

**Place:** Academic Hospital, Uppsala, Sweden.

**Attendees:**

Uppsala: Ann-Christine Syvänen, Tomas Axelsson, David Fange, Lars Backström, Andreas Dahlgren

Uppsala: Inger Jonasson, Jenny Jonsson, Ulf Gyllensten (arrived 13.30)

FGC: Aarno Palotie, Elisabeth Widén, Anne Leinonen (behalf of Juha Muilu)

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### **Microsatellite genotyping at Helsinki**

Elisabeth Widen reported that FGC has genotyped 3 plates of Danish samples and 3 plates of Finnish samples with chromosomes 1-11. Also some other TWIN projects have been genotyped with all markers and that data will be shared with genomeutwin later. FGC was ready to start genotyping earlier, because infrastructure was ready and marker set was familiar, also there was some Finnish twins available at that time. FGC did not have any experience in twin genotyping beforehand.

### **Microsatellite genotyping at Uppsala**

Inger Jonasson and Jenny Jonsson reported that they have genotyped 3 plates of Danish samples. Genotyping chromosomes 12-X will be finished by the end of summer. Finnish twin samples have to be plated for them also.

There was some problems before getting working PCR protocol (different marker set for them)

**Action Point:**

Success rates of markers (ch. 11-X ) will be compared with FGC data available.

### **Selection of twin samples for genotyping**

Aarno Palotie told that twin registers are now going over their samples. There is not enough DNA of Dutch samples now, so they will be out of question for some time. Center should have enough DNA for microsatellite scan, possible finemapping and SNP genotyping. Genotyping service will be offered to Australian samples next. Hopefully there is more information on samples and timetables after Steering committee meeting 13<sup>th</sup> of June.

## Discussion on Quality Control issues

Uppsala SNP genotyping laboratory is aiming to get accreditation for their laboratory. Different platforms and equipments needs different quality control checks. Laboratories should be compared to each others. Tomas Axelsson has been in touch with Markku Keinänen. Most of the SNPs will be different between Helsinki-Uppsala.

Microsatellite labs should have same quality control checks. Sample mix-ups and plate problems can be checked only at FGC, because the plates are same for both labs. All markers will be cross-checked. POOL sample will be runned once.

Action points:

- One plate of Corell DNA and around 9 SNPs will be done in both laboratories to compare the methods. Tomas Axelsson will pick up the samples and Helsinki will choose the SNPs.
- Elisabeth brough primers (ch.1-11) to Uppsala for some project. Ceph control results of those markers will be used in this project for cross-checking and merging data.

## Databases and LIMS

Genotyping data of all laboratories will be transferred into Submission database, which is one step before transferring results into Genotype Database (GtDB). GtDB is harmonised database that will be used for analysis. There will be just information on binned alleles and bases for SNPs. All the laboratory data of results will be stored locally.

Local systems at the moment:

- FGC microsatellite SQL\*LIMS (AB)
- FGC SNP BioBase
- UU SNP Chiasma database (alpha testing at the moment)
- UU microsatellite GeneMapper software (AB)

Before transferring any genotype result data into GtDB, information on samples, persons and markers has to be preloaded. Checks will be done to genotypes in Submission database. Harmonizing of GtDB microsatellite data will be done before transferring new data into GtDB. GtDB development is done as open source project.

Genotyping laboratories will produce first information on marker maps, but those who will analyze the data should always check and update the marker map information.

GENOMEUTWIN

Genotyping Core Meeting

11.06.2003

**Memo**

Anne Leinonen

### **Selection of candidate genes and SNPs**

Andreas Dahlgren presented how the selection of SNPs was done with SHOX (short stature homeobox-containing) gene in Uppsala.

### **Report to Brussels**

Each partner of genomeutwin project will do own report by August. Ann-Christine Syvänen will put together all Uppsala's laboratories report.

### **Next Meeting**

Next Genotype Core meeting will be end of September.