

# **Integration of Genomics into Public Policies and Health Services – European Perspectives and Initiatives**

Public Health Genomics

## **Public Health Genomics (PHG)**

**“Public Health Genomics (PHG) is the responsible and effective translation of genome-based knowledge and technologies into public policy and health services for the benefit of population health.”**

**(Bellagio Statement 2005: GRAPH/nt, PHGEN)**

## The Enterprise

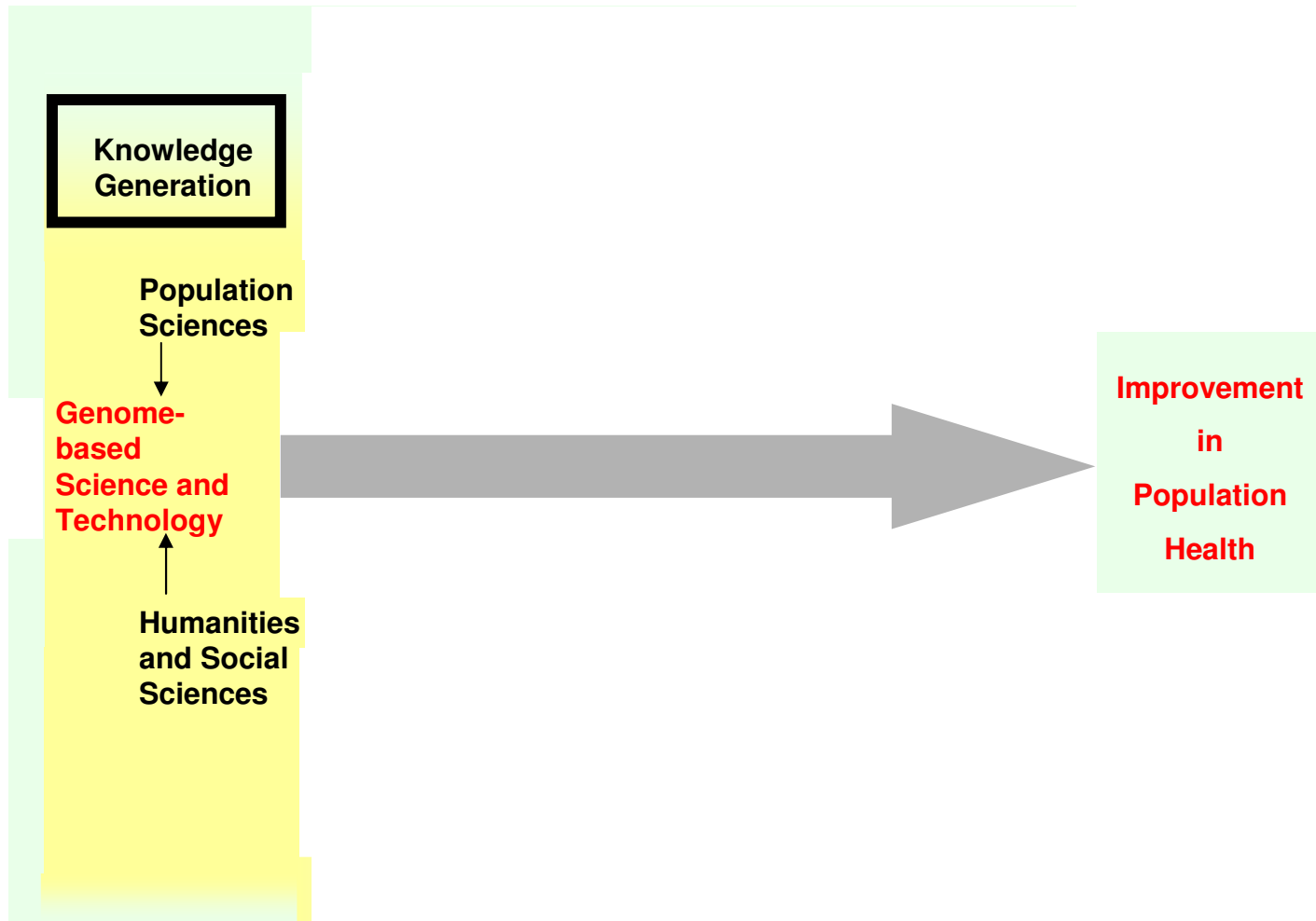
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**Genome-  
based  
Science and  
Technology**

