

ERNDIM Training Grant REPORT

Grantee: Esphie Grace D. Fodra

University Researcher and Team Captain

Biochemical Genetics Laboratory

Institute of Human Genetics, National Institutes of Health,

University of the Philippines Manila

Host Laboratory: Metabolic Laboratory, Dept. of Clinical Genetics,
Rigshospitalet, Copenhagen

Head of Host Laboratory: Dr. Flemming Wibrand

Inclusive Dates: 14-24 September 2015

My institute and I would like to express our sincere gratitude to ERNDIM for this very worthwhile opportunity. This help which was extended to us through this grant will be of great benefit to our laboratory and to the patients whom we serve.

I am equally grateful to the host lab under Dr. Flemming Wibrand whose generosity and kindness was very much sensed before and during my training, and I believe even as we go along our set-up of the new tests which I have learned.

I was primarily educated on the basic metabolic tests for lysosomal storage diseases: glycosaminoglycans assay and screening, and lysosomal enzyme assays. LSD is the second most prevalent IEM in our country. To be able to offer the necessary tests for this group of diseases locally (i.e. not send out overseas which we currently do), our patients would benefit from faster turn around of results hence faster assessment, and eventually lower costs. It will likewise pave the way for staff learning, development, and growth in terms of the IEM discipline in our country.

The highlights of my training to which I am all-grateful about were as follows:

• **Technology**

On the technical aspects, I believe that I was able to accomplish the purpose of my visit. With the spectrophotometric testing of GAGs by dimethylene blue assay, automation is apparent, and the importance of having a robust machine is emphasized. This, of course, should be complemented with execution of efficient laboratory procedures. For GAGs screening via electrophoresis which is in fact a “very old method” as most would agree, it still is a very good technique as the screening per se would save on costs and time since it determines which enzyme assay/s should be done. Should time allow, however, I think that developing this method into one with a shorter run time, would prove to be of worthy gain. The lysosomal enzyme assays which essentially make use of fluorimetric technology for the different mucopolysaccharidoses types are likewise quick

and effective. Doing hands-on exercises on these including that for GAGs screening was indeed a significant know-how. It is often a better learning tool to actually do than just observe, and I am thankful that the laboratory allowed me to do this (special thanks to Kirsten who was showing me the procedures most of the time). In addition, one of Dr. Wibrand's suggestion which could be of good use to us is to look for a machine which has both spectrophotometric and fluorimetric components. We may be able to use it for creatinine, DMB, and the enzyme assays alike should there be one in our local market.

- **Facility and Administration**

With regard to facility, I would say I have observed an ideal set-up at Dr. Wibrand's lab. Ideas on segregation of rooms for specific tests and equipment, designated instrumentation room for highly specialized machines, employee lockers and lounge, staff offices, and security concerns among others, may be used on the planning of our new building. I have also learned about practical techniques on doing routine lab assays, and the importance of having periodic laboratory meetings.

- **People**

May I, in the same way, comment on the staff in general- everyone is so friendly and helpful, for which I'm really glad about. Copenhagen as a city was similarly nice and food was awesome. Thanks also to Michelle who gave me a tour on some of the tourist spots. Beautiful people and place really!

- **Other Accomplishments**

During some time, I was also able to consult and discuss some of our own concerns regarding our tandem mass spectrometer which is used for acylcarnitine quantitation. Mette Christensen, one of the principal scientists there, was also very accommodating to address my questions. Dr. Jakob Ek likewise gave me a brief overview about their molecular genetics unit. On one instance, too, I was able to attend a short presentation about a genetic research study.

Overall, it was such a rewarding experience and I would like to express my genuine appreciation once again to ERNDIM, to Dr. Wibrand and team, to our laboratory head and institute director- Dr. Mary Anne Chiong, and our university for allowing me to be involved in an endeavor as this. I hope that the fruits of our joint efforts will manifest soonest. Glory to God.