to be productive.

Finally, **Prof. Dr. Şirin Güven** delivered the opening speech on behalf of the **Türkiye Maternity, Child and Adolescent Health Institute (TAÇESE)**, the host institution of the workshop. Prof. Dr. Güven touched upon the founding purpose of **TÜSEB**, established in 2014, and emphasized that studies based on a productive healthcare model are being conducted and that institutes are a significant part of the process leading "from idea to product" in health sciences. She conveyed the mission, vision, and motto (**"Healthy mother, Healthy child, Healthy generations"**) of **TAÇESE**, one of TÜSEB's 9 institutes, introduced her team, and provided information about other workshops and projects carried out by the Institute. Prof. Dr. Güven stated that as **TAÇESE**, they aim to develop evidence-based, multi-stakeholder, and solution-oriented approaches to problems in the field of maternal, child, and adolescent health. She expressed that the findings obtained from the **Experiences and Solution Proposals from the Patient's Perspective: DMD Awareness Meeting** held on September 17, 2025 formed the basis of this workshop, and the views emerging from here would guide new project and policy proposals. She stated that the meeting was planned to evaluate the current situation regarding the **Duchenne Muscular Dystrophy (DMD)** and **Spinal Muscular Atrophy (SMA)** diseases, address the problems experienced by patients and families, and develop solution proposals. Furthermore, she emphasized that such meetings held in collaboration with the **Ministry of Health**, universities, and **TÜSEB-TAÇESE** would contribute to strengthening the scientific and service-based approach in the field of rare diseases.

SPEAKER **Dr. Sabanur Çavdar - Türkiye Maternity, Child and Adolescent Health Institute (TAÇESE) - Researcher / Public Health Specialist**

- The DMD Awareness Meeting Final Report Presentation summarized the findings derived from the experiences of Duchenne Muscular Dystrophy (DMD) patients and their families, framing the thematic structure of the workshop.
- One of the most important outputs of this meeting, where DMD awareness was addressed not only medically but also in its social and psychosocial dimensions, is the direct inclusion of patient and family views into the process; this approach has enabled the development of policy and project proposals centered on the patient experience.
- The presentation shared the findings of the "DMD Awareness Meeting: Experiences and Solution Proposals from the Patient's Perspective" held on September 17, 2025. The meeting was organized in a hybrid format with 70 participants taking part; representatives of DMD associations, patients, families, academicians, TÜSPE R&D specialists from TÜSEB Institutes, and representatives of the Child, Adolescent and Reproductive Health Unit of the Ministry of Health Istanbul Provincial Health Directorate Presidency of Public Health Services were included in the process. A wider participation was achieved thanks to the participating associations sharing the meeting links with families via their own networks.
- Meeting data was evaluated by conducting content analysis on the voice recording and the transcription of online comments. Additionally, written opinions submitted later by the participants were included in the report. The findings were grouped under the following themes in line with the needs voiced by patients and families:
- **Multidisciplinary Muscle Diseases Centers**
    - Increasing the functionality and capacities of muscle disease centers currently existing in various cities of our country was brought to the agenda.
    - It was suggested that centers offering holistic services to DMD patients, containing neurology, cardiology, physical therapy, psychiatry, dietitians, and social workers, be established in university and city hospitals.
    - It was emphasized that managing examination, report, and treatment processes in a single center would both reduce the patient burden and ensure service continuity.
    - The need for effective delivery of genetic counseling was stated.
- **Home Care and Rehabilitation Services**
    - It was expressed that home physiotherapy practices, examples of which exist as pilots in some centers, should be expanded/reinitiated, and psychological support and device procurement processes should be managed at home.
    - Furthermore, it was suggested to strengthen coordination with home health services and deliver technical team support to families.
- **Facilitating Health Report Processes**

- It was stated that reports requiring annual renewal pose a severe burden on families, and considering the chronic and progressive nature of the disease, issuing long-term or permanent reports would be appropriate.
- It was also emphasized that the drops in disability rate evaluations after age 18 obstruct access to services.

