



**UNIVERSITY OF HEALTH SCIENCES TURKEY  
IZMIR FACULTY OF MEDICINE  
SCIENTIFIC MEETINGS – 32**



**INTERNATIONAL MULTIDISCIPLINARY RARE DISEASES  
SYMPOSIUM**



**ORGANIZING COMMITTEE**

**Prof. Dr. Enver İLHAN**

**Prof. Dr. Haluk MERGEN**

**Prof. Dr. Gülden GÖKÇAY**

**Assoc. Prof. Dr. Berna ERDOĞMUŞ MERGEN**

**SCIENTIFIC COMMITTEE**

**Prof. Dr. Agata POLIZZI, Catania, Italy**

**Prof. Dr. Martino RUGGERI, Catania, Italy**

**Prof. Dr. Viyaj TIWARI, United Kingdom**

**Prof. Dr. Huma ARSHAD CHEEMA, Pakistan**

**Prof. Dr. Gülden GÖKÇAY, Istanbul, Turkey**

**Prof. Dr. Fatmahan ATALAR, Istanbul, Turkey**

**Prof. Dr. Harun AKAR, Izmir, Turkey**

**Prof. Dr. Haluk MERGEN, Izmir, Turkey**

**Doç. Dr. Pelin TEKE KISA, Izmir, Turkey**

**Dr. Amalia EGGLE GENTILE, Rome, Italy**

**Presenters:**

**Assist. Dr. Evin Aydın Akyurt**

**Assist. Dr. Sırma YALAZ**

**Download Microsoft Teams**

**MAY 31<sup>st</sup>, 2023  
WEDNESDAY  
( ONLINE)**

**Participation free**

**Meeting ID:370 901 857 876**

**Passcode:n3KWD2**

[https://teams.microsoft.com/l/meetup-join/19%3a\\_wHoy9Sic2KtmWVC7asYj6nkgLITS7wpK-6SniHJiNM1%40thread.tacv2/1684743851250?context=%7b%22Tid%22%3a%226bbc2380-82df-4644-8338-4fa7a38cf205%22%2c%22Oid%22%3a%227f802ff2-6a98-4b0a-9f3a-f51491cfb87f%22%7d](https://teams.microsoft.com/l/meetup-join/19%3a_wHoy9Sic2KtmWVC7asYj6nkgLITS7wpK-6SniHJiNM1%40thread.tacv2/1684743851250?context=%7b%22Tid%22%3a%226bbc2380-82df-4644-8338-4fa7a38cf205%22%2c%22Oid%22%3a%227f802ff2-6a98-4b0a-9f3a-f51491cfb87f%22%7d)

# ***INTERNATIONAL MULTIDISCIPLINARY RARE DISEASES SYMPOSIUM***

**09.00 – 09.30**

## **NATIONAL ANTHEM & MOMENT OF SILENCE OPENING CEREMONY SPEECHES**

**Prof. Dr. Cevdet ERDÖL**, Rector of University of Health Sciences Turkey  
**Prof. Dr. Enver İLHAN**, University of Health Sciences Turkey , Dean of Izmir Faculty of Medicine  
**Prof. Dr. Haluk MERGEN**, University of Health Sciences Turkey, Izmir Faculty of Medicine

**09.30 – 09.50**

**Chair: Prof. Dr. Haluk MERGEN**, University of Health Sciences Turkey, Izmir Faculty of Medicine,

**Ataxia Teleangiectasia: based-registry data from the Italian Centres of Expertise**  
**Prof. Dr. Agata POLIZZI**, University of Catania (Catania, Italy)

**09.50 – 10.00**

**Discussion**

**10.00-10.20**

**Chairs: Prof. Dr. Agata POLIZZI**, University of Catania (Catania, Italy)  
**Prof. Dr. Süleyman GÖRPELİOĞLU**, Ankara Dişkapı Yıldırım Beyazıt Training and Research Hospital

