

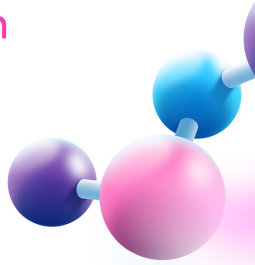
THE 2ND *TBL1XR1* GLOBAL FAMILY FORUM & THE 1ST INTERNATIONAL SCIENTIFIC CONFERENCE ON *TBL1XR1*



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

1-3
2026 **OCTOBER**

Acıbadem University
Maslak Hospital, İstanbul, 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
- Rare Disease Research at TÜSEB / TÜBİTAK
- Rare Disease Federation
- EURORDIS

National anthem, official welcome remarks from the organizers, Dr. Ayşegül Altınbaş

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



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: Dr. Başak Adaklı Aksoy

- Dr. Ayşegül Altınbaş & Nusret Altınbaş
- Sarah Hargadon
- Dr. Feyza Tuncer



10:30 - 11:00 COFFEE / TEA BREAK

11:30
12:00

Research Perspectives on TBL1XR1

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

Moderator: Dr. Tunç Fişgin

- Dr. Florian Eichler
- Dr. Yasemin Alanay



12:00 - 14:00 LUNCH BREAK



12:00 - 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 Augmentative Communication
- Q & A Session

Moderator: Dr. Semra Hız

- Dr. Özlem Akgün Doğan
- Dr. Amanda Nagy
- Dr. Pınar Gençpınar
- Dr. Anna - Katharina Rohlf
- Panel



15:30 - 16:00 COFFEE / TEA BREAK



16:30
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

Moderator: Dr. Uğur Özбек

- Dr. Florian Eichler
- Dr. Peter Ng / Dr. Neval Özdemir / Dr. Kaya Bilguvar
- Dr. Donatella Milani / Dr. Elisabetta Di Fede
- Dr. Cheng / Dr. Du



17:30 - 18:00 HOTEL TRANSFERS



18:00 - 19:00 FREE TIME



19:00 DINNER

DAY 2

02 OCTOBER 2026 FRIDAY: FAMILY FORUM

 09:00 - 10:00 BIOLOGICAL SAMPLE COLLECTION

10:00
10:30

**Everyday Life with TBLIXR1:
Diagnosis, Daily Life, and Shared Experience**
Moderated family panel with perspectives from
Türkiye and the international TBLIXR1 community

Moderator:
Dr. Yasemin Alanay
Dr. Ayşegül Altınbaş & Sarah Hargadon
& Çağla Boral & Dr. Bradley Edelman

 10:30 - 11:00 COFFEE / TEA BREAK

11:00
11:30

**Understanding TBLIXR1:
What Families Need to Know**
• Care and Follow-up in TBLIXR1-Related Neurodevelopmental Disorder
& Seizures, Medical Care, Emergency Preparedness
• TBLIXR1 and Genetic Therapies for Rare Neurodevelopmental Diseases

Moderator:
Dr. Kürşat Bora Çarman
• Dr. Türkan Uygur Şahin
• Dr. Florian Eichler

 12:00 - 13:30 LUNCH BREAK

Roundtable Discussions with Families and Experts

Opening Remarks: The Psychosocial Impact of Rare Neurodevelopmental Conditions on Children and Their Families, Dr. Yankı Yazgan
Topics for Discussion:
• Daily Living, Sensory Needs & Independence — Sensory Integration Therapist, Çağla Boral
• Communication, Language, Feeding & AAC — Speech & Language Therapist, Christina Laborde
• Mobility, Exercise & Physical Participation — Physiotherapist & Adapted Physical Activity Specialist, Duran Arslan & Çağla Boral
• Behaviour, Sleep, Emotional Regulation & Family Wellbeing — Psychologist / Psychiatrist, Dr. Yankı Yazgan & Dr. Süleyman Çakıroğlu & Dr. Onur Tuğçe Poyraz Fındık
• Education, School & Social Inclusion — Special Educator, Dr. Atilla Cavkaytar & Ebru Şen

Moderator:
**Dr. Tunç Fişgin &
Dr. Türkan Uygur Şahin**

- Dr. Yankı Yazgan,
- Dr. Süleyman Çakıroğlu,
- Dr. Onur Tuğçe Poyraz Fındık,
- Christina Laborde,
- Ebru Şen, Dr. Duran Arslan,
- Çağla Boral,
- Dr. Atilla Cavkaytar

 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 Diletta Ielacqua & Yasemin Memişoğlu

17:00
17:30

Closing Session: Summary and Next Steps

FINAL DECLARATION

 17:30 - 18:00 HOTEL TRANSFERS

17:30
18:00

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

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

17:30
18:00

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

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

 19:00 DINNER



DAY 3

03 OCTOBER 2026 SATURDAY

BOSPHORUS TOUR

(OR ALTERNATIVES DUE TO WEATHER)



DAY 4

04 OCTOBER 2026 SUNDAY

TRANSFERS TO AIRPORT



HERDEM CARE