

## Training Grant Report

I had, thanks to the ERNDIM Travel Grant, a possibility to spend two weeks (07.05.12 - 18.05.12) in VU Medisch Centrum in Amsterdam. My aim was to get experience of analysing sugars/polyols and 7-dehydrocholesterol using gas chromatographic (GC) methods with mass-spectrometry (MS).

At the moment we are using our GC-MS instrument for analysing Organic Acids, Very Long Chain Fatty Acids as well as Creatine/Guanidinoacetate. There are still more possibilities, and sugars/polyols and 7-dehydrocholesterol are some of the compounds for we could use GC-MS. We are measuring some of the sugars on HPLC, but GC-MS method allows to measure much more compounds simultaneously. Some polyols are very important to get information about pentose phosphate pathway. There are two new disorders and one of them, Transaldolase deficiency, seems to be not very rear. It is still quite difficult to estimate the incidence of it at the moment, because this disorder is so new. Also, the clinical picture is quite unspecific, hepatic dysfunction being main symptom, and can also overlap with that of classical Galactosemia. For those reasons it would be very useful to measure more compounds for patients with hepatic dysfunction to get more information and be able to exclude disorders in pentose phosphate pathway.

The second method was for analysing 7-dehydrocholesterol (and some other sterols like stigmasterol, sitosterol). There is already one patient in Estonia diagnosed with Smith Lemly Opitz syndrome, the most frequent disorder of cholesterol biosynthesis. Analysing 7-dehydrocholesterol in Estonia, we would reach quicker to the diagnosis.

I would like to thank very warmly all people in the laboratory and also ERNDIM to give me such a great opportunity. All the staff in VU Medisch Centre's Metabolic Lab was very friendly. I had possibility to watch how they perform the analysis. I got very useful mass-spectrums of the compounds. Also, we discussed with Dr. Mirjam Wamelik about interpretation. It is not easy to interpretate results in clinical analysis and it takes time to get skilled, but now a beginning has been made and it will be much easier to introduce those new methods as it would have been studying from a book.

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