

Document

Instructions to send samples and information for genotyping.

Version

1.6.3

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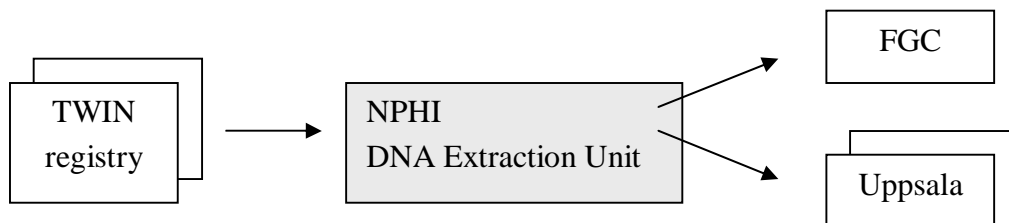
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1. General Information

The genotyping core operates at two geographical sites, one in Helsinki at the Biomedicum Helsinki, and one at Uppsala University. The tasks of the genotyping core are to extract DNA from blood samples and to perform the genotyping work in the laboratory. DNA is extracted using the Genra Puregene system. Both multiallelic (microsatellites) and biallelic (single nucleotide polymorphism, SNP) markers are analyzed using state-of-the-art high-throughput technology. Assigned genotypes are delivered from the genotyping core to a joint database for further statistical analysis.

http://www.genomeutwin.helsinki.fi/research_core_genotyping.htm

The samples will be received by Finnish NPHI's DNA Extraction Unit.



2. Sample Information needed DATA FORM 1

Transfer of DATA FORM 1 should be completed before transfer of samples.

Fields or columns can be left empty if the data is not available. DATA FORM 1 should be sent in excel format. The Centers sending DNA samples are to fill the DNA sample sheet. Centers sending blood samples are to fill the Blood sample sheet.

Please note, the questions related to the shipment. The DATA FORM 1 can be sent to Mrs. Outi Törnwall (outi.tornwall@ktl.fi).

DATA FORM 1

DNA sample sheet

Number information		Original sample information				
EUid number	CsampleID	Sample type before extraction	Anticoagulant (if whole blood)	Date of sample drawing	Date of freezing	Date of extraction
360026200113	1354	Whole blood	EDTA	01.12.2004	02.12.2004	6.1.2005
360026200225	5551	Whole blood	ACD	01.02.2005		1.2.2005
360026250091	2648	Buffy coat		03.11.2003	03.11.2003	5.6.2004

Please, include answers to these questions when sending the data to National Public Health Institute

Questions:

1. What number is in the DNA tube?
2. Are the samples bar coded or labelled some other way?
3. By which method is the DNA concentration measured?
4. Are the samples sent in tubes or plates?
5. How many samples are to be sent?

DNA information								
Extraction method	Concentration (ng/μl)	Volume (μl)	Type of hydration buffer	Storage temperature (C)	Gender	Indication of the Duplicate sample	Name of the Box/ plate	Location of the tube in the box/plate
Salt precipitation	135,00	100,00	TE	-20	1		Box 1	1
Phenol-Chloroform	50,00	260,00	dH2O	-80	1	DUP 1	Box 1	2
Gentra Kit	300,00	40,00	TE	-20	2		Box 2	1

Blood sample sheet

Number information		Blood information						
EUid number	CsampleID	Sample type	Anticoagulant (if whole blood)	Sample amount (ml)	Date of sample drawing	Date of freezing	Storage temperature (C)	Gender
36002620012	153456	Whole blood	EDTA	10,00	01.01.2005	01.01.2005	-20	1
36002620023	321548	Whole blood	EDTA	7,00	02.02.2002	03.02.2002	-20	2
36002625010	179513	Buffy coat		2,00	05.05.2005	05.05.2005	-20	1

Please, include answers to these questions when sending the data to National Public Health Institute

Questions:

1. What number is in the Blood tube?
2. Are the samples bar coded or labelled some other way?
3. What kind of tubes are used for the blood (plastic or glas)?
4. How many samples are to be sent?

Explanation of fields

EU id number	Eu code created for each sample (12 digits!)
CsampleID	Code used in the center/biobank
Sample type before extraction	The type of the sample before extraction
Anticoagulant	The name of the anticoagulant
Date of sample drawing/ freezing / extraction	DD.MM.YYYY
Extraction method	The method used for extraction of DNA
Concentration	ng/µl value of the measured concentration of DNA in the tube
Volume	Volume of the DNA sample in the tube
Type of hydration buffer	The name of the hydration buffer for the DNA
Storage temperature	Storage temperature of the DNA (Celcius)
Gender	1= Male, 2= Female This information is only used for QC purposes
Sample amount	The amount of blood in the tube (ml)

3. Duplicate Samples (DNA samples only)

Every shipment should contain one duplicate DNA sample per 92 samples.

Duplicates should be named as **DUP.**[1,2,3...]

e.g. for number of 276 samples there should be DUP.1, DUP.2, DUP.3

4. Person Information needed DATA FORM 2

Following information is needed to detect genotyping errors. If also parents or other siblings of twins will be genotyped, pedigree information is needed. Please, send the Data Form 2 information to Data Managers of genotyping centers (see contact information). Data should be encrypted if sending via email.

DATA FORM 2

Column	Description	Type	Values	Required
EUIDNUM	EUid number	NUMBER		X
CSAMPLEID	Sample ID in center/biopank	TEXT		X
GENDER	Sex of person	NUMBER	1 = male 2 = female	X
BIRTHDATE ^{*)}	Date of birth	DATE	(YYYYMMDD)	
FAMILY ID	Family identification	TEXT		
FID ^{#)}	Id of father	TEXT		
MID ^{#)}	Id of mother	TEXT		
ZYG	Zygoty	NUMBER	1 = MZ 2 = DZ 9 =unknown	only for twins

^{*)} If the **Date of Birth** information is incomplete, you can use one of the following ways

^{#)} Use EUid numbers, if parents have any phenotypic data available.

