

## BALTIC METABOLIC GROUP MEETING

5-6<sup>th</sup> April 2018,

Tartu

Estonia

### Program

**Meeting venue:** Conference hall Peterson, Dorpat Convention Centre, Turu 2, Tartu, Estonia

#### 5th of April

<b>9.30-10.00</b>	<b>Registration and welcome coffee</b>
10.00-10.15	Opening words from Prof. Katrin Õunap (Tartu, Estonia)
<b>Session I</b>	<b>Chair: Dr. Tiina Kahre</b>
10.15-11.00	<b>"Update on long-chain fatty acid oxidation defects: What is new?"</b> Prof. Ute Spiekerkoetter (Freiburg, Germany)
11.00-11.15	<b>"Cutoff Levels of 17-<math>\alpha</math>-Hydroxyprogesterone in Neonatal Screening for Congenital Adrenal Hyperplasia in Lithuania"</b> Karolis Sablauskas (Vilnius, Lithuania)
11.15-11.30	<b>"Diagnostic challenges of Lesh-Nyhan syndrome"</b> Dr. Laura Roht (Tallinn, Estonia)
<b>11.30-12.00</b>	<b>Coffee break</b>
12.00-12.45	<b>"Novel O-glycosylation defect"</b> (preliminary title, will be specified) Prof. Daan van Aalten (University of Dundee, Scotland, United Kingdom)
12.45-13.15	<b>Patient presentation</b> Prof. Katrin Õunap (Tartu, Estonia)
13.15-14.00	<b>"Diagnosis and Therapy of Late Infantile Neuronal Ceroid Lipofuscinosis (CLN2)"</b> Dr. Miriam Nickel (Hamburg, Germany)
<b>14.00 - 15.00</b>	<b>Lunch</b>
<b>Session II</b>	<b>Chair: Dr. Laura Roht</b>
15.00-15.20	<b>"Congenital disorders of N-glycosylation in Estonia"</b> Dr. Mari-Anne Vals (Tartu, Estonia)
15.20-15.40	<b>"Screening of congenital disorders of N-glycosylation in Latvia"</b> Ass. Prof. Zanda Daneberga (Riga, Latvia)
15.40-16.00	<b>"Congenital disorders of autophagy in epilepsy"</b> Birute Tumiene (Vilnius, Lithuania)
16.00-16.15	<b>"Familial hypertriglyceridemia"</b> Dr. Riina Žordania (Tallinn, Estonia)
16.15-16.30	<b>Dent disease and metabolic nephropathy. Case report."</b> Dr. Gita Tauriņa (Riga, Latvia)
16.30-16.45	<b>"Fanconi-Bickel syndrome presenting in neonatal screening"</b> Deimante Brazdziunaite (Vilnius, Lithuania)
16.45-17.30	Unknown cases discussion
18.30	Dinner, Tartu Ülikooli Kohvik (Ülikooli 20)

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**Chair:** Prof. Katrin Õunap

9.00-9.45	<b>"PKU and the Brain: pathogenesis of multidirectional phenylalanine neurotoxicity"</b> Prof. Maria Gizewska (Pomeranian Medical University, Szczecin, Poland)
9.45-10.15	<b>"PKU in Lithuania"</b> Laima Ambrozaityte (Vilnius, Lithuania)
10.15-10.35	<b>"PKU in Latvia"</b> Dr. Laura Ozolina, Prof. Rita Lugovska (Riga, Latvia)
<b>10.35-11.05</b>	<b>Coffee break</b>
11.05-11.25	<b>"Dietary treatment of PKU in Latvia"</b> Olga Lubina, Anna Kvasova (Riga, Latvia)
11.25-11.45	<b>"Hyperphenylalaninaemias in Estonia: Genotype-Phenotype Correlation and Comparative Overview of the Patient Cohort Before and After Nation-Wide Neonatal Screening."</b> Hardo Lilleväli (Tartu, Estonia)
11.45 - 12.05	<b>Dietary treatment in PKU in Estonia"</b> Siret Saarsalu (Tartu, Estonia)
12.05-12.35	<b>"Adult patient care"</b> Prof. Ute Spiekerkoetter (Freiburg, Germany)
<b>12.35-13.30</b>	<b>Lunch</b>
13.30-14.15	<b>"European Guidelines on diagnosis and treatment of phenylketonuria"</b> Maria Gizewska (Pomeranian Medical University, Szczecin, Poland)
14.15-14.45	Discussion about PKU guidelines in Europe and local Baltic guidelines
14.45-15.15	Baltic metabolic group meeting

17.00 Excursion to Eesti Rahva Muuseum/Estonian National Museum (Muuseumi tee 2, Tartu) <http://www.erm.ee/en>