**The impact of real world evidence on rare disease ecosystem: Applications, limitations and future perspectives**  
**Prof. Dr. Gülden GÖKÇAY**, Istanbul University, Faculty of Medicine

**10.20-10.30**

**Discussion**

**Chairs: Prof. Dr. Gülden GÖKÇAY**, Istanbul University , Faculty of Medicine,  
**Prof. Dr. Harun AKAR**, University of Health Sciences Turkey, Izmir Faculty of Medicine,

**10.30-10.50**

**Advances in Neurocutaneous disorders**  
**Prof. Dr. Martino RUGGERI**, University of Catania (Catania, Italy)

**10.50 – 11.00**

**Discussion**

**11.00-11.20**

**Chairs: Prof. Dr. Cengiz CEYLAN**, University of Health Sciences Turkey, Izmir Faculty of Medicine,  
**Prof. Dr. Hülya PARILDAR**, University of Health Sciences Turkey, Izmir Faculty of Medicine,

**Modern genomics techniques to study diseases**  
**Prof. Dr. Vijay TIWARI**, Queen's University (Belfast,United Kingdom)

**11.20 – 11.30**

**Discussion**

**11.30 – 11.50**

**Chairs: Prof. Dr. Adem ÖZKARA**, University of Health Sciences Turkey, Gülhane Faculty of Medicine,  
**Assoc. Prof. Duygu Ayhan BAŞER**, Hacettepe University, Faculty of Medicine,

**Establishing a Rare Disease center in a developing country with high burden of genetic disorders**  
**Prof. Dr. Huma ARSHAD CHEEMA**, University of Child Health Health Lahore Pakistan