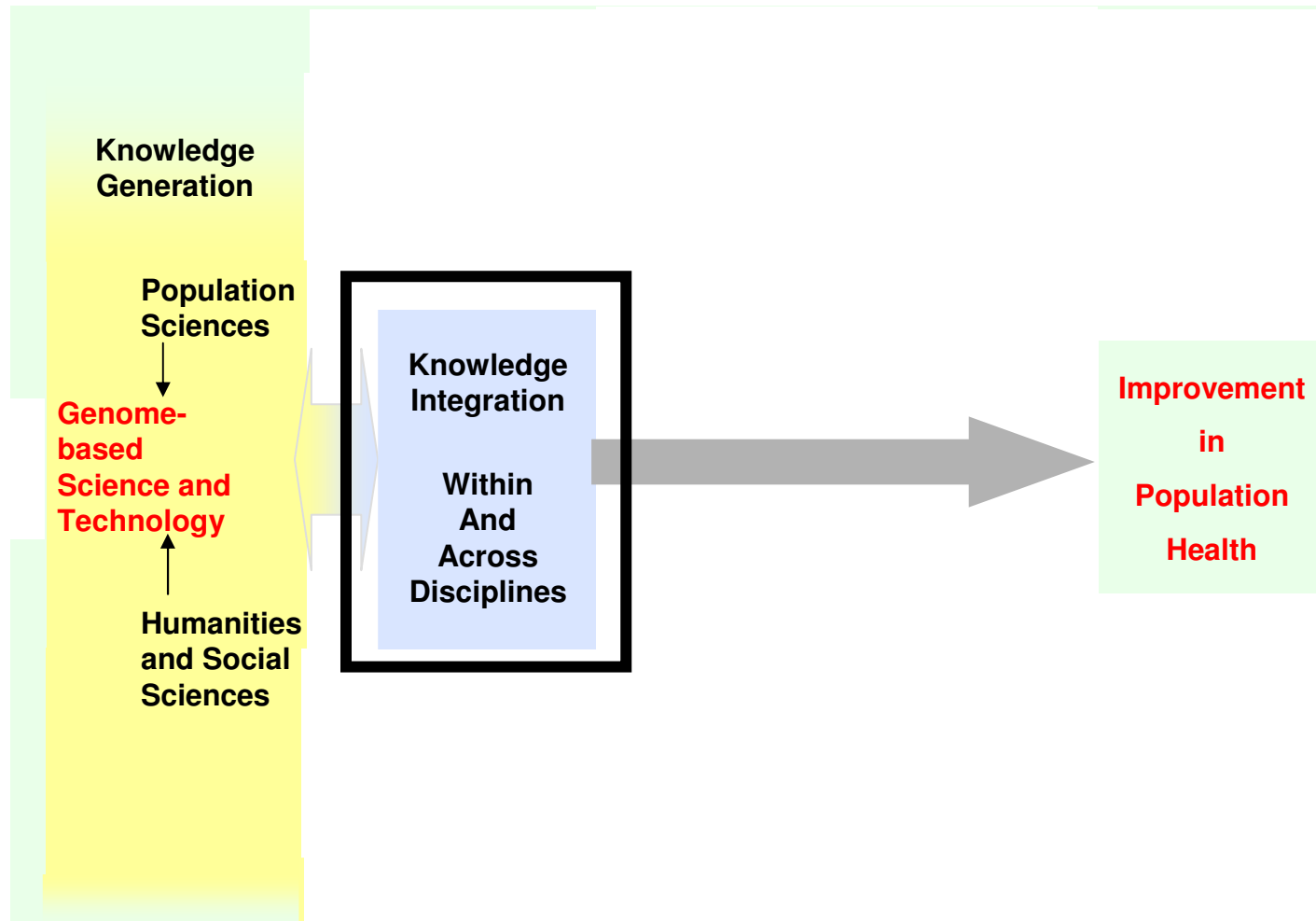


**Improvement  
in  
Population  
Health**

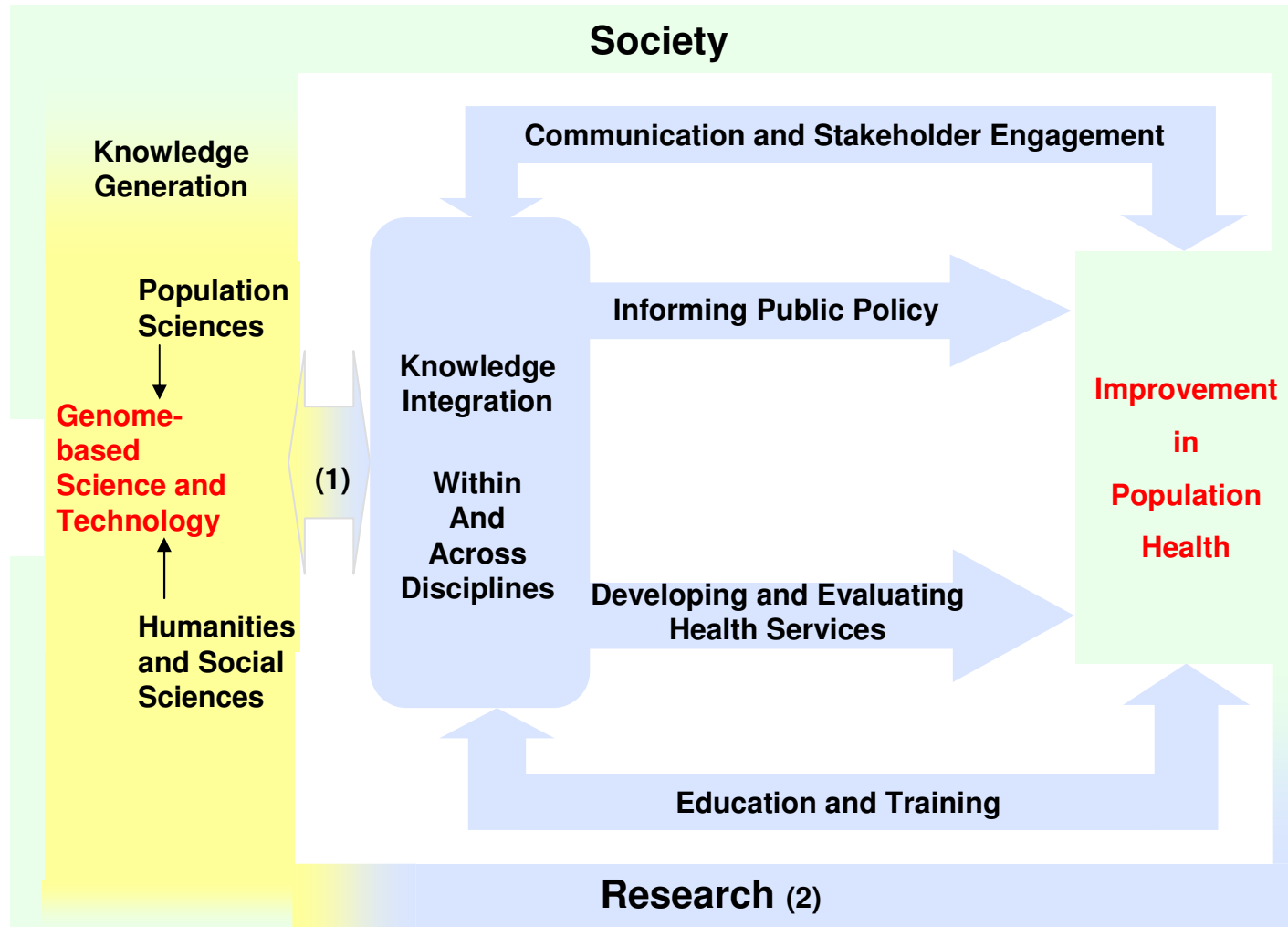
# The Enterprise



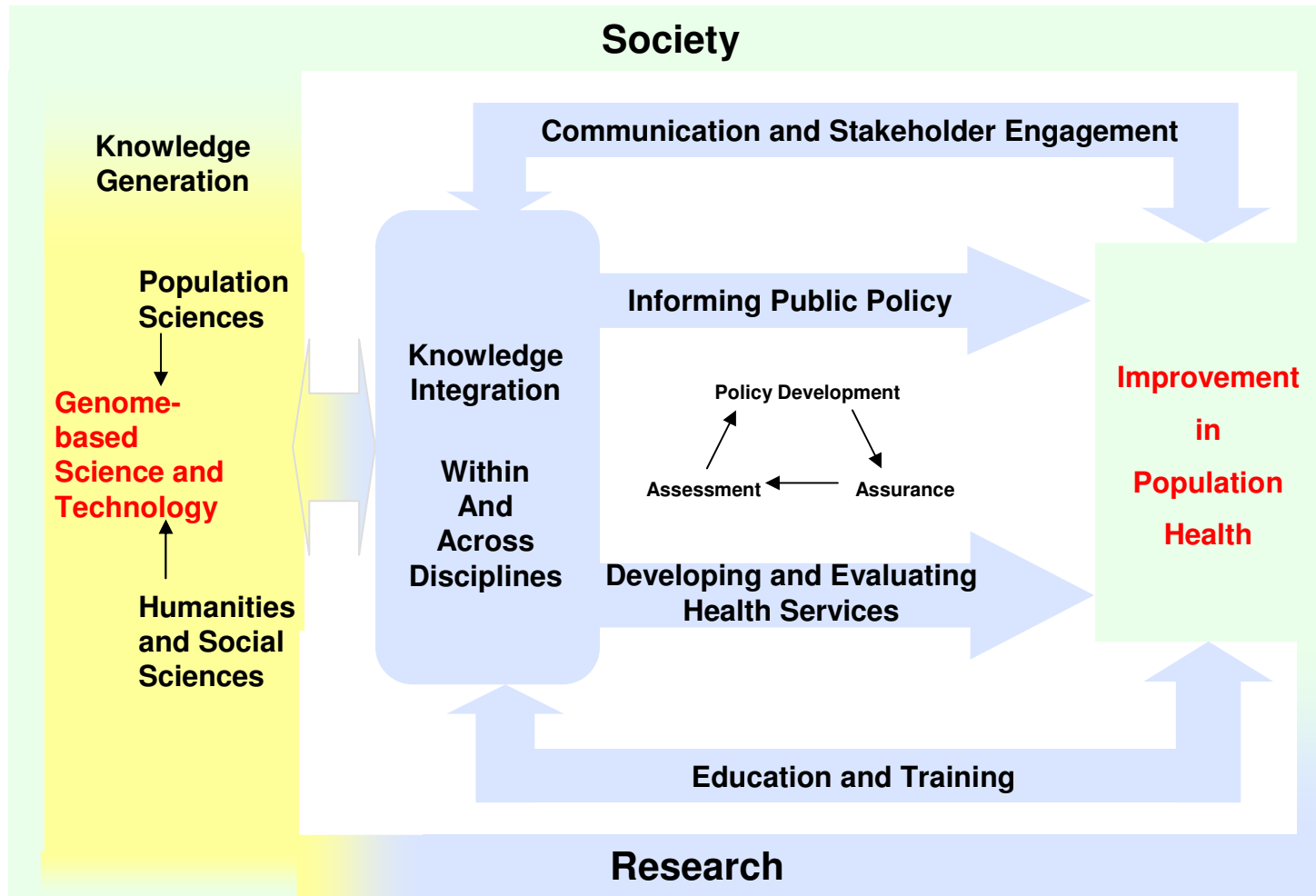
# The Enterprise



# The Enterprise



# The