Training report

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Host laboratory: Dept. of Biochemical Genetics, Academic Hospital Maastricht, 6202 AZ Maastricht, the Netherlands
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Already second time I would like to thank ERNDIM for the given opportunity to broaden my knowledge and experience in the laboratory diagnostics of inherited metabolic diseases.

For many years in Lithuania we had scanty possibilities in laboratory diagnostics of IEM. Except newborn screening for PKU and congenital hypothyroidism, almost all other diagnoses of IEM were made merely on the basis of the clinical picture or with help of laboratory diagnostics in laboratories abroad. This situation led to development of vicious circle: "no laboratory diagnostics – no IEM". And the majority of IEM were buried under the conceptual names. Blurred understanding about IEM lowered their significance in Health insurance system – as the result – poor financial support from the Government followed.

The first ERNDIM grant in 2006 opened for me the window to the field of laboratory diagnostics of IEM. Two weeks in the Academic Hospital Maastricht gave me a good start and understanding in organisation of the biochemical genetics laboratory and as well as experience in number of laboratory techniques and interpretation of results. Moreover, modelled by Dutch colleagues' example, foundation of the Baltic Metabolic group (colleagues from Estonia, Latvia, and Lithuania) was initiated with the purpose to discuss our specific problems, to exchange experience and achievements in clinical and laboratory diagnostics of IEM. The first Baltic metabolic meeting was held on March, 2007.

According to advices of the Dutch colleagues the purchase of the most required laboratory equipments was performed.

Purchase of a gas chromatograph – mass spectrometer by our Centre for Medical Genetics in 2007 raised necessity of the training in organic acids assay.

The second visit in the Biochemical genetics laboratory of Academic Hospital Maastricht, covered by ERNDIM grant gave me possibility to learn sample preparation for organic acids assay by GC-MS, calibration, method validation and interpretation of chromatograms. This was highly important for us, as our Centre for Medical Genetics was the first in Lithuania which started organic acids assay. The case of 4-OH-butyric aciduria found in our laboratory in Lithuania was an encouragement for me to continue.

Two days during my visit I spent in the Laboratory Genetic Metabolic Diseases of the Academic Medical Center of the University of Amsterdam with the aim to train myself in the creatine and guanidinoacetate assay by GC-MS.

Advices and examples given by Leo Spaapen, Ries Duran and Nico Abeling gave me ideas about rational arrangement of our new laboratory, which is planned to be build in 2010 and which architectural design was finished in 2008.

With growing enthusiasm I would like to admit that under the guidance and help of more experienced colleagues it is possible in a few years to step over more than a decade of stagnation and to start a new, productive period in the laboratory diagnostics of IEM. Moreover, I would like to thank ERNDIM not only for financial support; I would like to thank ERNDIM for activities which help to raise importance of laboratory diagnostics of IEM in my country.

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