- **Psychosocial Support Programs**
  - The existence of the need for psychosocial support for both patients and caregivers was emphasized.
  - It was recommended to provide regular psychological support especially for individuals with DMD in adolescence, and to establish group therapies and solidarity programs for families.
  - The necessity of developing support models that would alleviate the mental burden on caregivers was voiced.
- **Right to Education and Social Participation**
  - It was emphasized that obstacles seen in practice despite being stated in legislation, such as resolving the barriers children with DMD face in school enrollments and providing accessible classrooms in compliance with Ministry of National Education regulations, must be removed.
  - The need to implement practices that would guarantee equal opportunity in education for both those in formal education and young adult individuals with DMD wanting to continue higher education was stated.
- **Domestic Drug and Device Development**
  - Participants noted that the difficulties experienced in accessing drugs and medical devices used in the DMD disease impose a severe burden on families.
  - Delays in the procurement of products such as cough assist devices, respiratory support systems, supported wheelchairs, pressure-reducing cushions and beds, transfer lifts, medical bath/toilet apparatus, and communication support devices, and their exclusion from reimbursement coverage are among the most frequently voiced problems.
  - Families expressed that these devices are of vital importance for patients' quality of life and respiratory functions, but because a portion of them is solely imported, access has become difficult in terms of both cost and time.
- **Protection Against Unethical Practices**
  - It was recommended to strengthen information and auditing mechanisms against "peddling of false hope" practices lacking scientific basis, and the risks of overseas stem cell treatments and similar procedures.
- In conclusion, considering the priority expectations of DMD families, the priority study areas based on needs in DMD are summarized as follows:
  - Strengthening multidisciplinary centers (especially for Istanbul)

- Supporting domestic drug/device development processes
- Providing genetic screening programs and family counseling
- Developing psychosocial support systems
- Expanding the scope of home health services
- Ensuring equity in access to education, devices/drugs

- It was expressed that these headings derived from the meeting held with DMD patients and their families will provide input to new projects to be developed by TAÇESE and contribute to the Ministry of Health's rare diseases policies.

Figure 2. Priority Expectations of DMD Families (Source: TAÇESE "DMD Awareness Meeting: Experiences and Solution Proposals from the Patient's Perspective" Final Report Presentation)

SPEAKER **Dr. Murat Gülşen - Ministry of Health General Directorate of Health Services; Department of Autism, Mental Special Needs and Rare Diseases - Head of Department**

- Dr. Murat Gülşen greeted the participants and began his presentation, summarizing the structure and area of responsibility of the department. He noted that the department was established in 2020, its founding process was based on the Turkish Grand National Assembly (TBMM) Investigation Committee reports, and it covers rare diseases as well as autism and mental special needs units.
- He stated that the department's strategic plan and development plan duties are strengthening the capacity of healthcare services offered to individuals with special needs and rare diseases; expanding screening programs to diagnose rare diseases at an early stage; increasing the number of treatment and research centers regarding rare diseases; and

facilitating the access of individuals with mental special needs and their families to effective healthcare service delivery.

