

EuroGentest Training Report

Shivaram S. Shastri

I had the privilege to obtain training at the Cystic Fibrosis Centre, Institute of Biology and Medical Genetics, Prague, Czech Republic under Professor Milan Macek Jr., from 10th May 2008 to 1st June, 2008. The training was funded by EuroGentest, and coordinated by Prof. Jean-Jacques Cassiman of Leuven Catholic University, Belgium.

In those three weeks, I was able to learn some of the important and emerging techniques in the detection of pathogenic changes in the human genome that cause cystic fibrosis. The techniques I learnt included DNA sequencing, MLPA (Multiplex Ligation Dependent Probe Analysis) and HRMCA (High Resolution Melting Curve Analysis). I was able to run many experiments of HRMCA and MLPA on my own. I also had the opportunity to learn some of the routine protocols used for mutation screening in cystic fibrosis, including extraction of consistently high quality DNA from blood using automated systems, hybridization based assays etc. In addition, I could study the organization of the laboratory, generation of standard operating protocols, quality control in a molecular genetic laboratory set up.

The friendly and knowledgeable team at the centre taught me the intricacies of these techniques. I had many useful discussions with team members who took effort in clarifying all my doubts and gave many useful tips. The training was hands-on and it was possible to identify some of the mutations previously undetected by the SSCP (Single Strand Conformation Polymorphism) in a set of Indian patients' samples. The knowledge gained here will be invaluable in starting similar services for the cystic fibrosis patients in India.

I would like to thank all the team members of the CF centre, particularly Dr. Alexandra Stambergova, Dr. Jana Čamajova, Dr. Petra Krenkova, Dr. Patricia Norambuena, Dr. Mirka Balasckova and Dr. Irena Eliasova. I am immensely grateful to Prof. Jean-Jacques Cassiman, Professor Milan Macek Jr. and EuroGentest NoE for this worthwhile experience.

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