SMP: Genetic Epidemiological Methods (GEM)

Project: Interfacing of High-throughput Genotyping and Genetic Epidemiology Standards for Data Communication, Storage, Retrieval and Analysis

Thomas F. Wienker - **Friedrich Wilhelms University, Bonn - wienker@uni-bonn.de**

Introduction

Within the SMP-GEM this subproject has got a twofold mission: to develop and to implement standards for data exchange and data validation, and to provide support for genetic epidmiological projects run within NGFN-2 with respect to qualitiy control and quality management of genotype data generated by high-througput processes in specialized genotyping centers (Berlin, Kiel, Munich, companies). The working plan has to acknowledge the contradicting aspects of high-volume and high-throughput, low-cost processes on the one side and high quality standards and low drop-out rates on the other in a coordinated and balanced fashion. The subproject has to deliver its services in a changing working environment and with tools under permanent development.

Fig 1: *2D-Barcode*

Results/Project Status

The first half year of the second funding period was essentially focussed on finishing up projects from within the disease oriented networks of the first funding period. In order to do this, software for association analysis and family based linkage analysis was used, which had been developed during the last funding period.
At the same time, meetings took place with the genotyping centers in Bonn and Munich aiming at a standard data exchange format starting with the introduction of the new high-throughput genotyping platform by Illumina. In this context the use of standardized 2D-barcodes (according to ISO/IEC) for the labeling of blood samples was suggested and discussed in order to replace of the older person identifier (PID). Especially documenting and tracing of one person's multiple DNA-samples during the multi-layer process of genotyping and data analysis should be facilitated. In a first study at the university clinic of Bonn all the prerequisites have already been established and the new practice is being tested.

Outlook

Primarily suggested for the sector of clinical-epidemiological projects within NGFN, a universal sample-ID according to international ISO/IEC norms as it is suggested by this project can also be interesting for other sectors of the research network as well, e.g. for high-throughput research in genomics and proteomics, in general. The proposed solution constitutes the introduction of an industry standard into quality management of scientific research projects. High-throughput technologies involving machine-readable interfaces will play an ever growing role in genome research. From this point of view, these and other industry standards are indispensable in the long run, as far as quality and economy are concerned. Thus, in this field NGFN can perform important pioneering work.

Lit.: ***1.*** *Kurz, T., Strauch, K., Dietrich, H., Braun, S., Hierl, S., Jerkic, S.P., Wienker, T.F., Deichmann, K.A., Heinzmann, A. (2004) Multilocus haplotype analyses reveal association between 5 novel IL-15 polymorphisms and asthma. J. Allergy Clin Immunol .2004 May; 113(5): 896-901* ***2.*** *Becker T, Knapp M. Maximum-likelihood estimation of haplotype frequencies in nuclear families. Genet Epidemiol. 2004 Jul;27(1):21-32.* ***3.*** *Becker T, Knapp M: A powerful strategy to account for multiple testing in the context of haplotype analysis. Am J Hum Genet. 2004;75(4):561-70.*