BALTIC METABOLIC GROUP MEETING
MITOCHONDRIAL DISORDERS: CURRENT APPROACH IN DIAGNOSIS & TREATMENT
29th -30th of March, 2017
Vilnius

PROGRAM

***Meeting venue**: Conference hall (red auditorium), Vilnius University Hospital Santariskiu Clinics*
Santariskiu str.2, Vilnius, Lithuania

29th of March (Wednesday)

8.30 – 9.00	***Registration***
9.00 – 9.05	Welcome addresses
9.05-10.00	*Johannes Mayer* (Salzburg, Austria) **Overview – Mitochondria in human pathology**
10.00 – 11.00	*Saskia Wortmann* (Salzburg, Austria) **Clinics of inherited mitochondrial disorders**
11.00 – 11.15	*Laura Roht*, K. Õunap (Tartu, Estonia) **43-year-old male with MIDD syndrome caused by heteroplasmic m.15161T>C mutation in MT-CYB gene**
11.15 – 11.30	***Coffee break***
11.30 – 12.00	*Birutė Tumienė* (Vilnius, Lithuania) **Mimics of mitochondrial diseases**
12.00 – 12.20	*Sanna Puusepp* (Tartu, Estonia) R. Kovacs-Nagy , B. Alhaddad, M. Braunisch, G. F. Hoffmann, U. Kotzaeridou , L. Lichvarova, M. Liiv, C. Makowski , M. Mandel, T. Meitinger, S. Pajusalu, R. J. Rodenburg , D. Safiulina, T. M. Strom, I. Talvik, A. Vaarmann, C. Wilson, A. Kaasik, T. B. Haack , K. Õunap **Compound heterozygous SPATA5 mutations: phenotype overview and functional studies**
12.20 – 12.40	*Marija Rachlevičiūtė* (Vilnius, Lithuania) **Prenatal diagnostics of Barth syndrome. Case report.**
12.40 – 13.00	*Rūta Marcinkutė,* Aušra Matulevičienė (Vilnius, Lithuania). **Solved case: Atypical form of ataxia –telangiectasia**
13.00 – 14.00	***Lunch***
14.00 – 15.00	*Holger Prokisch* (München , Germany) **Diagnostics of mitochondrial disorders - classical and new approach**
15.00 - 15.15	*Inna Inashkina* (Riga, Latvia) **Using of cytoplasmic hybrid cell technique in characterization of mtDNA mutations.**
15.15 – 15.30	*Donatas Petroška*, Deimantė Brazdžiūnaitė (Vilnius, Lithuania) **Review of mitochondriopathy cases in skeletal muscle biopsies**
15.30 – 15.45	***Coffee break***
15.45 – 16.00	*Dita Pelnena* (Riga, Latvia) **OXPHOS enzyme activity measurements in peripheral blood leucocytes**
16.00 – 16.15	*Sander Pajusalu*, K. Reinson, S. Puusepp, Ü. Murumets, T. Reimand, R. Rodenburg (The Netherlands), K. Õunap (Tartu, Estonia) **The effectiveness of whole exome sequencing in previously unsolved patients with a suspected mitochondrial disorder**
16.15 – 17.30	Unknown cases: presentations form each country

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30th of March (Thursday)

9.00 – 9.15	*Katrin Õunap,* S. Pajusalu, K. Künnapas, H, Lilleväli (Tartu, Estonia) **Newborn with acute encephalopathy and 3-methylglutaconic aciduria**
9.15– 10.15	*Elena Pischik* (St. Petersburg, Russia) **Acute porphyria, hard to get?**
10.15 – 11.00	*Saskia Wortmann , Johannes Mayer* (Salzburg, Austria) **Hepatopathy and mitochondrial energy metabolism**
11.00 – 11.30	***Coffee break***
11.30 – 12.00	*Holger Prokisch* (München , Germany) **The Acute Liver Failure (ALF) project**
12.00 - 12.15	*Karit Reinson,* K. Künnapas, A. Kriisa, M.-A. Vals, K. Muru, K. Õunap. (Tartu, Estonia) **Estonian experience of expanded newborn screening.**
12.15 – 12.30	*Agnė Ulytė* (Vilnius, Lithuania) , A. Matulevičienė, L. Kupčinskas, L. Dvorakova, M. Hnizdova Bouckova, H. Klimentova, H. Vlaskova, R. Kaladytė-Lokominienė, V. Kozich, H.Y. Wu, K. Brammeier, H. Church **Niemann-Pick Disease ,Type C. Case report**
12.30 – 12.45	*Zita Krumina* (Riga, Latvia) **The stem cell transplantation in patient with alpha mannosidosis. Case report.**
12. 45 – 13.00	*Deimantė Brazdžiūnaitė*, Birutė Burnytė (Vilnius, Lithuania). **Late onset Pompe disease. Case report**
13.00 – 14.00	Unknown cases: presentations form each country
14.00	Closing remarks, discussions, future plans

Organizers

Lithuanian Society of Human and Medical genetics
Baltic Metabolic Group
Centre for Medical Genetics, Vilnius University Hospital Santariskiu Clinics
Dep. of Human and Medical Genetics, Medical Faculty, Vilnius University

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