

Travel Grant Report

Dr Jörgen Bierau (Head of Laboratory) kindly allowed me to spend four days in the Biochemical Genetics Laboratory (EMZ) at Maastricht University Medical Centre (azM) in October 2015. The purpose of my visit was to gain experience in the analysis and interpretation of purines and pyrimidines in urine and plasma by UPLC-MS/MS.

Disorders of purine and pyrimidine metabolism can have a very diverse and non-specific presentation. Symptoms can include anaemia, deafness, developmental delay, immunodeficiency, renal stones and seizures, to name a few. Because of this, and the requirement for purine and pyrimidine analysis to be a part of the DPT EQA scheme, laboratories should consider including analysis of purines and pyrimidines as part of their workup for inborn metabolic disease. Unfortunately, disorders of purine and pyrimidine metabolism are not widely tested for in the UK, and there is currently only one national laboratory that provides this analysis. The purpose of my visit to Maastricht was therefore to gain the knowledge to enable us to introduce testing for purine and pyrimidine disorders into our repertoire in Leeds.

During my time in Maastricht, the staff were incredibly helpful and friendly. Sandra and Huub were particularly accommodating, demonstrating sample processing and data analysis, and discussing method development. Jörgen and Daphna spent time with me discussing some of the basic interpretative aspects of purine and pyrimidine analysis, although it was impossible to cover it all as it is such a huge and mind-boggling field! Ping also spent some time with me covering some of the statistical aspects of the work, including some of the other very interesting projects currently underway at the centre. One aspect of my visit that was particularly memorable was finding out why the phrase “urine monsters” was so prevalent in the SOP! (I was informed that “monster” is Dutch for “sample”!).

I very much enjoyed my time in Maastricht, and I found the Netherlands to be a fascinating place. I would like to say a big thank you to Jörgen and his team for being so welcoming, and also to ERNDIM for providing the funding enabling me to attend.

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