- He stated that the department's duties and responsibilities within the scope of the General Directorate of Health Services Directive are awareness studies, screening studies, identifying problems experienced in the health system, determining service standards, creating and updating application guidelines, making and monitoring technical regulations, collaborating with national and international organizations, conducting studies aimed at facilitating access to healthcare services, establishing a national registry system, collecting and analyzing data, paving the way for scientific studies and supporting ongoing ones, and working on advanced centers to be established.
- He shared the studies carried out specifically on rare muscle diseases, which is the topic of today's workshop.
- He stated that the department has a Neuromuscular Diseases Scientific Committee, and that the studies conducted with the scientific committee are mostly related to SMA.
- He summarized the stages of what can be done by the health system for a rare disease as follows: Newborn screening → Early diagnosis → Early and free access to treatment → Carrier screening → Ensuring the birth of healthy babies via free PGT (Preimplantation Genetic Testing) and IVF → Guidelines → Specialized units → Access to possible treatment alternatives.
- He mentioned that there are studies indicating orphan drugs can be an area pharmaceutical companies focus on to cover financial deficits, and that globally, orphan drug sales are progressively increasing over the years. He stated that gene therapy studies are continuing all over the world.
- He noted that as the Department, they are conducting various studies regarding rare diseases:
    - **Reimbursement Model Study**: He stated that a study involving health technology assessment, modeling, factor analyses, and cost estimations was conducted, and this study was presented in the 2023 report titled "Making Reimbursement Decisions in Rare Diseases Using Health Technology Assessment Methods: Multi-Criteria Decision Analysis and Value-Based Payment Examples."
    - **Rare Diseases Health Strategy Document and Action Plan (2023-2027)**: He stated that an action plan to serve as a guide for planning in rare diseases was created for the first time for our country by evaluating country examples and receiving opinions from all relevant institutions and organizations. He mentioned that they worked with the relevant institutions for five main headings, 42 targets, and 44 activities.
    - **Rare Diseases Data System**: He stated that the rare diseases data system was established in April 2022 to respond to the need, as there were no diagnostic codes for rare muscle diseases in the ICD-10 diagnostic coding in the system. He said it was established in collaboration with TÜSEB and alongside the General Directorate of Information Systems. He noted that the data of all patients can be entered by

healthcare institutions, and so far since 2022, data pertaining to a total of 19,550 patients from 155 institutions have been processed. He expressed that there are over 8000 known rare diseases, and data entries belonging to 1802 different rare diseases are present in the system. However, he stated that the data entries are not sufficient. For example, he indicated that although we know the number of SMA patients is higher, there are 333 data entries for 175 unique patients in the system since 2022. He stated that data tracking can be ensured when clinicians seeing rare diseases communicate with the HBYS (Hospital Information Management System) representative and state their desire to make a Rare Disease data entry. He reminded that the data to be entered into the rare diseases data system is stored at the Ministry within the scope of KVKK (Personal Data Protection Law). He mentioned that the data entry of 597 unique DMD patients is also present in the system.

- He summarized the work done over the years in the field of SMA as follows:
    - He stated that we were one of the first countries to reimburse Type 1 SMA treatments in 2017, and Type 2 and 3 SMA treatments (Nusinersen Sodium) in 2019.
    - He noted that they ran a kit development call jointly with TÜSEB in February 2021, and subsequently, Premarital SMA Carrier Screening was launched in December 2021.
    - In 2021, the Neuromuscular Disease Scientific Committee Regulation was issued, in 2022 the data system was established, and the SMA Clinical Protocol was created (Revision efforts for the Clinical Protocol will be initiated).
    - He stated that the committee, initially established as the SMA Scientific Committee and holding its first meeting in 2020, continues its meetings under its new name as the Rare Neuromuscular Diseases Scientific Committee.
    - Evaluating the SMA screening numbers, he shared statistics indicating that according to August 2025 data, 2,157,000 individuals were screened premaritally, out of which 1,744 couples were suspected of SMA carriage, showing via calculation that for every 10,000 screenings, 8 couples have the potential to give birth to an SMA child. In the newborn SMA screening, 3 million babies were screened and 504 were referred.
    - He relayed that the SMA informative public service announcement work prepared with the scientific committee is ready, that PGT services can be offered in some public hospitals and private hospitals for diseases like SMA and DMD, and that SMA treatment centers have been designated. He noted that the designation of SMA treatment centers was done upon the request of SSI, due to the need to specify where the drug could be prescribed and administered.
    - He provided information about the neuromuscular disease units established across the country. He noted that there are currently 24 Neuromuscular Disease Units in 15 provinces. He expressed that the goal in the centers is to complete the examination, testing, consultation, and result processes on the same day.
    - He stated that the Center of Excellence Rare Diseases Field Criteria Study is being conducted with TÜSEB - TÜSKA (Turkish Institute for Quality and Accreditation in

Healthcare); and that one of the rare diseases sub-study areas is the Rare Neuromuscular Diseases Center of Excellence Sub-Working Group.

