

Estonian Genome Project

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Estonian Genome Project Foundation

**EU – Canada Workshop on Public Population
Project in Genetics - P³G**

14-15 December 2003

Manchester, UK



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What do we want to use the BioBank for?

- Gene discovery in complex diseases
- The role of the environment in complex diseases
- Pharmacogenetics for the existing drugs
- Promote gene- and biotechnology, bio- and medical informatics, international scientific collaboration and corporate development in Estonia



Estonian Gene Bank timeline

Genome Center Foundation 1999

Human Genome Research Act 2000

**Estonian Genome Project Foundation → EGeen Ltd.
EGeen Inc 2001**

Pilot Project September 2002 – March 2003

Main Project April 2003 -

Close to 9 000 individuals in the database

Genotyping First research projects will start in 2004

Funding will be shifted more towards public sources in order to have more long term stability from 2004



Scientific research

- Free access for Estonian Scientific and Research Institutes and together with Estonian academic researchers.
- Approved by Ethics Committee of EGPF and IRB
- Only anonymous records (not less than 5)
- IP rights will be shared with EGPF
- As a rule the DNA samples have to stay in ESTONIA



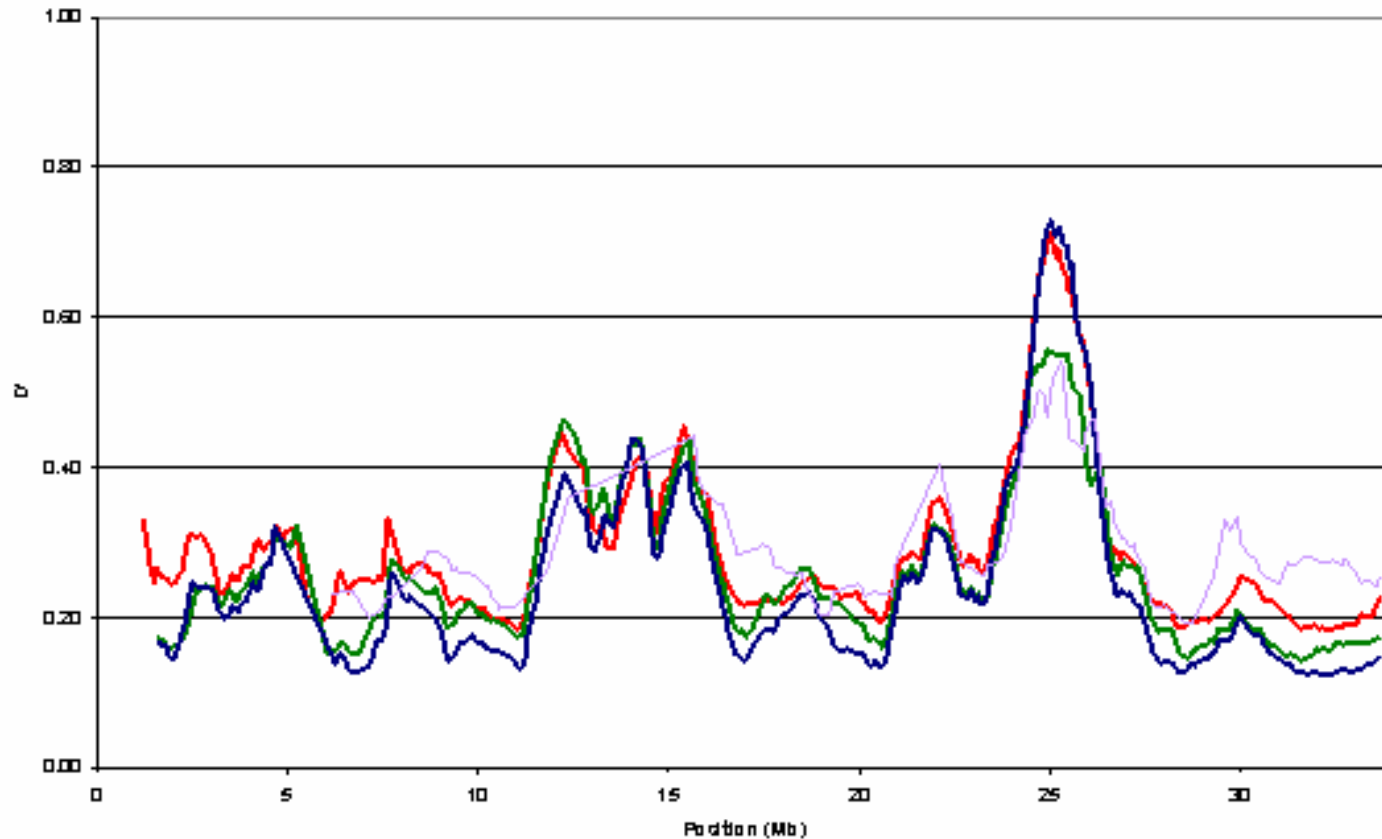
Is Estonia a good place for a biobank?