Enterprise



## **Public Health Genomics (PHG) - no revolution, but evolution!**

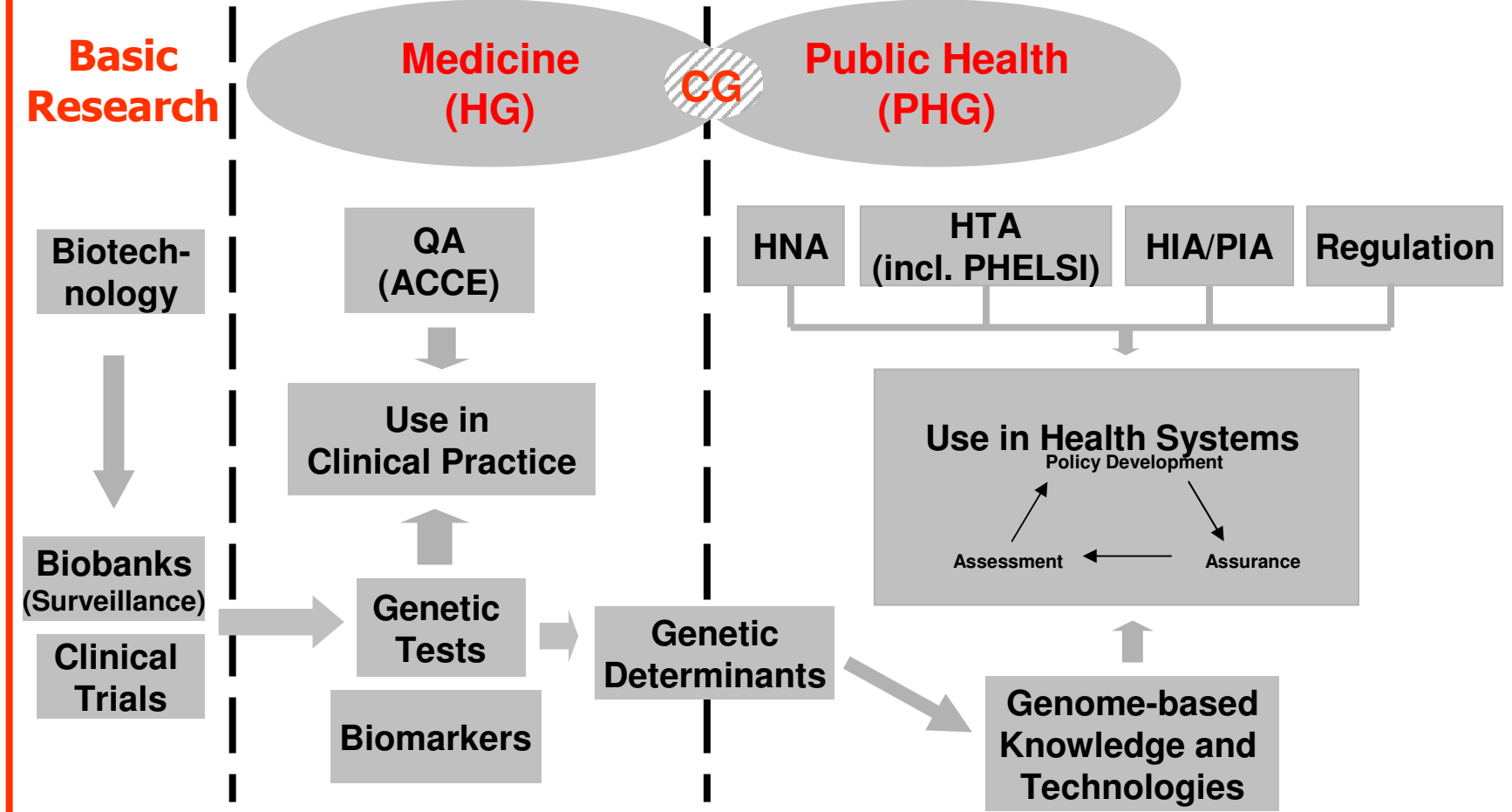
**“One specific task of PHG is to reconsider and systematically evaluate every condition of public health interest...” , e.g.**

- **evidence-based (stratified) prevention**
- **balance between self-responsibility and social welfare**
- **reduction of (genetic) inequalities in health**
- **health protection (e.g., recombinant vaccines, bioremediation, envirogenomics)**
- **consumer protection (e.g., nutrigenomics)**
- **health management (e.g., stakeholder responsibilities, public policy, regulation, risk communication, public participation, professional education and training)**

