SCIENTIFIC – PRACTICAL CONFERENCE
DYSMORPHOLOGY SUMMER 2024

MOKSLINĖ – PRAKTINĖ KONFERENCIJA
DISMORFOLOGIJOS VASARA 2024

15-17th August 2024
Hotel Gabija,
Vytauto g. 40, Palanga, Lithuania

Thursday – 15th August

10:30 – 11:00 REGISTRATION	
11:00 – 12:30 FIRST SESSION	
Unknown cases (live consultations)	
11:00 – 11:30	Patient 1 (live consultation)
11:30 – 12:00	Patient 2 (live consultation)
12:00 – 12:30	Patient 3 (live consultation)
12:30 – 13:50 LUNCH	
13:50 – 15:50 SECOND SESSION	
13:50 – 14:00	Aušra Matulevičienė **OPENING ADDRESS** Vilnius, Lithuania
14:00 – 15:00	Emilia K Bijlsma **FOLLOW-UP STUDY AFTER REACHING A GENETIC DIAGNOSIS IN 82 ELDERLY PATIENTS** Leiden, The Netherlands
15:00 – 15:30	Aleš Maver **HOW NGS TRANSFORMED DIAGNOSTICS AND OUTCOMES OF RARE DISEASE PATIENTS IN SLOVENIA - LESSONS LEARNED THE PAST 10 YEARS** Ljubljana, Slovenia
15:30 – 15:50	Sander Pajusalu **BALTIC DISEASE HERITAGE!?** Tartu, Estonia
15:50 – 16:20 COFFEE BREAK	
16:20 – 18:20 THIRD SESSION	
16:20 – 16:50	Algirdas Utkus **IS IT IMPORTANT FOR DENTISTS TO KNOW THE GENETIC BASIS OF ORAL HEALTH?** Vilnius, Lithuania
16:50 – 17:10	Rasa Ugenskienė **FAMILIAL CASE OF INTELLECTUAL DISABILITY-FACIAL DYSMORPHISM SYNDROME CAUSED *SETD5* GENE PATHOGENIC VARIANT.** Kaunas, Lithuania
17:10 – 17:30	Ewelina Preizner-Rzucidło **DISORDERS OF SEX DEVELOPMENT – ANALYSIS OF A GROUP OF 40 INDIVIDUALS** Krakow, Poland
17:30 – 17:50	Violeta Mikštienė **PHIP-ASSOCIATED CHUNG-JANSEN SYNDROME, CASE REPORT** Vilnius, Lithuania
17:50 – 18:20	**DISCUSSION**

Friday – 16th August

9:00 – 11:00 FOURTH SESSION	
9:00 – 10:00	Jeroen Breckpot **MECHANISMS UNDERLYING PHENOTYPIC VARIABILITY IN 22q11.2 DELETION SYNDROME AND GUIDELINES FOR FOLLOW-UP.** Leuven, Belgium
10:00 – 10:20	Renata Posmyk **CHANGING PHENOTYPE IN PATIENTS WITH OVERGROWTH SYNDROME** Bialystok, Poland
10:20 – 10:40	Rasa Traberg **REVIEW OF 22Q11.2 DELETION SYNDROME PATIENTS DIAGNOSED IN LUHS HOSPITAL KAUNO KLINIKOS DURING 10 YEARS PERIOD.** Kaunas, Lithuania
10:40 – 11:00	Kaisa Teele Oja **NEURODEVELOPMENTAL SYNDROME DUE TO BI-ALLELIC LOSS-OF-FUNCTION VARIANTS IN WBP4** Tartu, Estonia
11:00 – 11:30 COFFEE BREAK	
11:30 – 13:00 FIFTH SESSION	
11:30 – 12:00	Aleš Maver **IMPROVING THE DIAGNOSTIC EVALUATION IN SYNDROMIC PATIENTS BY BRIDGING THE CLINICS AND THE NGS LABORATORY** Ljubljana, Slovenia
12:00 – 12:20	Evelina Vaitėnienė **A CASE OF A COMPLEX CHROMOSOMAL ANOMALY CAUSING DYSMORPHIC FEATURES AND NEURODEVELOPMENTAL DISORDER** Vilnius, Lithuania
12:20 – 12:40	Anna Kutkowska-Kaźmierczak **RAHMAN SYNDROME - CLINICAL AND MOLECULAR ANALYSIS OF 5 PATIENTS** Warsaw, Poland
12:40 – 13:00	Deimantė Braždžiūnaitė **NEPHRONOPHTHISIS AND RELATED SYNDROMES: CLINICAL AND GENETIC OVERVIEW** Vilnius, Lithuania
13:40 – 14:30 LUNCH	
14:30 – 16:30 SIXTH SESSION	
14:30 – 15:30	Emilia K Bijlsma **EXAMPLES OF DIAGNOSES WITH THERAPEUTIC CONSEQUENCES** Leiden, The Netherlands
15:30 – 15:50	Monika Gos **TARGETED SEQUENCING IN RASOPATHY PATIENTS** Warsaw, Poland
15:50 – 16:10	Eglė Narmontienė **22q11.2 DELETION AND DUPLICATION SYNDROMES: A REVIEW OF THE LITERATURE AND CLINICAL CASES** Vilnius, Lithuania
16:10 – 16:30	Laura Mauring **THE DIAGNOSTIC CHALLENGE OF LIKELY VAN DEN ENDE-GUPTA SYNDROME** Tartu, Estonia
16:30 – 17:00 COFFEE BREAK	
17:00 – 18:30 SEVENTH SESSION	
17:00 – 17:30	Aleksander Jamsheer **CRANIOSYNOSTOSIS – AN OVERVIEW OF FREQUENT AND RARE GENETIC SYNDROMES** Poznan, Poland
17:30 – 17:50	Austėja Semėnaitė **SYT1 - ASSOCIATED NEURODEVELOPMENT DISORDER: LONG JOURNEY TO DIAGNOSIS** Vilnius, Lithuania
17:50 – 18:10	Renata Posmyk **CASE REPORT OF CORNELIA DE LANGE SYNDROME** Bialystok, Poland
18:10 – 18:30	**DISCUSSION**

Saturday – 17th August

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