- Representative European population
- Statistically relevant sample base – 1.35M
- Proper legal and ethical structure
- Universal centralized healthcare
- Government and popular support
- Affordable logistics, good infrastructure



LD across chromosome 22

(CEPH families. Porton Down. Estonian and combined samples)



Dawson et al. (2002) Nature 418: 544



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Legal and ethical framework

HGRA regulates

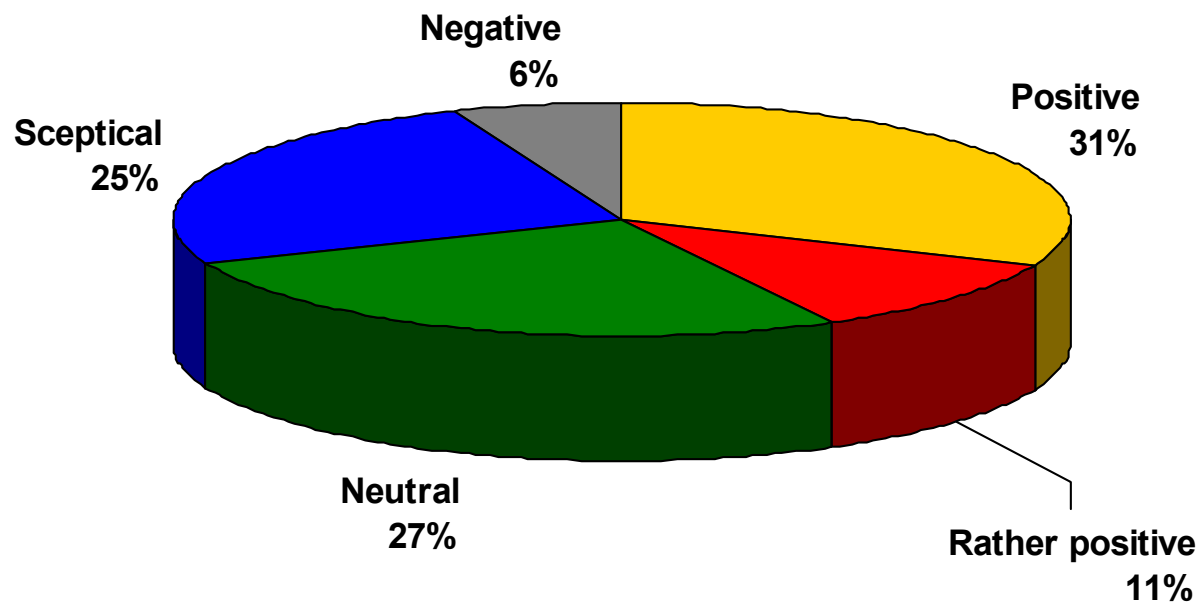
- scientific research on human genetics
- establishment and maintenance of the GeneBank
- supervision of ethical issues (Ethics Committee)
- access to data and use of genetic information

