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## To the Executive Committee of ERNDIM

## **Re: ERNDIM Training Grant Report**

This is a report referring to the ERNDIM funding of my visit to the Biochemical Genetics Laboratory, in St James's University Hospital, in Leeds, UK, for the period 14 - 25/10/2013.

I am a Medical Doctor, board certified in Medical Biopathology, and a Lecturer of Medical Biochemistry in the Faculty of Medicine of the Aristotle University of Thessaloniki, in Greece. For the last two years (2012 and 2013), after a year of training, I am a collaborator in the Metabolic Laboratory of the 1<sup>st</sup> Pediatric University Department, in Hippokration Hospital, Thessaloniki, Greece.

So far, during this two-year period, I have attended SSIEM Academy courses 2012 and 2013 and I have been trained in our lab in techniques, such as TLC (1D and 2D) for amino acids, HPLC for amino acids in biological samples, such as plasma, urine and CSF, using OPA for derivatization. The need to deepen my knowledge in these assays and broaden my horizons into new techniques urged me to ask Dr. Mick Henderson to accept me as a visitor in the Biochemical Genetics Laboratory in Leeds, UK, for a fortnight. He kindly accepted me to visit the Lab and offered me the opportunity to get acquainted with neonatal screening assays and urine organic acid determination.

Neonatal screening for Phenylketonuria, Hypothyroidism, Galactosemia and G-6-PD deficiency is performed in Greece, but only in Athens and not in Northern Greece. My visit in Leeds gave me the opportunity to get to know the techniques used in NHS for Newborn Screening and for the Expanded Screening Programme, using Tandem MS and fluoroimmunometric methods. This way I feel confident in performing screening in case a Tandem-MS is provided to our lab.

Moreover, urine organic acid analysis is an assay of high importance for pediatric diagnostics and, in particular, for possible diagnosis of an IEM. In our Metabolic Lab, and generally in Northern Greece, this assay is not performed. During my visit in Leeds, I have been trained in urine organic acid analysis, using GC-MS. I got acquainted with the analytical procedure, as well as with the interpretation of the results and I had the opportunity to discuss any queries on analytical methods already used in our Lab. Moreover, I gained experience in the diagnostic service for neuroblastoma using HVA and HMMA, with the use of GC-MS. During my two-week long visit in Biochemical Genetics Lab in Leeds, funded by ERNDIM, I gained a lot of experience. I now feel confident in introducing the methods in which I have been trained, in our Lab and, in this way, facilitate the clinical diagnostic procedure in the area of Northern Greece, as long as Hippokration Hospital is the largest hospital in Northern Greece.

I would, particularly, like to thank Dr. Mick Henderson, Head of the Laboratory in Leeds, for all the support during my visit in the Lab, for all the time he spent with me in tutorials, discussion and advice on issues in relation to IEM and on the analytical procedures, and all the hospitality, in general. I would also like to thank all Advanced Clinical and Biomedical Scientists, as well as all laboratory staff members, who helped me in my training and made my visit pleasant and useful, sharing generously their expertise; and most important, I thank them all for reassuring me of helping me out with any query in my initial steps performing the techniques I learned in our Metabolic Lab in Thessaloniki.

Last but not least, I would like to thank ERNDIM Executive Committee for approving my application for the financial support during my training in the Biochemical Genetics Lab in Leeds.

Sincerely,

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Lecturer of Medical Biochemistry