



מרכז רפואי הדסה ירושלים
Hadassah Medical Center Jerusalem

The Department
of Genetics
and Metabolic Disease

המחלקה
לגנטיקה
ומחלות מטבוליות

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REPORT ON ERNDIM TRAINING GRANT

I'm most grateful to ERNDIM for providing me the opportunity to visit and receive training at CMMS- The Center for inborn metabolic diseases at Karolinska University hospital in Stockholm, which the main metabolic disease and NBS screening department in Sweden. The CMMS laboratories are equipped with state-of-the-art instruments and facilities, and are staffed by highly trained, expert professionals.

The purpose of my visit was to receive training in techniques important for the diagnosis and treatment of IEM, with the main focus on methodologies related to organic acids, acylcarnitines and amino acids

At CMMS, I interacted with several experienced staff members learning different aspects from each profession. From the physicians, I gained insight in how to interpret results. From the chemists, I learned about the instrumentation and computer programs. Finally, last but certainly not least, experienced biomedical analysts showed the several methodologies including sample preparation and analysis.

Additionally, I participated in several internal meetings and discussing related to diagnostic cases and technical issues. From these meetings, I learned the importance of real team work; laboratory management and the necessity of writing good standardize operating procedures.

I really appreciate the kindness of the whole staff and I'm especially thankful to Dr. Martin Engvall and Dr. Rolf Wibom for taking some of their precious time to guide me around in their laboratories, and to Prof. Anna Wedell, head of CMMS for allowing me to visit and to participate in meetings.

The experiences gained from this enjoyable visit, will enable us to expand, improve and standardize metabolic tests performed in the Metabolic and Enzyme laboratory at the Hadassah-Hebrew University Medical Center in Jerusalem (serving a large part of the Israeli and Palestinian populations)

Sincerely yours

Ann

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