HGRA protects

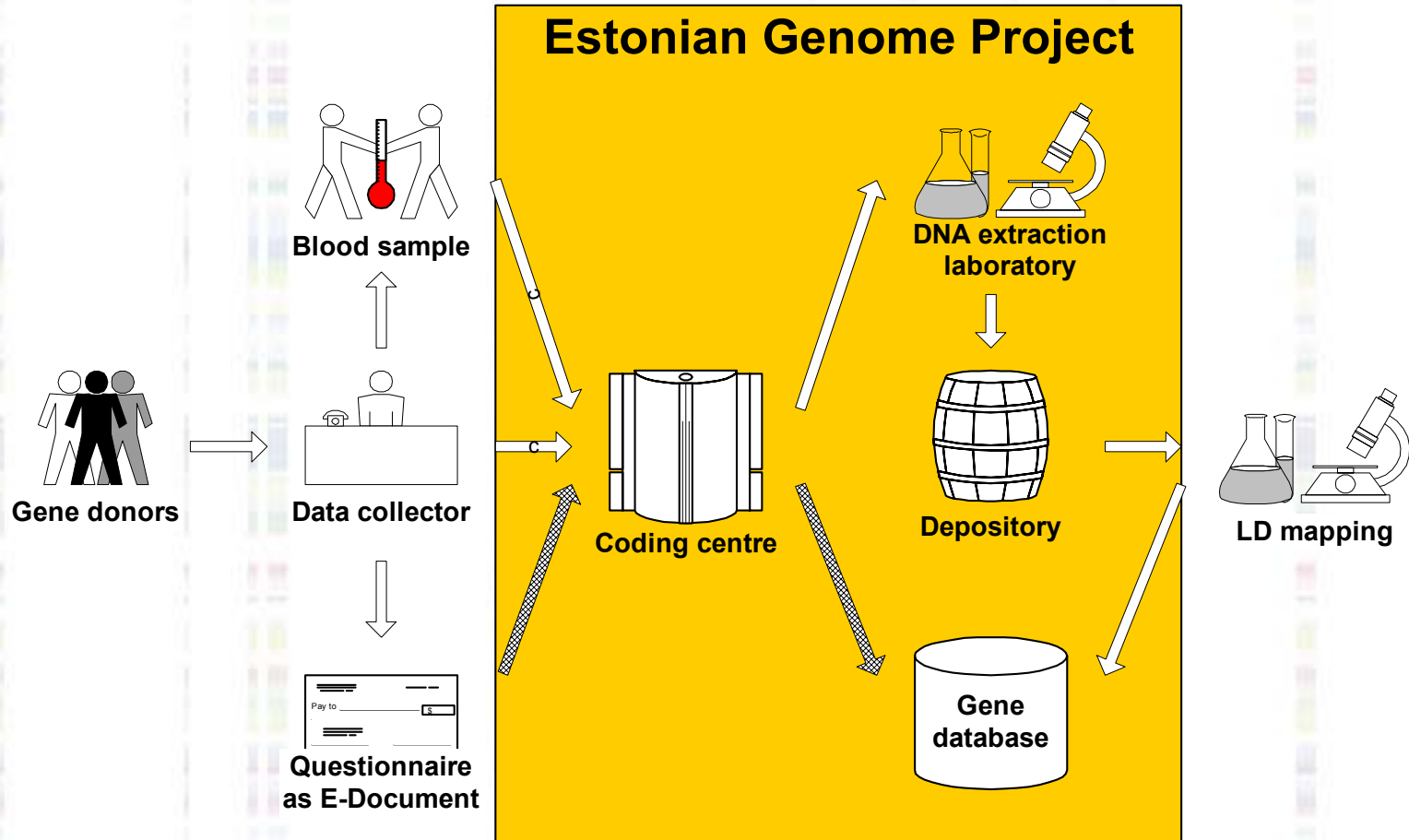
- person from obligation and influence to participate
- confidentiality of gene donor
- public from the misuse of genetic information
- gene donor from genetic discrimination



Attitudes of Estonian Media (2001-2002)



