Baltic Metabolic Group Meeting

" The journey of a patient with mitochondrial disorder - from diagnosis to treatment"

21.-22.03 2024

Tallinn, Estonia. Original Sokos Hotel Viru, Viru väljak 4.

21.03.2024

10:45 - 11:10 Gathering

11:10 - 13:15 Leigh syndrome

11:10 - 11:15 Dr. J. Songailiene - opening remarks
11:15 - 12:15 Prof. S. Rahman - "Leigh syndrome"
12:15 - 12:35 Dr. A. Seménaité and Dr. A. Matulevičiene - "Leigh syndrome: case report and patient presentation"
12:35 - 12:55 Dr. H. Freijere-Pope - "Leigh syndrome: a case presentation"
12:55 - 13:15 Prof. K. Õunap - " Leigh syndrome caused by complex I deficiency: patient presentation"

13:15 - 14:15 Lunch at Merineitsi restaurant

14:15 - 16:15 Mitochondrial disorders

14:15 - 15:15 Dr. H. Prokisch - "Novel biomarkers and the development of specific signatures in the metabolomics, lipidomics and epigenetics data from mitochondrial disease patients"
15:15 - 16:15 Prof. S. Rahman - "(Gene) therapy for mitochondrial disorders"

16:15 - 16:45 Coffee break in lobby

16:45 - 17:45 Rare Disease Centers in the Baltic States

16:45 - 17:05 Ass. Prof. B. Tumiené - "Center for Rare Diseases in Lithuania"
17:05 - 17:25 Dr. I. Mičule - "Center for Rare Diseases in Latvia"
17:25 - 17:45 Dr. K. Tael - "The Competence Centre of Rare Diseases at Tartu University Hospital, Estonia"
17:45 - 18:00 Discussion

19:00 Dinner at Olde Hansa, Vana turg 1

22.03.2024

9:00 - 11:00 New developments in the diagnostics of herediatry metabolic diseases

9:00 - 10:00 Dr. H. Prokisch - "Integrative omics approaches to advance rare disease diagnostics"
10:00 - 10:30 Prof. K. Õunap - "The discovery of new metabolic diseases"
10:30 - 10:45 Dr. K. T. Oja - "Transcriptomic and metabolomic approach in rare diseases discovery"

10:45 - 11:15 Coffee break in lobby

11:15 - 13:15 Complex cases

11:15 - 11:35 Ass. Prof. B. Tumiené - "Multidisciplinary care of patients with inherited metabolic diseases and epilepsy: current perspectives"
11:35 - 11:55 Dr. L. Skrabule - "MT-ND5: a case presentation"
11:55 - 12:15 Dr. M. Masinska - "Child born with 16p12.2 del syndrome and maternal PKU: a case presentation"
12:15 - 12:35 Dr. A. Seménaité - "Lysinuric protein intolerance and pregnancy: case report"
12:35 - 12:55 Dr. S. Laktina - "Branched chain ketoacid dehydrogenase kinase deficiency"
12:55 - 13:15 Dr. L. Vasilevska - "PEPCK deficiency: a case presentation"

13:15 - 14:15 Lunch at Merineitsi restaurant

14:15 - 15:00 Complex cases

14:15 - 14:35 Dr. K. Muru - " An unusual profile in oligosaccharides analysis"
14:35 - 14:55 Dr. L. Roht - "A 5-year-old girl with suspected Pearson syndrome: an unsolved case"
14:55 - 15:00 Closing remarks

15:30 - 17:00 Tour with guide in the Tallinn Old Town