

Subject: Adil's Eurogentest Report

In Oman it is estimated that 1 in 6000 is born with a defect in cystic fibrosis transmembrane regulator (CFTR) gene. The clinical features of cystic fibrosis (CF) individually (bacterial infection, lung inflammation, and elevated electrolyte levels in sweat) resemble those of other diseases such as pneumonia, bronchiectasis, asthma, failure to thrive and celiac disease. Indeed, where these conditions are prevalent CF may still lie unrecognized. Moreover, if clinicians believe that CF is absent from their population they will not consider it in a differential diagnosis. A better awareness of CF and the increasing availability of diagnostic tests such as the sweat test and/or DNA tests, frequently leads to the identification of a higher number of affected individuals.

Additionally, in Oman approximately 20% of couples are infertile and more than 50% of them are due to men being sterile, therefore, we thought to investigate the molecular aspect of male infertility by testing for microdeletions in chromosome Y which have never been yet studied in Oman. Therefore, I undertake a project under Prof. Milan Macek Jr. M.D. Ph.D (*Chairman of the Institute of Biology and Medical Genetics, Charles University Prague*) to investigate CF and men infertility in Omani population. The training was held at the Prague Institute of Biology and Medical Genetics – Centre for Reproductive Genetics (Charles University, Prague) under the sponsor of Eurogentest.

The aim of the training was to learn the basic molecular techniques required for analyzing the most common mutations in CFTR gene and how to test for microdeletions in chromosome Y. The training was focused on how to isolate DNA from blood sample, observing and performing techniques, include PCR, high-resolution DNA melting analysis, and sequencing. Due to some problems with the genetic analyzer I only observed how multiplex ligation-dependent probe amplification (MLPA) is performed. The training was very helpful and fruitful because I came to know about new techniques that I did not come across (High-resolution DNA melting analysis & MLPA).

Oman is a multicultural society that includes people from different origin (e.g. African, Baluchi, Pakistani, Indian, and Iranian), so we expect to get various mutations in CFTR

gene. Therefore, this study will help in recognize the common CFTR mutations in Oman and it will provide a definite diagnosis of CF. It will also help us to know the genetic alterations responsible for men infertility in the Omani population.

I would like to take this opportunity and thank Prof. Milan Macek for his kind support and guidance and all the technicians, M.Sc students, Ph.D students and staff for teaching me the techniques I require to know and for their help and support. I am very grateful for Eurogentest for giving me this opportunity.