Operations of the EGP

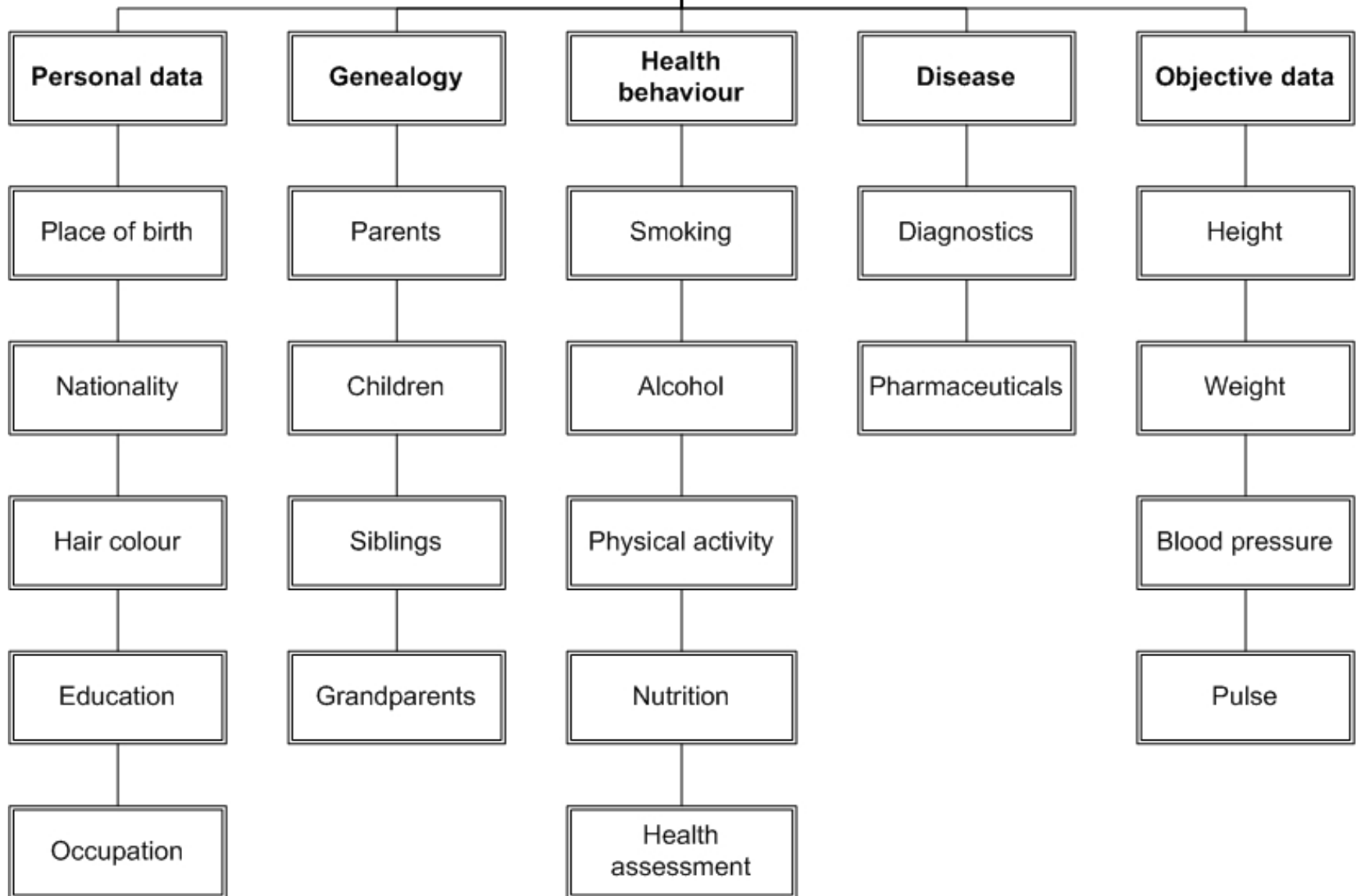


Phenotypic data

- **Digital records** – personal data, environment, behaviour, health, treatment, genealogical data (3 generations)
- **Data collection** - interview + existent health records
- **Data monitoring according to GCP** - verification, health registries, hospitals databases
- **Dynamic data updating**



EGFQ



21.veebruar.2003

11:48:01

Isiku otsimine isikukoodi järgi

Geenidoonorite hulgast:

 Otsi

Perearsti süsteemist:

 Otsi

Isikuandmed perearsti süsteemis

Isikukood

Eesnimi

Perekonnanimi

Sünniaeg

Sugu

Pikkus

Kaal

Elukoha kood

Address

[Lisa geenidoonoriks =>](#)

Geenidoonori andmed







Isikukood Eesnimi Perekonnanimi Sünniaeg Sugu Pikkus Kaal

Elukoht

Maakond Vald Elukoha kood

Address

Nõusoleku aeg

Transpordikood  Nõusoleku trükieelvaade... Trüki nõusolek Koeproov... Küsimustik...  Lisa Kustuta Muuda Salvesta Iühista

Ischemic stroke

- ICD-10
- Describes the pathogenesis, localisation of the vessels (emboli, thrombosis)
- Used in everyday clinical practice
- I63.0-I63.9 ischemic stroke
- I61.0-I61.9 hemorrhagic stroke
- TOAST
- Describes the etiology of the ischemic stroke
- Used only in scientific work
- Subcategories:
 - Large vessels (the criteria is 90% stenosis)
 - small vessels
 - Cardiogenic
 - Others (coagulopathies etc)
 - The reason is not identified



Harmonisation of ICD-10 and TOAST

ICD-10 I.63.0-I.63.9



TOAST

- Large vessels
- Small vessels
- Cardiogenic



Key to the success of the EGP

- General education of the population (and physicians!)
⇒
(TV, radio, newspapers, presentations, booklets, Web, hot line)
- Careful preparatory work - **IT solutions**
- Advances in **population genetics** (e.g.HapMap)
- Transparency of management and **financing**
- **Quality** of data - QMS ISO 9001:2000
(health data questionnaire, training of data collectors, monitoring, operative trouble-shooting, qualified staff)
- NO hype.
- **Funding!**