1) As in GenomEUtwin's Phenotype Database Prototype v. 3.2.2.

BIRTHDATE	Date of birth	Date	8	YYYYMMDD - Year, month, day YYYYMM15 - Day unknown YYYY0701 - Day and month unknown NULL/blank - Irrelevant/non-participant/non-response, Irrelevant/structural, Do not know, Unknown
BIRTHDATE_ ERROR	Error code for Date of birth	Number	1	1 - Date is complete 2 - Day is missing, coded 15 3 - Day and month are missing, coded 0701 6 - Irrelevant/non-participant/non-response 7 - Irrelevant, structural 8 - Do not know 9 - Unknown

2) Two separate fields.

BMONTH	Birth month	TEXT	NULL /month is missing
BYEAR	Birth year	TEXT	

5. Shipment

The aliquotted DNA or blood samples should be send via courier to:

National Public Health Institute
Dept. of Molecular Medicine
Mrs. Arja Terola
Biomedicum
Haartmaninkatu 8
00290 HELSINKI
FINLAND

The samples should be packed into a clean cardboard or plastic secondary container preferably in the same order than in Data Form 1. Print of samplelist should be included.

Shipping document should include at least following information: description of the goods, statement of the value of the goods (emphasis on only scientific value). Required items depends on country. Please, check needed information, if you are shipping outside the EU. Consult your local courier about shipping human derived material and legislation of your country.

The shipping should take place in the beginning of the week. If the samples will arrive during the weekend, this has to be agreed with Mrs Outi Törnwall

Please, notify Mrs Outi Törnwall (outi.tornwall@ktl.fi) via email, when the samples have left your facility.

Include information on:

- tracking number for the courier
- number of samples sent
- name and contact information of the sender
- estimated time of arrival
- sample list file

Return receipt will be sent by receiver.

5.1 Shipment of DNA

The amount of DNA to transfer should be agreed with GenomEUtwin steering group, genotyping lab and participating center. The agreed amount is aliquotted into a new tube in the center.

Suitable type of tubes

- Sarstedt (cat. 72.694.100) 2.0 ml screw cap tube
- Eppendorf (eg. Micro Test Tube 3810) 1.5 ml flat lid tube

Screw cap tube can be shipped as it is, but standard Eppendorf tube must be individually sealed with Parafilm or equivalent. NPHI DNA Extraction Unit can send you clean unsterilized 2.0 ml tubes on your request.

Identification code should be placed with bar code to the side of the tube. Teemu Perheentupa (teemu.perheentupa@ktl.fi) can give information of available barcode types. Also numeric code should appear on the tube. Please, use the Sample ID of your center/biopank in barcode if you are uncertain with EUtwin numbers (EUIDNUM). (Please see figure 1. for instructions of how to place the label.)

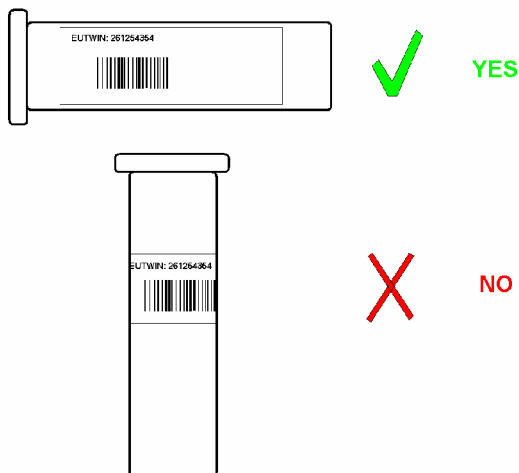


Figure 1. Orientation of identification label on DNA tube.

5.2 Shipment of blood samples

ALL SAMPLES MUST REMAIN FROZEN DURING SHIPMENT

A frozen blood sample may not be thawed for aliquotting. DNA extraction will not produce enough DNA of required quality and quantity if the sample has been repeatedly frozen and thawed.

A thawed blood sample must be extracted within a day.

Heavy contact between samples should be avoided either by desing of the box or by padding each sample. Breakage of the tubes should be avoided by any cost (if the tubes are glass).

The boxes should be packed into a styrofoam boxes and ample amount of dry ice should be included.

Consult your local courier about the insulation properties of the box you are about to use and about the amount of ice to be included. Biotransported Cargo - Airseecontainers - is recommended

The temperature of the shipment should remain under -10°C during transportation.

6. Contact information

NPHI DNA Extraction and Sample Handling (Finland)

<p>Outi Törnwall DNA co-ordinator, sample data management</p> <p>Tel: +358 – 9-4744 8265 E-mail: outi.tornwall@ktl.fi</p>	<p>Minttu Jussila DNA sample management</p> <p>Tel: +358 -9-4744 8277 E-mail: minttu.jussila@ktl.fi</p>
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Finnish Genome Center - microsatellite genotyping (Finland)

<p>Prof. Aarno Palotie Director</p> <p>Tel: +358 -9-191 25470 E-mail: aarno.palotie@helsinki.fi</p>	<p>Elisabeth Widén Associate Director (genotyping)</p> <p>Tel: +358 -9-191 25477 E-mail: elisabeth.widen@helsinki.fi</p>
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Uppsala Univeristy - SNP genotyping (Sweden)

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