- He noted that there were more than a total of 25 thousand applications to the neuromuscular disease units in 2024.
- The SMA Clinical Quality Indicators were published in February 2025. This guideline includes premarital and newborn screening rates, premarital and genetic counseling rates, time between newborn screening and starting medication, survival of SMA patients, and multidisciplinary evaluation statuses. This guideline, prepared in collaboration with the scientific committee, is considered a good effort aimed at increasing service quality.
- He stated that the Department conducts statistical studies. He expressed that in their evaluation, looking at findings regarding the efficacy of Nusinersen treatment, they demonstrated that those who did not receive a loading dose of Nusinersen had a 3.8 times higher risk of death over 2 years compared to those who did. Furthermore, evaluating the effect of early treatment tied to screening, he stated they observed that the risk of death at any time within a 2-year period was 2 times higher in the pre-screening group compared to the post-screening (delayed treatment) group. He also stated that they observed a decrease in SMA incidence after 2020 as a result of awareness campaigns and screenings.

- **Zolgensma (Gene Therapy) and Peddling of False Hope**
  - It is necessary to carry out data communication with the Ministry of Interior regarding Zolgensma. Attempts are made to track patients receiving Zolgensma treatment by linking them with campaign records. It is known that some children receiving this treatment are also receiving Nusinersen and Risdiplam treatments, whereas normally a patient receiving Zolgensma should not receive Nusinersen.
  - He mentioned that the SMA Clinical Protocol was issued in collaboration with the scientific committee, and the DMD Clinical Guideline was issued in collaboration with the relevant department (Department of Research and Development and Health Technology Assessment).
  - He expressed that there are findings showing that the clinical status of SMA Type 1 patients improves with the administration of Nusinersen treatment in the early period after screening from among the SMA treatments (stating that before treatment, 90% of Type 1 patients were lost by age 2, whereas with screening and early treatment, there are now Type 1 patients who can walk).
  - He mentioned that one of the conducted studies is an article where 4-year follow-up results of 310 SMA Type 1 patients in Türkiye were published, presenting an increase in CHOP-INTEND scores in patients who started treatment early (0-3 months).
  - On the point of "peddling of false hope," he expressed that a physician in Dubai was meeting with SMA patients, and this physician even accepted tracheostomy patients (who have no indication for Zolgensma); stating that this was done in hotels like a tourist trip.

- Since campaign permissions are granted by governorships, governorships can seek the opinion of the Ministry of Health. He relayed that even if a negative opinion is stated when asked, courts permit charity campaigns under the justification of "what if it helps, we cannot know," and because society is compassionate, funds are collected and patients can go receive the treatment and return.
- He noted that campaigns can be initiated for tracheostomy (off-label) patients.
- To monitor these campaigns, since applications for charity campaign requests are made to them, he stated that studies regarding the fate of patients who go abroad and receive treatment are conducted by requesting data from the Ministry of Interior.
- Additionally, he stated that as of October 2025, the condition of specifying the Zolgensma receipt status has been added to reports. He expressed that in the future, there might be a practice of not reimbursing other drugs to patients who have received Zolgensma.

- **DMD (Duchenne Muscular Dystrophy) Treatment:**

  - He emphasized that the DMD patient group has also been actively working to get gene therapy for the past year, but there is still no clinical study proving the efficacy of this drug.
  - Regarding the Elevidys gene therapy (Sarepta), he stated that there is no EMA-NICE approval, Phase 3 studies are available, and FDA approval exists. However, he shared the information that US shipments were halted due to 3 developing deaths, and it was later opened to a restricted group.
  - It was stated that there are immense doubts about the drug's efficacy and no study showing its efficacy; a Nature study revealed no significant improvement in the NSAA score at 52 weeks, and although some secondary outcomes appeared numerically in favor of treatment, there was no statistical significance.
  - Upon the directive of our Minister of Health to increase communication with TÜSEB, he stated that under the guidance of the Scientific Committee, they have initiated two studies planned to be conducted in collaboration with TÜSEB: (1) Post-gene replacement therapy follow-up of SMA disease; (2) Results of Nusinersen treatment following newborn SMA screening.