**[Brand A.: Public health and genetics – a dangerous combination? EJPH 15(2):113-116, 2005]**



**Human Genetics (HG), Community Genetics (CG) and Public Health Genomics (PHG) have complementary tasks!**



## The call



The European Commission called for a “networking exercise ... to lead to an inventory report on genetic determinants relevant to public health. This network will identify public health issues linked to current national practices in applying genetic testing and on that basis will contribute to developing best practice in applying genetic testing.”

Community action in the field of public health (2003-2008), Work plan 2005

**PHGEN**

**Public Health Genomics  
European Network**

**January 1st 2006 – December 31st 2008 (36 months)**

**EU Project: 2005313**

## **Aims of PHGEN (1)**

- **To conduct a networking exercise on PHG covering all EU Member States, Applicant Countries, and EFTA-EEA Countries**
- **To identify and list key experts and institutions relevant to PHG in these countries**
- **To provide an inventory of genetic determinants relevant to public health**
- **To provide an inventory of PHG-issues and priorities in Europe**

## **Aims of PHGEN (2)**

- **To identify legal diversities and barriers in a cross-border market**
- **To analyse the relevance of EU treaties for PHG**
- **To contribute to the co-operation and exchange of information in order to enhance coherence and disseminate best practice in Europe**
- **To promote and stimulate the countries' efforts in this emerging field by developing PHGEN and by supporting effective networking in order to reach sustainability (e.g. national task forces on PHG)**

## **Aims of PHGEN (3)**

**„In the long run  
PHGEN will serve the European Commission  
as an ‚early detection unit‘  
for horizon scanning, fact finding, and monitoring  
of the integration of genome-based knowledge  
and technologies into public health.“**

## **Main Partner & Associated Partners**

**Institute of Public Health  
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Public Health Genomics (DZPHG)**  
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**Public Health Genetics Unit (PHGU)**  
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## Steering Group

- Angela Brand, German Center for Public Health Genomics, Bielefeld, Germany (Speaker)
- Hilary Burton & Ron Zimmern, Public Health Genetics Unit, Cambridge, UK
- Helmut Brand, Institute of Public Health NRW (Iögd), Bielefeld, Germany
- Jean-Jacques Cassiman, Center for Human Genetics, Catholic University Leuven, Belgium
- Bartha Knoppers, Law Faculty, University of Montreal, Canada
- Marcella Rietschel, Central Institute of Mental Health, Mannheim, Germany
- Jörg T. Epplen, Department of Human Genetics, Ruhr-University Bochum, Germany
- Peter Dabrock, Department of Social Ethics, Philipps University Marburg, Germany
- Finn Kristensen, EUnetHTA, Danish Centre for Evaluation and HTA, Copenhagen, Denmark
- Andrej Marusic, Institute of Public Health, Ljubljana, Slovenia
- Dolores Ibarreta, Institute for Prospective Technological Studies (IPTS), Sevilla, Spain
- Ingolf Pernice, Walter Hallstein Institute for European Constitutional Law, Berlin, Germany



