

14.09.2004, Uppsala

Attendees

KLT: Minttu Jussila, Outi Repola, Kaisa Silander
 FGC: Aarno Palotie, Elisabeth Widén, Anne Leinonen, Juha Muilu
 UGC: Ann-Sofi Strand, Charlotte Johansson, Jenny Jonsson,
 Inger Jonasson
 Mol.Med U.U: Ann-Christin Syvänen, Tomas Axelsson, Andreas Dahlgren,
 Per Lundmark, Mats Jonsson

DNA extraction

Outi Repola and Minttu Repola reported about the status at KTL concerning delivered DNA samples from the different cohorts.

COHORT	Samples total @ KTL	Processed for genotyping	On plates	Sent to FGC	Sent to Uppsala	Sent for SNP-analysis	Not on plate, comment
DANISH LSDAT-97	335	325	Eutwin_P1-P3 Eutwin_P7	325	325	49	10, MZ
DANISH Geminikar	332	322	Eutwin_P4-P7	322	322	322	10 samples on P11
DANISH Geminikar-Hvidovre	261	0	0	0	0	0	Only in 29 samples enough DNA
DANISH Odense	400	398	Eutwin_P18- Eutwin_P22	372	372	396	P22 not filled, not ready for scanning
UK	238	231	Eutwin_P8 – Eutwin_P10	231	0	0	7, not enough DNA
FINNISH cohorts	1507	1507	Several plates with different names	1441	548	0	66, P11 not filled, not ready for scanning

DNA samples available at KTL for SNP genotyping

	Processed for genotyping	On plates	Sent to FGC	Sent to Uppsala	Sent for SNP-analysis	Not on plate, comment
DUTCH Trios	72	Eutwin_SNP_P1	0	0	72	3, not enough DNA
FINNISH Trios	185	Eutwin_SNP_P2 Eutwin_SNP_P3	0	0	185	

Samples from Australian twin cohort (n=750) expected to arrive before Christmas 2004.
 Additional samples expected in the future.
 5 plates with trios from Sweden will probably be extracted in Sweden.

Progress in Genotyping

FGC, Helsinki, (microsatellite)

Elisabeth Widén reported that in total 638 000 genotypes has been produced at FGC according to table.

Danish Twins			
Genotyped	7 plates	139 000 gt	Chr.1-11
In progress	4 plates		Chr. 1-11
Finnish twins			
Genotyped	10 plates	313 000 gt	Whole genome
Genotypes	6 plates	119 000 gt	Chr. 1-11
UK twins			
Genotyped	2 plates	67 000 gt	Whole genome
In progress	½ plate		Whole genome

UGC, Uppsala, (microsatellite)

Jenny Jonsson reported the status from Uppsala Genome Center

Danish twins			
Genotyped	7 plates	111 600 gt	Chr. 12-22, X
(30 markers still in progress from plate 4-7)			
Start progress	Oct-04	4 plates	Chr. 12-22, X
Finnish Twins			
Start progress	Oct-04	6 plates	Chr. 12-22, X

Jenny reported that one male sample from Danish plate 5 shows heterozygosity for 6 of 12 chr. X markers. It was decided that markers from chr. Y should be run on this samples in Uppsala and some more chr. X markers should be run at KTL for gender control of this sample.

Jenny reported that UGC have problems with low signal intensity for some markers. It was decided that UGC should use 8ng DNA per reaction instead of 5 ng as have been used for the plates processed up to now.

Helsinki, SNP

No twins typed in genomEUtwin. Status in genotypes produced in Morgam study was reported.

- Typed ~1000 FINRISK92 samples on 136 SNPs
Approximately 129000 successful genotypes*
- FINRISK92 genotype data in database:
80761 genotypes (average 94.9% success rate)
- Blind duplicate discrepancies:
10/3970 successful pairs
genotyping reproducibility of 99.72%

[* Data does not include 180 controls & 60 trios]

Uppsala, SNP

Andreas Dahlgren reported about progress in genotyping candidate genes for stature.

For genotyping the Short stature Homoeobox containing (*SHOX*) gene 25 in house validated SNP markers have been selected. Danish twins, 371 dizygotic controls, have been genotyped and 396 cases will be genotyped in the nearest weeks. Future plans are to genotype samples from Finland and to identify new candidate genes looking at QTLs from genome scans.

Per Lundmark reported from the migrain study, "haplotype/LDstudy of GRID2".

442 samples (trios) from Australia, Finland & Netherlands will be compared regarding LD patterns in CEPH samples from HapMap project.

Report from SNP / QC project

10 laboratories using different technology for SNP genotyping have been involved in a study comparing results between the labs. In May-04, 48 samples were sent out to the different laboratories to be genotyped with 18 SNP markers. Results were sent to Labtechnology who made the report.

The result showed 98-100% accuracy between the participating labs.

Database issues

Juha Miulu reported the status for the genotype database.

Currently 172 000 genotypes has been uploaded to the db. Nothing is deleted.

Juha gave an on line demonstration of the db.

Comments;

Wish to include line for SNP markers with possibility to make comments for markers deviating from Hardy Weinberg equilibrium.

RS numbers should be used in db for SNP markers.

Report from the Etichal Core meeting in Olso by Kaisa Silander

Data Security issues

Action item #1: Have a committee that will make recommendations (to be approved by the Steering Group) on the following topics:

- Guidelines for short-term data sharing (e.g. encryption)
- Access rights to data at present (no common database), and access rights to the federated database
- Guidelines for storing combined genotype & phenotype data sets, e.g. for analysis and following a publication
- Backup of data produced during project

Agreements between participants

Action item #2: Ethics Core, with help and advice from MORGAM, will develop "material and data transfer agreement" documents that will specify ownership rights to samples and data, sample transfer and storage procedures, etc.

These agreements should be in compliance with participating countries' existing laws.

Q: Do we need "Intellectual property rights" agreements?

DNA Sample issues

Action item #3: A system for tracking of DNA samples received and sent could be developed by DNA & Genotyping Core

Action item #4: Streamline protocols for DNA isolation, aliquoting, quality control, storage, etc. between KTL DNA Core and other EUtwin participants, e.g. the Swedish Biobank.

End of meeting