3. Discussion Session - Findings and Suggestions

At the beginning of the discussion session, it was stated that the presentations made in the earlier parts of the workshop complemented each other and that the purpose of this section was to develop concrete solution proposals with views coming from the field and academia, based on the information presented. It was emphasized that the session was not merely an evaluation where problems would be relisted, but a phase where actionable policy and project proposals would be shaped.

Participants were asked to share their views and suggestions within the framework of the topics that stood out at the **Duchenne Muscular Dystrophy (DMD)** awareness meeting. It was reminded that these headings cover multidisciplinary centers, home care services, health report processes, psychosocial support, the right to education, domestic product development, and protection against ethical practices.

The aim of the session was expressed as clarifying the collaboration and project subjects that could be carried out under the coordination of **TAÇESE** by discussing the current situation and needs under these headings. It was emphasized that the discussion, carried out with the contributions of the participants, would be an application-oriented session aiming to transform feedback from the field into policy proposals.

This section of the report presents summarized contents regarding the findings and suggestions of the workshop participants.

- **Rare Diseases Data System and Data Entry Deficiencies:** The failure of physicians to make rare disease data entries due to busyness at the centers lowers data quality. To increase data quality, it is suggested to assign a dedicated medical secretary responsible for this task at the centers.
- **R&D Manpower:** It was noted that physicians in Türkiye work largely in a "service" oriented manner, and that a research and domestic product development ecosystem like in global pharmaceutical companies and the number of researcher physicians are insufficient. On the other hand, it was also emphasized that there are many academicians and teams working in the field of rare diseases in our country, and there are teams ahead of many countries closely following international and national developments.
- **Planned Clinical Studies:** It was discussed that multi-center clinical studies could be planned in collaboration with TÜSEB and the Ministry of Health. Some project management suggestions were shared for multi-center studies. To accelerate and facilitate

the study, it was proposed to appoint one chair (project manager) from each center and collect all data through that responsible person. This will both reduce ethical issues and speed up the data collection process.

- **Importance of Preventive Medicine:** It was emphasized that the prevention of rare diseases (especially SMA and DMD) is more critical than early treatment.
- **PGT and Genetic Counseling:** To increase the effectiveness of the screening program and ensure families do not give birth to another child with SMA, it was argued that the Ministry of Health and the state need to direct these families to free PGT (Preimplantation Genetic Diagnosis) and IVF opportunities. It was suggested to identify families needing genetic counseling with a coding like a "red light" in the Family Medicine system, thereby making this counseling mandatory.
- **DMD Genetic Screenings:** In the current situation, it was stated that newborn screening for DMD is not a priority, because the purpose of newborn screening is to detect treatable diseases, and DMD currently has no definitive cure. Instead, it was emphasized that the focus should be on genetic counseling. Premarital screenings, on the other hand, ensure that genetic counseling is provided to families. These screenings will also prevent high treatment costs.
- **Importance of Early Diagnosis and Genetic Screening Programs:** It was stated that early diagnosis could be achieved through carrier screenings for DMD just as in SMA, which would both decrease the clinical burden and facilitate the early management of the disease. Newborn screening for DMD was stated as not a priority at this stage (on the grounds that it has no definitive cure yet and the majority of cases are passed down from the mother). However, it was suggested that "mandatory genetic counseling" should be provided to families with affected children and preimplantation genetic diagnosis (PGT) should be expanded in a free/controlled manner.
- **Domestic Genetic Diagnostic Kits:** Furthermore, it was expressed that developing domestic genetic testing kits would both speed up the diagnostic process within the country and reduce foreign dependency. It was emphasized that investments in this direction would strengthen both the scientific capacity and the financial sustainability of the health system.
- **Non-Screening Cases (SMA):** Although mandatory screening is done for new marriages, cases are seen in the second or third children of previously married couples (those married before the screening program), and this situation was thought to be the reason for the glitch in the incidence drop post-screening.
- **Effectiveness of Multidisciplinary Muscle Disease Centers:** While the existence of centers is beneficial, it was expressed that some centers operate passively or cannot host the necessary experts on a full-time basis.
- **Practice Differences in Centers:** It was noted that there can be practice differences between centers in the field of neuromuscular diseases. In some centers, genetic counseling, psychosocial support, and physiotherapy services are delivered strongly, whereas in others, these services are offered in a limited capacity. For this reason, it was

