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ESPE Sabbatical Leave Programme 2014
Recipient:
Associate Professor Giedre Grigelioniene
Dept of Clinical Genetics/Dept of Molecular Medicine and Surgery
Karolinska University Hospital/Karolinska Institutet Stockholm, Sweden

Host: Professor Harald Jueppner,
Endocrine Unit, Endocrine Unit; Department of Medicine, Massachusetts General Hospital, Boston USA
20150501-20160731

Aim: Searching for genetic causes of pseudohypoparathyroidism type Ib.

Progress and achievements:
I had the great privilege of spending a stimulating year at the Endocrine Unit, MGH, Boston, MA, USA. This sabbatical year was made possible by the grant from the ESPE Sabbatical Leave Programme, supported by Eli-Lilly International Corp, USA. The new academic environment was very stimulating. I regularly attended laboratory and clinical meetings at the Endocrine Lab, MGH, unit academic activities as well as Skeletal Society seminars and workshops.

I worked on the project looking for the genetic cause of PHP Ib in a Finnish family with PHP Ib with an affected mother and her two children, in whom previously known genetic causes for PHP Ib were excluded. We have discovered that a genetic rearrangement, an inversion which broke apart the GNAS locus, was the cause of PHP Ib in this family. Such a genetic rearrangement as cause of PHP Ib is discovered for the first time and the results of this study are published in Journal of Bone and Mineral Research (PMID: 28084650).

In addition to the project described above I also had the opportunity to visit clinical education sessions at the department of Clinical Genetics, observe work of professors Angela Lin and Paula Goldenberg. Furthermore I have established collaboration with Associate Prof Tatsuya Kobayashi group and we started a project together on two novel skeletal dysplasias. The functional studies on both are ongoing.

I have much enjoyed and benefitted from this sabbatical period, learning new techniques (analysis of WGS, CRISPR/Cas9 and RNA sequencing) as well as novel approaches to translational research. I am very grateful to the ESPE Sabbatical Leave Committee and to Lilly Pharmaceuticals for making my sabbatical year possible.

I am very grateful to ESPE for this excellent opportunity at this stage of my career to learn new research skills, while expanding my collaborative network and gathering new ideas for continued research.

Sincerely,
Giedre Grigelioniene