

SCIENTIFIC – PRACTICAL CONFERENCE  
**DYSMORPHOLOGY SUMMER**  
**2024**



MOKSLINĖ – PRAKTINĖ KONFERENCIJA  
**DISMORFOLOGIJOS VASARA**  
**2024**

15-17<sup>th</sup> August 2024  
 Hotel Gabija,  
 Vytauto g. 40, Palanga, Lithuania

Thursday – 15<sup>th</sup> August

<i>10:30 – 11:00 REGISTRATION</i>	
<b>11:00 – 12:30 FIRST SESSION</b>	
<i>Unknown cases (live consultations)</i>	
<i>11:00 – 11:30</i>	Patient 1 (live consultation)
<i>11:30 – 12:00</i>	Patient 2 (live consultation)
<i>12:00 – 12:30</i>	Patient 3 (live consultation)
<i>12:30 – 13:50 LUNCH</i>	
<b>13:50 – 16:00 SECOND SESSION Chair Eglė Preikšaitienė</b>	
<i>13:50 – 14:00</i>	Aušra Matulevičienė <b>OPENING ADDRESS</b> Vilnius, Lithuania
<i>14:00 – 15:00</i>	Emilia K Bijlsma <b>FOLLOW-UP STUDY AFTER REACHING A GENETIC DIAGNOSIS IN 82 ELDERLY PATIENTS</b> Leiden, The Netherlands
<i>15:00 – 15:30</i>	Aleš Maver <b>HOW NGS TRANSFORMED DIAGNOSTICS AND OUTCOMES OF RARE DISEASE PATIENTS IN SLOVENIA – LESSONS LEARNED THE PAST 10 YEARS</b> Ljubljana, Slovenia
<i>15:30 – 16:00</i>	Sander Pajusalu <b>BALTIC DISEASE HERITAGE!?</b> Tartu, Estonia
<i>16:00 – 16:30 COFFEE BREAK</i>	
<b>16:30 – 18:30 THIRD SESSION Chair Sander Pajusalu</b>	
<i>16:30 – 17:00</i>	Piotr Iwanowski <b>FROM GESTALT TO DATA: ALIGNING DIVERSE OBSERVATIONS IN DYSMORPHOLOGY</b> Warsaw, Poland
<i>17:00 – 17:30</i>	Algirdas Utkus <b>IS IT IMPORTANT FOR DENTISTS TO KNOW THE GENETIC BASIS OF ORAL HEALTH?</b> Vilnius, Lithuania
<i>17:30 – 17:50</i>	Renata Posmyk <b>CHANGING PHENOTYPE IN PATIENTS WITH OVERGROWTH SYNDROME</b> Białystok, Poland
<i>17:50 – 18:10</i>	Evelina Marija Vaitėnienė <b>A CASE OF A COMPLEX CHROMOSOMAL ANOMALY CAUSING DYSMORPHIC FEATURES AND NEURODEVELOPMENTAL DISORDER</b> Vilnius, Lithuania
<i>18:10 – 18:30</i>	<b>DISCUSSION</b>