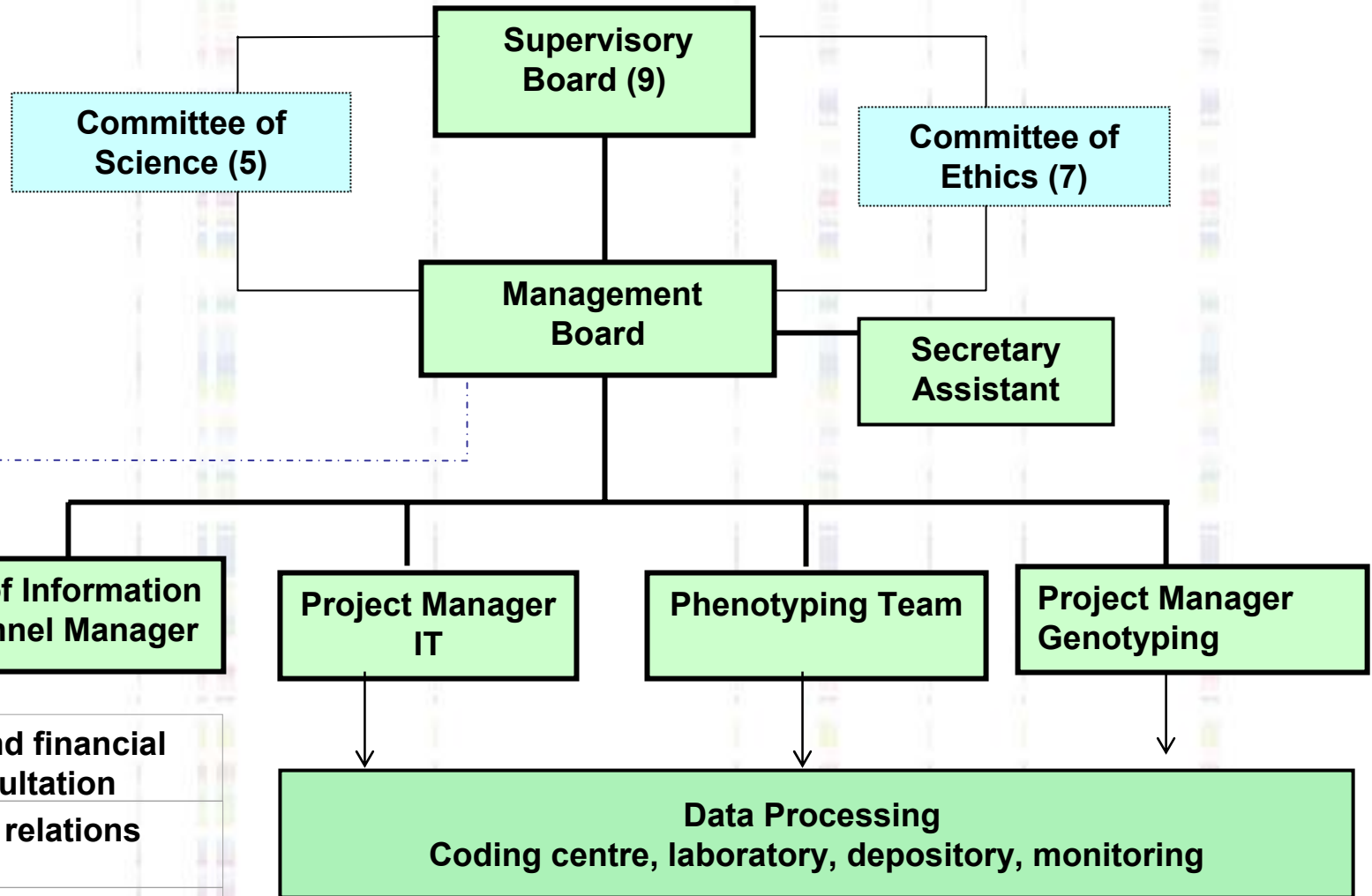


Problems?

- Funding - academia and industry have different expectations
- Infrastructure for the next generation of scientists (hard to convince funding agencies)
- Power - to increase, collaboration between biobanks e.g. P3G is needed
- Management of the public –private project (risk e.g. UmanGenomics)



Structure of the EGP Foundation

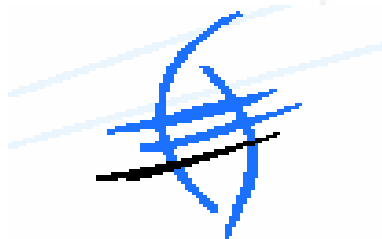


www.geenivaramu.ee



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www.egeeninc.com



€ **EGeen** ct