Training report

2014.11.18

Host laboratory: Laboratory for Biochemical Genetics (EMZ), Department of Clinical Genetics, Maastricht University Medical Centre (AZM), Maastricht, the Netherlands

Training period: 2014.10.27 – 2014.11.14

First of all, I would like to thank ERNDIM for giving me an opportunity to have a training in the state-of-the-art metabolic laboratory with modern analytical techniques and highly professional staff. I believe that the knowledge and experience that I have gained during this training will definitely help our laboratory to speed up the progress of mastering (U)HPLC-MS/MS technique for diagnosing genetic metabolic diseases.

Testing for metabolic diseases in Lithuania is still developing and our lab has just recently started to implement tandem MS tor clinical diagnostics. That is why we needed to have this training and to get advice from more experienced colleagues about the analytical methods, their application for routine testing.

During my stay I have seen and studied several analytical methods on the tandem MS. The most important one for our lab is analysis of acylcarnitines in plasma, which we are planning to implement in the near future. Seeing how the analysis is performed by skilled technicians and being able to discuss various technical details with them was very helpful in gaining deeper understanding of the process.

I have also studied the analysis of very long chain fatty acids in plasma and analysis of purines/pyrimidines in urine and plasma, which are of great interest for our laboratory too.

What is more, I had a chance to study analytical methods which use an ion-pairing reagent (analysis of homocysteine/cysteine in plasma and urine; analysis of creatine, guanidine acetate and creatinine in urine and plasma). At the moment we do not use methods with ion-pairing, but the other methods for these analytes – GC/MS to quantify creatine and guanidine acetate in urine, and qualitative test for HCys/Cys in urine. The same can be said about the analysis of sialic acid, which we now perform only qualitatively by TLC, while the quantitative analysis of both free and total sialic acid on tandem MS seems to be a simple and reliable method for revealing such diseases as sialidosis, SASD, etc. Knowing more about different alternatives to the methods we use will help to compare various analytical techniques and their performance capabilities. This information will definitely be useful in the future, for improving our methodology and expanding the analytical and diagnostic capabilities of our laboratory.

In addition, learning the method of analysis of methylmalonic acid in plasma for diagnosing the deficiency of vitamin B_{12} was of great advantage, as more and more physicians show interest in this analysis and implementing it in our lab could be beneficial.

It was also of great importance to get advice on the interpretation of analytical results. The colleagues from Maastricht have provided me with the pathological patterns, characteristic for various metabolic diseases, especially the ones that can be diagnosed by measuring acylcarnitines concentrations in plasma. As metabolic diseases are very rare, having the results of analysis of real patients is surely a big advantage, so we wouldn't miss some patients in our future work.

To sum up, during this three weeks I have seen and learned a lot of things that will be of great importance for the future of our lab and I believe would have a positive impact on all our Centre of Medical Genetics. Staying in touch with colleagues in Maastricht will also help to get advice in case of various questions in the future.

Many thanks to everyone working in the EMZ for being very friendly and helpful during my stay: Jean-Pierre, Ann, Sandra, Hoob, Karen, Daphna, Ping, Daisy, Marjon, Ardy, Loes, Jeannine, Carla, Vivian, Marc and others! And of course, my biggest gratitude to Jorgen, who agreed to have me in the lab for this training. It was great pleasure working and learning from you all!

Yours sincerely,

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