Biobanks and Genomic Resources in Europe 2007-2013

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Background

- Genetics has the potential to enable effective personalised medicine
- There is a rapid emergence of new methodologies to study genetic variation
- These must be applied in conjunction with well-designed studies of large population cohorts (disease and health)

- Europe has the best cohorts and the health care systems for these studies worldwide
- Europe has strong traditions in human genetics, epidemiology and other skills

Europe can be leader in personalised medicine

Conditions:

Coordinate existing genetic-epidemiological collections and establishing and expanding the new biobanks, particularly in Central and Eastern Europe, to determine and validate the principal genetic and environmental determinants of major diseases

Identifying the principal genetic determinants of major disease

TCGAGATCGTCGATTCGGA
TCGAGATCGCCGATTCGGA
Single Nucleotide Polymorphism SNP

People have different genome sequences

In consequence they:

a. Have susceptibility to different diseases
b. Have different complications of their disease
c. Respond differently to treatment
d. React differently to different environment

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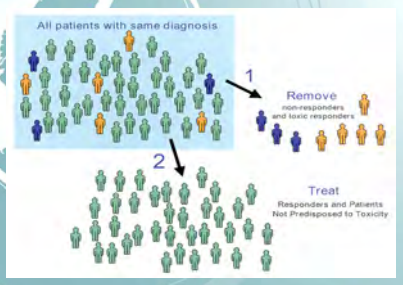
Leading to:

a. New taxonomy of human disease
b. Diagnostics
c. Parmacogenomics
d. Genetic epidemiology



From McLeod and Evans, Ann Rev of Pharmacol and Toxicol, 2001: 41,101-121

National spending (some examples)

▪ Spain	8 M € /year
▪ UK (case/control study)	5 M € /year
▪ Scandinavia platforms	2 M € /year
▪ Estonia (biobank and genomics infrastructure)	4 M €/2005
▪ Germany	10 M€/year
▪ France	7 M €/year

Present situation

- National efforts, IP and NoEs have prepared or started the biobank collections that has to be expanded and demand an analysis infrastructure; ethical engagement to explore these optimally
- National fragmentation and lack of sustainability for both genotyping infrastructure and samples
- Even major efforts are underpowered; difficulty of scientific and translational follow-up
- Access not available throughout the EU

Identifying the principal genetic determinants of major disease we need :

- Powerful study designs
- Large biobanks with DNA collections and good information about health, disease and other variables
- Other biological data (transcripts, proteins etc.)
- Even better genotyping technologies

How to support projects that require access to large-scale genotyping with rapidly shifting technologies?

<u>Existing instruments are not sufficient to achieve:</u>

- Harmonisation of procedures
- Standard approach to ethics and legal aspects
- Powerful studies combined with economies of scale
- Prioritisation
- Interpretation of results (statistical genetics)
- Integrated approach to bioinformatics and databases
- Access to data and cohorts