## **Collaborating Partners, Networks, Organizations & Experts**

### **Collaborating Partners from all EU Member States, Applicant Countries, EFTA-EEA Countries:**

public health, human genetics, competent authorities

### **Representatives of EU Networks:**

e.g. EuroGentest, EUnetHTA, Orphanet, NuGo

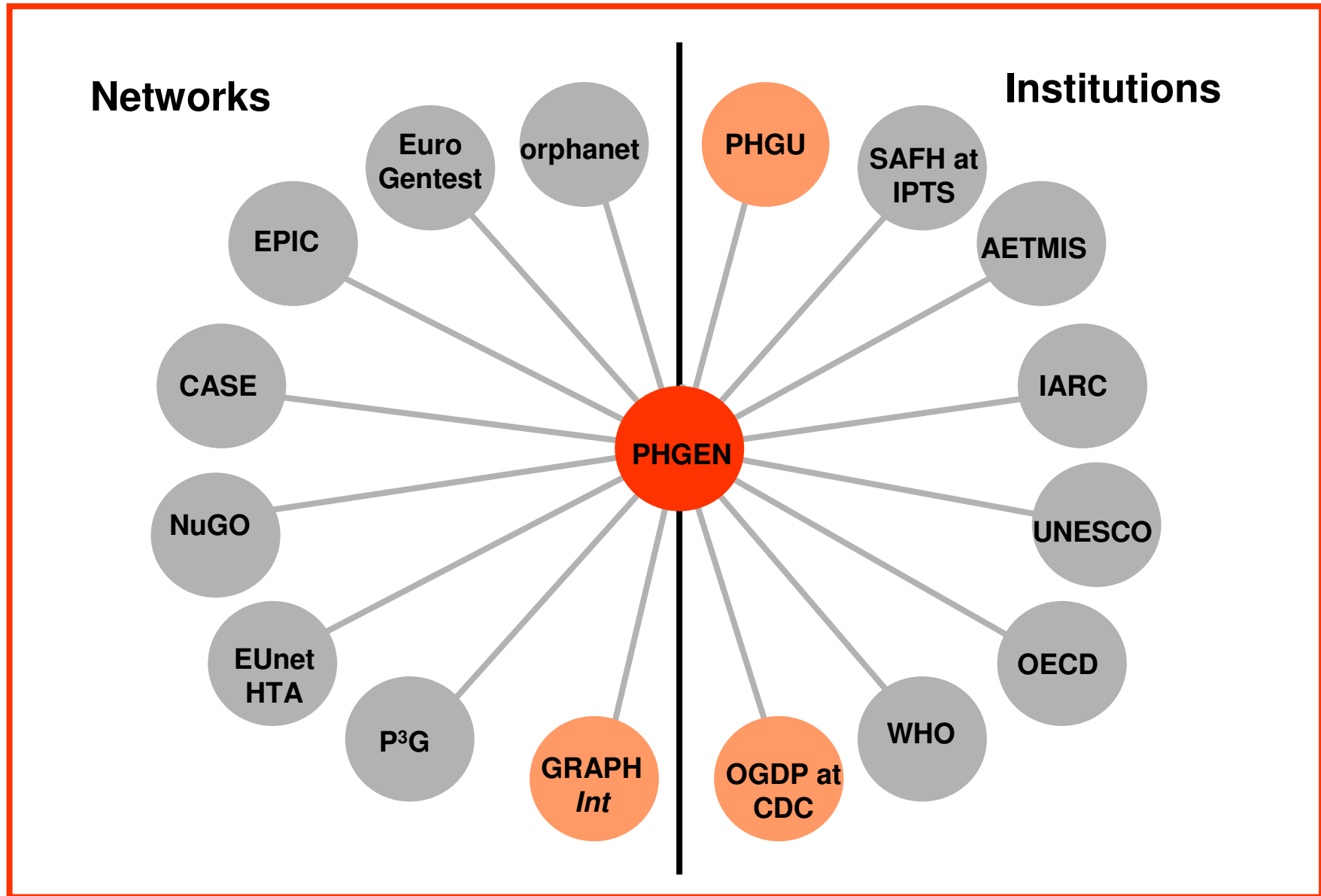
### **Representatives of Organizations:**

e.g. WHO, WTO, OECD, UNESCO, STOA

### **Experts on European Law**

### **Experts from outside EU:**

e.g. CDC, AETMIS, P<sup>3</sup>G



Public Health Genomics European Network (PHGEN) –  
[www.phgen.nrw.de](http://www.phgen.nrw.de)

## Roadmap: 10 Work Packages

1. Project Coordination
2. Evaluation
3. Dissemination
4. Network Meetings
5. Steering Group Meetings
- 6. Assessment**
- 7. Policy Development**
- 8. Assurance**
9. Final Conference
10. Final Report

## **Assessment (WP6)**

**Systematic collection, assembly & analysis of genome-based information relevant to Public Health**

- **Analysis of PHG concepts (e.g. definition of PHG, genetic determinants, risk stratification)**
- **Identification of PHG issues & priorities**
- **Identification and „best practice“ of PH methods relevant to PHG (e.g. HNA, HTA, HIA/PIA)**
- **Identification of networks and institutions relevant to PHG**