emphasized that setting a common service standard at the national level is mandatory. It was voiced that establishing a shared knowledge platform where centers could share their internal protocols and good practice examples would both ensure that newly diagnosed patients are directed to the right centers and increase participation in clinical trials.

- **Center Criteria:** During the establishment of multidisciplinary centers, it was noted that feasibility studies are conducted and the presence of necessary team members is checked. However, a review of these criteria can be considered.
- **Follow-up Arrangements:** It was suggested that having patients tracked en masse by a transdisciplinary team in large centers (like universities), perhaps every six months instead of all the time, might be more efficient. An application could be designed by planning for them to come for observation at defined tracking times at certain intervals to each center, ensuring that physicians specialized in the neuromuscular field are together during that period. It was stated that telehealth applications could also be initiated in this way.
- **Follow-up Processes:** Attention was drawn to the differences between the follow-up processes of SMA and DMD patients. It was stated that strong coordination is achieved between centers following gene therapy applications in SMA, whereas this cohesion does not exist for DMD patients. The lack of a common algorithm for DMD regarding which tests will be done at what intervals, which branches will be involved in the follow-up, and how patient care will be organized was brought to the agenda. Therefore, it was stressed that creating a national follow-up algorithm for DMD is a priority need. Additionally, it was voiced that families are forced to apply to different hospitals every year during report renewal processes, creating a situation that is exhausting both financially and psychologically.
- **Continuous Care:** It was noted that DMD patients become untraceable in the system during the transition process to adult health services after age 18, so an uninterrupted chain of care between childhood and adulthood must be created. Defining this transition process with an official guideline will ensure continuity in patient follow-up.
- **Home Care and Rehabilitation:** It was strongly emphasized that "Home Care" and "Rehabilitation" (a multi-disciplinary job that needs to be done at a hospital) must absolutely be separated from each other. Continuing the practice of physiotherapists going to the home, which families previously received via municipalities or pilot hospitals, should be evaluated. For qualified service delivery, providing rehabilitation services at a hospital takes priority.
- **Health Reports (ÇÖZGER) Problems:** It was stated that especially due to the differences in approaches in evaluating disability rates after age 18, there is a drop in disability rates despite the progressive nature of the disease, leading to grievances.
- **Education and Transportation:** Problems related to the Ministry of National Education (MoNE) (school access, lack of teacher/administrator awareness) were brought to the agenda.
- **School Integration:** A school integration project, like in the US model, where a school health team manages the patient at school and a team from the hospital receives

feedback to prevent the child from being deprived of education, could be initiated.