**11.50 – 12.00**

**Discussion**

**12.00 – 12.30**

**LUNCH TIME**

<b>12.30-12.50</b>	<p><b>Chairs: Prof. Dr. Nilgün ÖZÇAKAR</b>, 9 Eylül University Faculty of Medicine  <b>Assoc. Prof. Dr. Umut GÖK BALCI</b>, University of Health Sciences Turkey, Izmir Faculty of Medicine  <b>Analytic systems in rare disease ecosystem: Metabolomics, genetic diagnostics and future perspectives</b>  <b>Prof. Dr. Fatmahan ATALAR</b>, Istanbul University, Institute of Child Health</p>
<b>12.50-13.00</b>	<p><b>Discussion</b></p>
<b>13.00 – 13.20</b>	<p><b>Chairs: Assoc. Prof. Dr. Yasemin KILIÇ ÖZTÜRK</b>, University of Health Sciences Turkey, Izmir Faculty of Medicine  <b>Assoc. Prof. Dr. Nil TEKİN</b>, University of Health Sciences Turkey, Izmir Faculty of Medicine</p> <p><b>Health humanities in practice: the project PandemicaMente at a glance</b>  <b>Dr. Amalia EGLE GENTILE</b>, Istituto Superiore di Sanità (Rome, Italy)</p>
<b>13.20 – 13.30</b>	<p><b>Discussion</b></p>
<b>13.30– 13.50</b>	<p><b>Chairs: Assoc. Prof. Dr. Berna ERDOĞMUŞ MERGEN</b>, Atatürk Research&amp;Training Hosp.  <b>Assoc. Prof. Dr. Hilal ÖZKAYA</b>, University of Health Sciences Turkey , Hamidiye Faculty of Medicine</p> <p><b>An example of rare disease: Alcaptonuria</b>  <b>Assoc. Prof. Dr. Pelin TEKE KISA</b>, University of Health Sciences Turkey, Dr Behçet Uz Child Diseases and Surgery Research and Training Hospital</p>
<b>13.50 – 14.00</b>	<p><b>Discussion</b></p>
<b>14.00 – 14.20</b>	<p><b>Chairs: Assoc. Prof. Dr. Erol YAYLA</b>, University of Health Sciences Turkey , Adana Faculty of Medicine  <b>Assoc. Prof. Dr. Tarık EREN</b>, University of Health Sciences Turkey , Gülhane Faculty of Medicine</p> <p><b>Compulsory Neonatal screening tests at Family Medicine Centers in Türkiye</b>  <b>Prof. Dr. Haluk MERGEN</b>, University of Health Sciences Turkey , Izmir Faculty of Medicine</p>
<b>14.20 – 14.30</b>	<p><b>Discussion</b></p>
<b>14.30 – 14.50</b>	<p><b>Chairs: Assoc. Prof. Dr. Yusuf Adnan GÜÇLÜ</b>, University of Health Sciences Turkey, Izmir Faculty of Medicine  <b>Assoc. Prof. Dr. Nurdan TEKGÜL</b>, University of Health Sciences Turkey , Izmir Tepecik Research &amp; Training Hospital</p> <p><b>A study across international strategies and action plans for disabilities Beliefs, expectations and attitude toward the use of digital tools and videogames to support child development in healthcare and education</b>  <b>Dr. Eleonora DESOGUS, Dr. Gennaro ANASTASIO, Dr. Lucrezia MICHELI, Dr. Amalia Egle GENTILE</b>,  Istituto Superiore di Sanità (Rome, Italy)</p>
<b>14.50 – 15.00</b>	<p><b>Discussion</b></p>
<b>15.00-15.20</b>	<p><b>Chairs: Assoc. Prof. Dr. Ashı BAYINDIR</b>, University of Health Sciences Turkey , Izmir Bozyaka Research &amp; Training Hospital  <b>Dr. Işık GÖNENÇ</b>, Haydarpaşa Numune Research&amp;Training Hospital</p> <p><b>Beliefs, expectations and attitude toward the use of digital tools and videogames to support child development in healthcare and education</b>  <b>Dr. Sergio RINELLA*</b>, <b>Dr. Tornike KHOSHTARIA**</b>, <b>Prof. Dr. Agata POLIZZI**</b>  University of Catania (Catania, Italy)*, Geomedi University (Tbilisi, Georgia) **</p>
<b>15.20– 15.30</b>	<p><b>Discussion</b></p>
<b>15.30– 15.50</b>	<p><b>Chairs: Dr. Özge TUNCER</b>, University of Health Sciences Turkey , Izmir Bozyaka Research &amp; Training Hospital  <b>Dr. Demet MERDER</b>, University of Health Sciences Turkey , Izmir Tepecik Research &amp; Training Hospital</p> <p><b>McCune-Albright syndrome (MAS): the tale of two genes</b>  <b>Dr. Manuela LO BIANCO, Dr. Roberta LEONARDI</b>,  University of Catania (Catania, Italy)</p>
<b>15.50– 16.00</b>	<p><b>Discussion</b></p>
<b>16.00– 16.20</b>	<p><b>Chairs: Dr. Olgu AYGÜN</b>, University of Health Sciences Turkey, Izmir Bozyaka Research &amp; Training Hospital  <b>Asist. Dr. Evin Aydın Akyurt</b>, University of Health Sciences Turkey, Izmir Tepecik Research &amp; Training Hospital</p> <p><b>Extensive Newborn screening: the experience of the Eastern Sicily</b>  <b>Dr. Marianna MESSINA, Concetta MELI</b>, University of Catania (Catania, Italy)</p>
<b>16.20-16.30</b>	<p><b>Discussion</b></p>
<b>16.30-16.50</b>	<p><b>Chairs: Dr. Hande İLERİ</b>, University of Health Sciences Turkey , Izmir Tepecik Research &amp; Training Hospital  <b>Asist. Dr. Sırma YALAZ</b>, University of Health Sciences Turkey, Izmir Tepecik Research &amp; Training Hospital</p> <p><b>Novel neurometabolic therapies in the pediatric setting</b>  <b>Dr. Annamaria SAPUPPO, Dr. Luisa LA SPINA</b>, University of Catania (Catania, Italy)</p>
<b>16.50-17.00</b>	<p><b>Discussion</b></p>
<b>17.00-17.10</b>	<p><b>Closing Ceremony</b></p>