



UNIVERSIDAD AUTÓNOMA DE NUEVO LEÓN 🔲 FACULTAD DE MEDICINA Y HOSPITAL UNIVERSITARIO / Departamento de Genética

Travel Grant Report

I would like to thank ERNDIM for the opportunity I had to attend the molecular diseases laboratory (CEDEM), Faculty of Sciences, module 10 at the Autonomous University of Madrid, and the great support provided by Dr. Pedro Salas and the staff of the laboratory for sharing its great experience in the interpretation and quantification of organic compounds in urine. In addition, Dr. Begoña Marinero was the main contact to carry out this stay in her laboratory.

I would also like to thank Dr. Brayan Fowler who recommended me to apply for a scholarship to the ERNDIM to carry out this stay and to Dr. Magdalena Ugarte who is the head of CEDEM and accepted my request.

The main objective of my visit in the CEDEM was to review the technique of quantification of organic compounds in urine, since we only perform the qualitative analysis for it in our department and that is very important to us. To do this, I learnt how to elaborate the calibration curves of certain analytes and support with experience of Dr. Pedro Salas, using the Agilent equipment, which has a different software from our laboratory (CG / MS Clarus 500 by Perkin Elmer).

To realize the calibration and quantification curves of several analytes, I learnt that a mixture of them can be made, for the process, in such a way that I will implement it in my laboratory.

Second objective of my visit, to reinforce the knowledge of interpretation the chromatograms that you face every day: to achieve this objective, I checked multiple chromatograms of real patients and quality control of the ERNDIM. It was very helpful to consider the - of my results interpretation, to generate a reliable report.

In the future I would like to participate in quality control of the ERNDIM in scheme for the analysis of organic acids in urine, if possible on filter paper since in our country we have many problems for the importation of biological samples in liquid state.

During this stay, I had the opportunity to learn about other techniques such as quantification of methylmalonic acid in plasma, galactitol and galactonate in urine, very long chain fatty acids in plasma, acylcarnitines in plasma, Lactate, Pyruvate, 3-OH Butyrate and Acetoacetate in plasma and of Galactose 1 Phosphate Uridyltransferase in erythrocytes and we are considering the possibility of implementing these techniques in my lab.

Finally, they asked to interpret 3 patient chromatograms of a quality control from France and later to be present in the discussion and conclusion of the case to send the report to the quality control. It seemed very important to me that all the personnel involved are incorporated to achieve the diagnosis and report the case.

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