



# **Latvijas Medicīniskās Ģenētikas asociācija**

*Latvijas Ārstu biedrības Sabiedriskās organizācijas reģistrācijas apliecība Nr.70.*

Medicīniskās ģenētikas klīnika, VAS Bērnu klīniskā universitātes slimnīca, Bērnu slimnīca Gailezerā  
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Latvian Medical Genetics Association together with Baltic Metabolic Group,  
“Biomarine” and “Swedish Orphan International AB”  
will organise metabolic meeting at 23<sup>rd</sup> -24<sup>th</sup> March 2010 in  
Children's University Hospital “Gailezers”, Juglas str. 20, Riga.

## The 23<sup>rd</sup> March

11.00-11.30	Registration, coffee
11.30-11.45	MPS VI theoretical aspects (Dr.Med. A.Jurecka)
11.45-12.15	MPS VI clinical aspects (Prof. A. Tylki-Szymanska)
12.15-12.35	Gaucher disease (Dr.Med. Grazina Kleinotiene)
12.35-13.35	Galactosemia session: Ass. Prof. K. Õunap, Clinical experience in Tartu 15 min Dr. K.Joost, Clinical experience in Tallinn, 15 min K. Krabbi, Results of urinary analyses of Estonian galactosemia patients, 15 min. Discussion
14.00-15.00	Lunch
15.00-15.35	Nieman Pick type C (Prof. A. Tylki-Szymanska)
15.35-17.30	Unknown cases (3 till 4 from each country)
18.00	Dinner

## The 24<sup>th</sup> March

9.00-9.30	Fabry patients in Estonia (Ass. Prof. K. Õunap)
9.30-10.00	Clue guidelines on metabolic diseases identification: experience (Prof. A. Tylki-Szymanska)
10.00-10.15	Krabbe leukodystrophy: case report (Dr. K. Muru)
10.15-10.30	Glycerolkinase complex deficiency (Dr.I.Mičule)
10.30-10.45	GM1 gangliosidosis (Dr.Z.Krumina)
10.45-11.00	Mitochondrial myopathy: case report (Dr. E. Vaidla)
11.00-11.15	LCHAD deficiency in 1-year-old girl: case report (Dr. M.-L. Uudelepp)
11.15-11.30	A girl with Schwartz – Jampel syndrome, type I. Case report (Dr. B.Skerliene)
11.30-11.45	Pyruvate dehydrogenase deficiency - a case report (Birute Tumiene)
12.00	Closing remarks
12.30	Lunch