

## Laboratory Training with Prof. Ralph Fingerhut at Neonatal Screening laboratory, University Kinderspital, Zurich, Switzerland (March 9 – April 4, 2015)

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I attended Prof. Ralph Fingerhut's Neonatal Screening Laboratory at the University Kinderspital, Zurich, Switzerland, from March 9, 2015 to April 3, 2015 for training in screening for metabolic disorders through Tandem Mass Spectrophotometry (TMS), Fluorometric Immunoassays and Gas Chromatography-Mass Spectrophotometry (GC-MS).

As a part of the ongoing collaboration for a pilot study to screen newborns in Nepal for metabolic disorders between Ralph Fingerhut, Brian Fowler (UKBB, Basel, Switzerland) and myself, 1500 Guthrie cards had been shipped to Prof. Fingerhut's Laboratory by the time I arrived at his lab. This gave me an opportunity to carry out the different assays included in our pilot study, on the samples from Nepal. I got first hand experience in carrying out the assays available at Prof. Fingerhut's lab. The samples were assayed for congenital hypothyroidism (TSH), congenital adrenal hyperplasia (CAH), cystic fibrosis (IRT), galactosemia (galactose and Galactose 1- phosphate uridyl transferase (GALT)) and biotinidase deficiency on GSP instruments using GSP kits from Perkin Elmer. Amino acids and acylcarnitines were measured to screen for Phenylketonuria, Glutaric acidemia, Medium chain acyl carnitine deficiency and Maple syrup urine disease using Tandem mass spectrometry.

It requires a high deal of organization and a sophisticated tracking system to ensure the correct placement of data with respect to the samples. The Tandem Mass Spectrometer, GSP instruments and punching machines are on a network and communicate with each other. As the samples are punched into the wells, the barcode representing that sample is read into the machine, which is read by the rest of the machines on the network. I learnt about the use and importance of quality controls and their influence on the measurements. Calculations are performed with close attention to the QC values and ensure the correct conduction of the assays.

The lab uses the software "Lifecycle" to integrate all demographic and assay data. The dissemination of information is done carefully after a confirmatory second assay along with a second tier test. There is a strong system of communication with other hospitals and attending physicians, so that disorders with serious consequences are dealt with in time to prevent severe damage to the infant.

Organization of a tracking system, conduction of assays, calculations and interpretations, second tier testing and communication with the sample source institution constitute the running of a neonatal screening program. The pilot study in progress in Nepal will help identify the disorders screening for which will benefit the health and survival of the Nepalese infant. This information will form the basis for starting screening services starting with my employer institute Kathmandu Medical College and Teaching Hospital. Although the automated system is neither affordable, nor required in Nepal, it has demonstrated to me the need for the merging of data and analysis therein within a tight tracking framework.

Gas Chromatography Mass Spectrophotometry (GC-MS): I learnt in detail the method followed at the Clinical Laboratory at the Zurich Kinderspital to extract organic acids from urine and measure them in GC-MS. I was introduced to the analysis of the GC-MS chromatograms in comparison with the normal and the identification of peaks for common drug metabolites. With one of the hospitals in Kathmandu having acquired the GC-MS equipment, testing urine samples for organic acidemias using this machine is now a possibility.

Zurich is a beautiful city with a unique architecture and historical churches. I enjoyed the city despite the rain and the highlight was my visit to the Pizol Ski Resort with Dr. Susanna Sluka. I enjoyed getting to know Yvonne, Corazon, Esther, Karin, Ruth, Alinka, Katie, Sandra (both) and Monica. I think I have made some very good friends. I was fortunate to be a part of the 50<sup>th</sup> anniversary of Neonatal Screening in Switzerland organized at the kinderspital by Prof. Ralph Fingerhut. In the end, my heartfelt thanks to Prof. Ralph Fingerhut for giving me the opportunity to visit his lab and be a part of it for a month. I also thank ERNDIM for making this a possibility.

