Preliminary program

BALTIC METABOLIC GROUP MEETING: INHERITED GLYCOGEN STORAGE DISORDERS AND DISORDERS OF PURINES AND PYRIMIDINES.

27th -28th of March, 2014

Vilnius

Program

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

27th of March (Thursday)

10.30 – 12.00	***Registration and coffee***
12.00 – 12.05	Welcome addresses: prof. Algirdas Utkus
12.05 – 13.05	Rene Santer (Hamburg): Glycogen storage disorders
13.05 – 13.30	K. Joost: The experience of diagnostics of glycogen storage diseases in Estonia
13.30 – 13.45	Birutė Tumienė (Vilnius): Fanconi – Bickel syndrome: two unrelated patients with the same novel *SLC2A2* gene mutation
13.45 – 14.00	Gražina Kleinotiene (Vilnius): Follow up of patients with Gaucher disease in Lithuania
14.00– 15.00	***Lunch***
15.00 – 15.15	M-L. Uudelepp (Tartu): Case report – adult male with rhabdomyolysis and glycogen storage in muscles
15.15 – 15.30	Zita Krumina (Riga): Classic infantile Pompe disease - case report
15.30 – 15.45	Ieva Grinfelde (Riga): Hurler-Sheie syndrome - case report
15.45 – 16.45	Workshop: Glycogen storage disorders

28th of March (Friday)

9.00 – 10.00	Jorgen Bierau (Maastricht): Inborn Errors of purines/pyrimidines: a diagnostic challenge
10.00 – 10.30	Aušra Matulevičienė (Vilnius): Lesch-Nyhan syndrome. Treatment with SAMe
10.45 – 11.00	***Coffee break***
11.00 – 11.30	K. Õunap (Tartu): Hereditary fructose intolerance in Estonia.
11.30 – 12.00	Baiba Lace (Riga): Leigh syndrome, different mutations, different phenotypes
12.00 – 13.00	Workshop: purines/pyrimidines
13.00 – 14.00	***Lunch***
14.00 – 14.15	K. Päärson - Reinson: Selective newborn screening by tandem MS technology in Estonia
14.15 – 14.30	Violeta Mikštienė (Vilnius): Thiamine responsive megaloblastic anemia syndrome
14.30 – 16.00	Unknown cases: 3-4 presentations form each country
16.00	Closing remarks, discussions, future plans