

CENTER FOR MEDICAL GENETICS VILNIUS UNIVERSITY HOSPITAL SANTARIŠKIŲ KLINIKOS

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2006-06-30

Dear professor Brian Fowler,

Once again I would like to thank you for the given possibility to broaden my knowledge and experience in the laboratory diagnostics of inherited metabolic diseases.

Two weeks $(06\ 04\ -\ 06\ 18)$ in Dept. of Biochemical Genetics Academic Hospital Maastricht were highly important for my understanding in laboratory work organisation, experience in a number of analytical techniques and interpretation of the results.

Laboratory co-workers favoured me by with full openness and cooperativeness. They have demonstrated me numerous features of the analytical procedures and interpretations of the results. I have participated in the performance of a large number of laboratory techniques: thin-layer chromatography of urinary amino acids, imidazoles, oligosaccharides, sialic acid containing oligosaccharides, electrophoresis and quantification of mucopolysaccharides (glycosaminoglycans), and isoelectric focusing of sialotransferrins.

Furthermore, I have discussed on plasma amino acid analysis with experts of HPLC analytical techniques and have followed demonstration of the GCMS analysis of urinary organic acids. We plan to purchase HPLC and GC/MS at the end of summer for our laboratory in Lithuania, so all methodological details and also a number of required laboratory accessories were very important.

Unfortunately, the period of traineeship was far too short to obtain sufficient insight to interpret the results of complete diagnostic programme and to get enough experience to perform GCMS analyses of organic acids. Hopefully, good contacts and continuing cooperation with Dutch colleagues will help to dissolve any possible problems of our future laboratory works.

In addition I have participated in a meeting of Dutch colleagues from Rotterdam, Amsterdam and Utrecht. Several hour discussions about complicated cases in the laboratory diagnostics of inherited metabolic diseases and demonstration of the way towards final diagnosis during this meeting have left me a deep impression.

The first real benefits of this traineeship just after my return to Lithuania are:

- I could reasonably expand the list of required laboratory equipments in our new project "Improvement of genetic consulting and diagnostics in Lithuania".
- 2. I have translated into Lithuanian and modified several methods (thin-layer chromatography of urinary amino acids, imidazoles, oligosaccharides, sialic acid containing oligosaccharides, electrophoresis and quantification of mucopolysaccharides (glycosaminoglycans).
- 3. I have ordered lacking reagents in order to improve existing and introduce new methods in our laboratory.

The nearest plans would be to introduce HPLC and GC/MS methods in our laboratory here in Lithuania, as soon as the equipments will be purchased, and after that proudly to join ERNDIM quality control.

I would ask to convey special thanks to dr. Leo Spaapen and his colleagues in Dept. of Biochemical Genetics Academic Hospital Maastricht for warm-hearted and highly professional assistance, multiple support, continuing attention and fruitful cooperation.

Please, see financial documents of a major part of my traineeship expenditure, which was funded by ERNDIM (the grant was 691,99 EUR), attached payment

confirmations for: lodges in Guest House of the Maastricht University -430,00 EUR, costs of the flight Vilnius - Amsterdam –Vilnius -343,00 EUR and train airport - Maastricht v/v - 54,60 EUR.

Truly grateful,

Jurgita Songailiene, MD. PhD

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