

THE 2ND *TBL1XR1* GLOBAL FAMILY FORUM & THE 1ST INTERNATIONAL SCIENTIFIC CONFERENCE ON *TBL1XR1*



Families, Research, and Future Directions
Building the Clinical and Research
Agenda Together

1-3
2026 **OCTOBER**

Acibadem University
Maslak Hospital, Istanbul, Türkiye



HERDEM CARE

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DAY 1

01 OCTOBER 2026 THURSDAY:

SCIENTIFIC MEETING



10:00
10:30

Opening Ceremony

- Rare Disease in Türkiye: Ministry of Health Perspective
- Rare Disease Research at TÜSEB
- Rare Disease Research at TÜBİTAK
- Rare Disease Federation of Türkiye
- EURORDIS - Rare Disease Europe

National Anthem & Official Welcome Remarks from the Organizers

- Ministry of Health
- TÜSEB (Health Institutes of Türkiye)
- TÜBİTAK (The Scientific and Technological Research Council of Türkiye)
- Güneş Uyar
- Gülçin Gümüş, PhD



10:30
11:00

Why We Are Here: Building a Global TBL1XR1 Community

- HERDEM CARE
- Fly Little Bird Foundation
- The Importance of Establishing a Registry

Moderator: Başak Adaklı Aksoy, MD

- Ayşegül Altınbaş, PhD / Nusret Altınbaş
- Sarah Hargadon
- Feyza Nur Tuncer Kılınc, PhD



11:00 - 11:30 COFFEE / TEA BREAK

11:30
12:30

Research Perspectives on TBL1XR1

- TBL1XR1: Current Directions in Genetic Research
- TBL1XR1: Building the Clinical Research Agenda for TBL1XR1

Moderator: Hilal Yazıcı Malkoçoğlu, PhD

- Florian Eichler, MD
- Yasemin Alanay, MD, PhD



12:30 - 14:00 LUNCH BREAK



12:30 - 14:00 BIOLOGICAL SAMPLE COLLECTION



14:00
15:30

What Have We Learned So Far

- TBL1XR1-Clinical Spectrum, Genotype-Phenotype
- Epilepsy in TBL1XR1-Related NDD: Phenotype, EEG and Management
- Epilepsy in TBL1XR1-Related NDD: Intractable Epilepsy Treatment
- The Communicative Spectrum in TBL1XR1 disorders: From Hearing Loss to Augmentive Communication
- Q & A Session

Moderator: Semra Ayşe Hız Kurul, MD, PhD

- Özlem Akgün Doğan, MD, PhD
- Amanda Nagy, MD
- Pınar Gençpınar, MD
- Anna - Katharina Rohlf, MD
- Panel



15.30 - 16:00 COFFEE / TEA BREAK



16:00
17:30

Emerging Molecular and Functional Research

- Ongoing Molecular Research in TBL1XR1-Related Disorders
- Establishing a Brain Organoid Research Platform for TBL1XR1
- Clinical and molecular insights into the Italian cohort of Pierpont Syndrome
- Functional Rescue of TBL1XR1 haploinsufficiency: Bridging Basic science and translational Gene Therapy
- CRISPR - Cas and Genome Editing Technologies
- Q & A Session

Uğur Özbek, MD, PhD

- Florian Eichler, MD
- Yuk Yin Peter Ng, PhD / Neval Sevinç Özdemir, PhD / Kaya Bilgüvar, MD, PhD
- Donatella Milani, MD / Elisabetta Di Fede, PhD
- Yong Cheng, PhD
- Ahsen Özcan, PhD
- Panel



17:30 - 18:00 HOTEL TRANSFERS



FROM 18:00 FREE EVENING

DAY 2

02 OCTOBER 2026 FRIDAY: FAMILY FORUM



09:00 - 10:00 BIOLOGICAL SAMPLE COLLECTION

10:00
10:30

Everyday Life with *TBL1XR1*: Diagnosis, Daily Life, and Shared Experience

Moderated family panel with perspectives from Türkiye and the international *TBL1XR1* community

Moderator:
Yasemin Alanay, MD, PhD

Ayşegül Altınbaş, PhD / Sarah Hargadon /
Çağla Boral / Bradley Edelman, PhD



10:30 - 11:00 COFFEE / TEA BREAK

11:00
11:30

Understanding *TBL1XR1*: What Families Need to Know

- Care and Follow-up in *TBL1XR1*-Related Neurodevelopmental Disorder & Seizures, Medical Care, Emergency Preparedness
- *TBL1XR1* and Genetic Therapies for Rare Neurodevelopmental Diseases

Moderator:
Ercan Mihçı, MD

- Türkan Uygur Şahin, MD
- Florian Eichler, MD



12:00 - 13:30 LUNCH BREAK



12:00 - 13:30 BIOLOGICAL SAMPLE COLLECTION

Roundtable Discussions with Families and Experts

Moderator: Tunç Fışgın, MD &
Türkan Uygur Şahin, MD

Opening Remarks

The Psychosocial Impact of Rare Neurodevelopmental Conditions on Children and Their Families

Yankı Yazgan, MD

Panel Discussion Q&A

Daily Living, Sensory Needs & Independence —
Sensory Integration Therapist

Çağla Boral

Communication, Language, Feeding & AAC —
Speech & Language Therapist

Christina LaBorde-Altuğ M.S. CCC/SLP

Physical Participation & Mobility, Exercise -
Adapted Physical Activity Specialist & Physiotherapist

Duran Arslan, PhD / Çağla Boral

Behaviour, Sleep, Emotional Regulation & Family
Wellbeing — Psychiatrist / Psychologist

Yankı Yazgan, MD / Süleyman Çakıroğlu,
MD / Onur Tuğçe Poyraz Fındık, MD

Education, School & Social Inclusion —
Special Educator

Atilla Cavkaytar, PhD / Ebru Şen



15:30 - 16:00 COFFEE / TEA BREAK

16:00
17:00

Research Roadmap & Priorities for the Next Phase

Interactive brainstorming with experts & families

Moderator: Giovanna D. Ielacqua / Yasemin Memişoğlu

17:00
18:00

Closing Session: Summary and Next Steps

FINAL DECLARATION



18:00 - 18:30 HOTEL TRANSFERS

18:00
19:00

Working Group: Natural History, Longitudinal Follow-up &
Shared Data Collection

Herdem / FLBF / TÜSEB / TÜBİTAK / ACURARE /
MGH / Ped. Neurology Society / Ped. Genetics Society

18:00
19:00

Working Group: Research Infrastructure:
Registries, Biosamples & International Collaboration

Herdem / FLBF / TÜSEB / TÜBİTAK / ACURARE /
MGH / Ped. Neurology Society / Ped. Genetics Society



19:00 DINNER



DAY 3

03 OCTOBER 2026 SATURDAY

BOSPHORUS TOUR

(OR ALTERNATIVES DUE TO WEATHER)



DAY 4

04 OCTOBER 2026 SUNDAY

INDIVIDUAL DEPARTURES



HERDEM CARE