- **Transportation Problem:** The transportation problem, the need for patients to be transferred to access all kinds of services, was highlighted as one of the most prominent problems families experience; it was suggested to develop a specific model (perhaps through a pilot project) to solve this issue.
- **Medical Device Call:** He suggested that TÜSEB open a general project call for medical devices related to rare diseases (especially those outside SSI reimbursement, like cough assist devices). This could increase domestic production opportunities.
- **mRNA Therapy Discussion:** The mRNA-based domestic drug study in DMD treatment was brought to the agenda. Scientific Committee members stated that mRNA poses major question marks as a therapeutic molecule due to it being an epigenetic mechanism and its expression variability depending on tissue and time. Furthermore, it was emphasized that merely increasing dystrophin amount does not mean it is a functional dystrophin. It was stated that written opinions could be requested from specialist academicians along with study results.
- **Artificial Intelligence Applications:** It was suggested that inviting TÜSEB Artificial Intelligence Institute experts to future meetings and receiving AI support on advanced innovative subjects like drug development and molecule creation would be beneficial.
- **Peddling of False Hope and Treatment Decisions:** In neuromuscular diseases (especially SMA), patients making treatment decisions themselves and bypassing doctors/scientists was identified as the biggest problem. Courts granting permission for charity campaigns with the justification of "what if it helps" causes patients to travel abroad (for example, even for Zolgensma for tracheostomy patients who have no indication). It was noted that there are obstacles to preventing this situation. It was predicted that DMD patients, like SMA patients, will actively launch campaigns for gene therapy, but there is a lack of proven clinical studies demonstrating the efficacy of the mentioned drug.
- **Charity Campaigns:** It was highlighted that charity campaigns for SMA in society might not be evidence-based, that social sensitivity could contribute to the support of off-label practices, therefore, correctly informing society on the subject is critical, and the importance of increasing health literacy was emphasized.
- **Genetics Project Workshop:** To evaluate projects in the Genetics and Biotechnology area, the intent to organize a separate workshop with the participation of neurology and genetics experts, together with TAÇESE, the Biotechnology Institute, and the Public Health Institute, was stated.
- **Clinical Study Team:** Specialist academicians expressed their readiness to contribute to a joint SMA clinical study and to serve as project managers.
- **Future Meeting Framework:** Noting that most of the problems discussed in the workshop (access to treatment and care) are under the responsibility of the General Directorate of Health Services and Public Hospitals, it was emphasized that TÜSEB and the Scientific

Committee should focus on more forward-looking, innovative projects. It was stated that the findings in the workshop would be shared with relevant parties.

- **Increasing Burden of Rare Diseases:** It was reminded that while the topics discussed in the workshop focused on SMA and DMD, rare diseases are on the rise. It was noted that future-oriented studies are critical.

4. Evaluation and Conclusion

The **Childhood Rare Muscle Diseases Workshop** held on October 20, 2025 has set a concrete roadmap for the future by analyzing the current situation, especially in the field of **Duchenne Muscular Dystrophy (DMD)** and **Spinal Muscular Atrophy (SMA)**.

- **Multidisciplinary Approach and Strengthening Centers:** One of the most important outputs of the workshop is the necessity of not just increasing the number of muscle disease centers, but also standardizing their functionality and capacities.
- **Screening Programs and Genetic Counseling:** Prevention is critical in the fight against rare diseases. The positive results (incidence drop and early treatment success) of premarital and newborn screening programs successfully implemented in SMA have been demonstrated with data. For DMD, due to the current lack of definitive treatment, prioritizing carrier screening, genetic counseling, and free PGT (Preimplantation Genetic Testing) opportunities over newborn screening takes priority.
- **Domestic Drug and Device Development:** To reduce foreign dependency, the goal is to strengthen the R&D ecosystem with TÜSEB's support for domestic genetic diagnostic kits, medical devices (cough assist devices, etc.), and mRNA-based drug studies.
- **Data Management and Quality:** To increase the effectiveness of the "Rare Diseases Data System," clinicians should be encouraged to enter data, and the assignment of medical secretaries responsible for this process at the centers should be evaluated.
- **Service Continuity:** An official guideline organizing the transition of DMD patients to adult health services after the age of 18 must be created.
- **Ethical and Social Protection:** Health literacy should be increased, and auditing mechanisms tightened against unscientifically grounded or off-label gene therapy campaigns publicly termed "peddling of false hope".
- **Project Collaborations:** Under the coordination of TÜSEB and the Ministry of Health, the initiation of multicenter clinical trials such as the post-gene therapy follow-up of SMA and the analysis of newborn screening results was evaluated.
- **Psychosocial and Educational Support:** The social needs of patients and families—such as facilitating report processes, providing appropriate physical conditions in schools, and expanding home care services—should be brought to the agenda in policies and practices, not just medical ones.

In conclusion, this workshop presents a roadmap centering on preventive health services, standardized care processes, and domestic production in the management of childhood rare muscle diseases. In the context of the topics addressed, it has established a scientific and

service-based foundation that will contribute to **Türkiye's vision of a productive healthcare model**.


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