Friday – 16<sup>th</sup> August

<b>9:00 – 11:00 FOURTH SESSION</b> <i>Chair: Emilia K Bijlsma</i>	
9:00 – 10:00	Jeroen Breckpot <b>MECHANISMS UNDERLYING PHENOTYPIC VARIABILITY IN 22q11.2 DELETION SYNDROME AND GUIDELINES FOR FOLLOW-UP.</b> Leuven, Belgium
10:00 – 10:20	Eglė Narmontienė <b>22q11.2 DELETION AND DUPLICATION SYNDROMES: A REVIEW OF THE LITERATURE AND CLINICAL CASES</b> Vilnius, Lithuania
10:20 – 10:40	Rasa Traberg <b>REVIEW OF 22Q11.2 DELETION SYNDROME PATIENTS DIAGNOSED IN LUHS HOSPITAL KAUNO KLINIKOS DURING 10 YEARS PERIOD.</b> Kaunas, Lithuania
10:40 – 11:00	Laura Mauring <b>THE DIAGNOSTIC CHALLENGE OF LIKELY VAN DEN ENDE-GUPTA SYNDROME</b> Tartu, Estonia
<i>11:00 – 11:30 COFFEE BREAK</i>	
<b>11:30 – 13:00 FIFTH SESSION</b> <i>Chair Rasa Ugenskienė</i>	
11:30 – 12:00	Aleš Maver <b>IMPROVING THE DIAGNOSTIC EVALUATION IN SYNDROMIC PATIENTS BY BRIDGING THE CLINICS AND THE NGS LABORATORY</b> Ljubljana, Slovenia
12:00 – 12:20	Monika Gos <b>TARGETED SEQUENCING IN RASOPATHY PATIENTS</b> Warsaw, Poland
12:20 – 12:40	Aušra Morkūnienė <b>SYT1 - ASSOCIATED NEURODEVELOPMENT DISORDER: LONG JOURNEY TO DIAGNOSIS</b> Vilnius, Lithuania
12:40 – 13:00	Anna Kutkowska-Kaźmierczak <b>RAHMAN SYNDROME - CLINICAL AND MOLECULAR ANALYSIS OF 5 PATIENTS</b> Warsaw, Poland
<i>13:00 – 14:30 LUNCH</i>	
<b>14:30 – 16:30 SIXTH SESSION</b> <i>Chair Piotr Iwanowski</i>	
14:30 – 15:30	Emilia K Bijlsma <b>EXAMPLES OF DIAGNOSES WITH THERAPEUTIC CONSEQUENCES</b> Leiden, The Netherlands
15:30 – 15:50	Rasa Ugenskienė <b>FAMILIAL CASE OF INTELLECTUAL DISABILITY-FACIAL DYSMORPHISM SYNDROME CAUSED SETD5 GENE PATHOGENIC VARIANT.</b> Kaunas, Lithuania
15:50 – 16:10	Violeta Mikštienė <b>PHIP-ASSOCIATED CHUNG-JANSEN SYNDROME: CASE REPORT</b> Vilnius, Lithuania
16:10 – 16:30	Deimantė Braždžiūnaitė <b>NEPHRONOPHTHISIS AND RELATED SYNDROMES: CLINICAL AND GENETIC OVERVIEW</b> Vilnius, Lithuania
<i>16:30 – 17:00 COFFEE BREAK</i>	
<b>17:00 – 18:30 SEVENTH SESSION</b> <i>Chair Jeroen Breckpot</i>	
17:00 – 17:30	Aleksander Jamsheer <b>CRANIOSYNOSTOSIS – AN OVERVIEW OF FREQUENT AND RARE GENETIC SYNDROMES</b> Poznan, Poland
17:30 – 17:50	Kaisa Teele Oja <b>NEURODEVELOPMENTAL SYNDROME DUE TO BI-ALLELIC LOSS-OF-FUNCTION VARIANTS IN WBP4</b> Tartu, Estonia
17:50 – 18:10	Renata Posmyk <b>CASE REPORT OF CORNELIA DE LANGE SYNDROME</b> Białystok, Poland
18:10 – 18:30	<b>DISCUSSION</b>

Saturday – 17<sup>th</sup> August

<b>9:00 – 11:00 EIGHTH SESSION</b> <i>Chair Vytautas Štiužas</i>	
9:00 – 9:30	Ewelina Preizner-Rzucidło <b>DISORDERS OF SEX DEVELOPMENT – ANALYSIS OF A GROUP OF 40 INDIVIDUALS</b> Krakow, Poland
9:30 – 10:00	Ode Laik <b>TWO SISTERS WITH MAYER-ROKITANSKY-KÜSTER-HAUSER SYNDROME – SEMA3F GENE BIALLELIC VARIANTS AS A POSSIBLE CAUSE</b> Tartu, Estonia
10:00 – 10:20	Deimantė Braždžiūnaitė <b>EXPLORING HNF1B-RELATED PATHOLOGIES: CLINICAL AND GENETIC SPECTRUM AND CASE SERIES</b> Vilnius, Lithuania
10:20 – 10:40	Kai Muru <b>PATIENT WITH A DE NOVO LOSS-OF-FUNCTION VARIANT IN USP9X GENE</b> Tartu, Estonia
10:40 – 11:00	Michael Sabsovich <b>POTOCKI – LUPSKI SYNDROME: CASE REPORT</b> Vilnius, Lithuania
<i>11:00 – 11:30 COFFEE BREAK</i>	
<b>11:30 – 13:00 NINTH SESSION</b> <i>Chair Algirdas Utkus</i>	
11:30 – 11:50	Lina Kievišienė, Jovita Urbaitė-Bernotė <b>X-LINKED KABUKI SYNDROME: CASE REPORT</b> Kaunas, Lithuania
11:50 – 12:10	Madara Mašinska <b>CASE REPORT</b> Riga, Latvia
12:10 – 12:30	Rugilė Stankevičiūtė <b>PITT HOPKINS SYNDROME IN LITHUANIA</b> Vilnius, Lithuania
12:30 – 12:50	Renata Posmyk <b>COFFIN-SIRIS SYNDROME IN 4 PATIENTS – NATURAL HISTORY</b> Białystok, Poland
12:50 – 13:20	<b>FINAL DISCUSSION</b>



Lietuvos  
mokslo  
taryba



biovita