QENDRA SPITALORE UNIVERSITARE "NËNË TEREZA"

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UNIVERSITETI I MJEKËSISË TIRANË

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SHOQATA E SËMUNDJEVE TË RRALLA

The 3-rd Scientific Conference on Rare Diseases

"Towards clinical implementation of new generation genetic tests (NGS) in diagnosis of rare genetic diseases"

Rare Diseases Association and
Clinical Genetics Laboratory Service

19 February 2025
Marriott Hotel

8:30 – 9:00 REGISTRATION

9:00 – 9:30 WELCOME SPEECH

- Mrs. Albana KOÇIU / Minister of Health and Social Protection
- Prof. Xheladin DRAÇINI / Rector, University of Medicine, Tirana
- Prof. Alma CANI / General Director, University Hospital Center "Mother Teresa"
- Prof. Margarita GJATA / Dean, Faculty of Medicine, UMT
- Prof. Ermira KOLA / Head of the Pediatric Department

SESSION I

Chairperson: *Prof. Anila Babameto – Laku, Prof. Rob Pieters, Prof. Ermira Kola, Prof. Anila Godo, Prof. Donjeta Bali*

09:30 – 09:45 Do we have data on rare genetic diseases in Albania? Diagnostic challenges
Prof. Dr. Anila Babameto-Laku

09:45 – 10:00 Clinical implementation of sequencing studies in childhood cancer care; Results from the Princess Maxima Center *Prof. Rob Pieters*

10:00 – 10:15 Is reading DNA enough to make a clinical diagnosis? *Prof. Giuseppe Novelli*

10:15 – 10:30 Our experience in the molecular diagnosis of the frequent genetic diseases in Albania. Towards implementation of NGS in diagnosis of rare genetic diseases.
Msc. Besmira Basholli

10:30 – 10:45 Building bridges of hope: The role of NGOs in transforming pediatric cancer care
Mrs. Carmen Uscatum

10:45 – 11:00 Questions and Discussions

11:00 – 11:15 *Coffe Break*

SESSION II

Chairperson: *Prof. Gjeorgjina Kuli -Lito, Prof. Paskal Cullufi, Dr. Shk. Aida Bushati, Prof. Numila Kuneshka, Prof. Asc. Irena Kasmi*

11:30 – 11:45 Congenital immunodeficiencies, their diagnosis and classification
Prof. Gjeorgjina Kuli - Lito

11:45 – 12:00 Changing etiopathogenesis of neurological disorders in the new era of NGS
Dr. Shk. Aida Bushati

12:00 – 12:15 The Impact of metabolic and genetic screening in clinical practice: two case reports *Prof. Asc. Alketa Hoxha*

12:15 – 12:30 The importance of molecular diagnosis in pediatric endocrine diseases; diagnosis, treatment and progression *D.Shk. Laurant Kollcaku*

12:30 – 12:45 Fructose-1,6-bisphosphatase deficiency: Case report *Dr. Gladiola Hoxha*

12:45 – 13:00 Questions and Discussions

13:00 – 14:00 *Coctail/ Finger food*

13:00 – 13:40 *Symposium supported by* SwixxBiopharma

13:45 – 14:00 *Symposium supported by Roche:"Advancement in SMA; Key insights from FIREFISH study in pediatric Population"*

SESSION III

Chairperson: *Prof. Arben Ivanaj, Prof. Manika Kreka, Dr. Shk. Adela Perolla, Dr.Shk. Alma Cili*

14:00 – 14:15 Luspatercept: A therapeutic innovation for patients with Beta-Thalassemia Major *Prof. Manika Kreka*

14:15 – 14:30 Molecular cytogenetics (FISH) in diagnosis and treatment of hematological malignancies – our experience *Dr. Shk. Dorina Roko*

14:30 – 14:45 Systemic mastocytosis: Case report and literature review *Dr. Shk. Adela Perolla*

14:45 – 15:00 Adult Langerhans cell histiocytosis: Literature review through a case report *Dr. Shk. Alma Cili*

15:00 – 15:30 *Questions and Discussions*

15:00 – 15:30 *Symposium supported by Centogene: "Importance of genetic diagnosis in clinical practice, what is new in NGS"*

SESSION IV

Chairperson: *Prof. Agnieszka Madej-Pilarczyk, Prof. Margarita Gjata, Prof. Alma Idrizi, Prof. Asc. Ariana Strakosha*

15:30 – 15:45 Neurodevelopmental disorders: A constant challenge in genetics *Prof. Agnieszka Madej-Pilarczyk*

15:45 – 16:00 C 3 glomerulopathy *Prof. Margarita Gjata*

16:00 – 16:15 Classic hypertrophic osteoarthropathy primary autosomal dominant (PHOAD) and a heterozygous pathogenic variant in SLCO2A1 gene: Case report *Dr. Barbara Shkodrani*

16:15 – 16:30 Atypical hemolytic uremic syndrome *Prof. Alma Idrizi*

16:30 – 16:45 Rare cystinuria: Pathophysiology, diagnostic approaches, and current treatment strategies" *Prof. Asc. Ariana Strakosha*

16:45 – 17:00 Hereditary renal cystic disease *Prof. Diamant Shtiza*

17:00 – 17:30 *Questions and Discussions*