**→ Assessment Paper**

## **Policy Development (WP7)**

**Developing European standards and guidelines which promote the responsible and effective use of genome-based information and technologies in European health systems**

- **Analysis of legal diversities (e.g. conflicting laws) and barriers in a cross-border market**
- **Analysis of EU treaties for PHG**
- **Analysis of European minimal standards, guidelines & laws**
- **Analysis of economic implications & PHELSI**
- **Development of policies on education, information and empowerment**

**→ Policy Development Paper**

## **Assurance (WP8)**

### **Appropriate use of genome-based information and technologies in European health services**

- **Need for enforcement of new laws and/or regulations?**
- **Assurance of stakeholders' responsibilities in the application of genome-based information and technologies**
- **Assurance of a competent workforce**
- **Evaluation of health services (e.g. health promotion, disease prevention, therapy, rehabilitation)**

**→ Assurance Paper**

## **PHGEN, EGAN & Eastern European Countries: challenges I/II**

- Involvement of EGAN in PHGEN II?
- Involvement of representatives of EECs as CPs in PHGEN since 1/2006
- Development of European policies supporting national policies:  
eg. patient rights in biobanks
- Implementation of responsible, effective and efficient health services:  
from a prospective perspective in EECs  
versus  
from a retrospective perspective in Western European Countries (WECs)  
(innovation versus imitation)

## **PHGEN, EGAN & Eastern European Countries: challenges II/II**

- **Use of already existing strong infrastructures in EECs:  
e.g. Institutes of Public Health (IPHs)**
- **Implementation of EU regulations  
barriers regarding „Tourism on genome-based health services“ from  
WECs to EECs**



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**PHGEN is funded by the European Union**  
Duration period: January 1st, 2006 - December 31st, 2008, EU Project No 2005313

**Steering Group:**

German Center for Public Health Genomics,  
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**PHGEN**

**Public Health Genomics  
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**Aims**

The aims of PHGEN are:

- To conduct a networking exercise on Public Health Genomics (PHG) covering all EU Member States, Applicant Countries, and EFTA-EEA countries.
- To identify and list key experts relevant to PHG in these countries.
- To provide an inventory of PHG-issues and priorities in Europe.
- To contribute to the co-operation and exchange of information in order to enhance coherence and disseminate best practice.
- To identify legal diversities and barriers in a cross-border market.
- To promote and stimulate the countries' efforts in this emerging field by developing PHGEN and by supporting effective networking in order to reach sustainability.

Thus, in the long run, PHGEN will serve the European Commission as an "early detection unit" for horizon scanning, fact finding, and monitoring of the integration of genome-based knowledge into public health.

**Approach**

PHGEN involves experts as collaborating partners from the fields of human genetics and public health as well as representatives of relevant competent authorities from all EU Member States, Applicant Countries and EFTA-EEA countries. Experts on European law will be involved in the networking exercise identifying legal diversities and barriers in the different countries and contributing to a European added value by thus working toward best practice recommendations.

Representatives of other European networks (e.g. EuroGenest, EUseHTA, Orphanet, NuGo) are involved in the network to ensure complementarity between EU-funded projects. In addition, representatives from health-relevant organizations (e.g. WHO, WTO, OECD, STOA) will be invited to participate.

With these experts, regular meetings will be held to work toward the aims named above. The overarching guiding principle for a systematic analysis of the tasks set out will be the application of the public health trias (assessment, policy development, assurance) which has already been approved in other fields of public health.

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**Community Genetics 2006, Vol. 9, No. 1**

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## Group Ponders Genomics and Public Health

Xavier Bosch, MD, PhD

THE EUROPEAN UNION (EU) HAS launched a new project to ensure that as public health systems exploit advances in genetics, the rights of individuals are protected. The Public Health Genomics European Network (PHGEN) "will not only identify where and how the EU has to approach challenges and changes posed by public health genomics but will also give concrete recommendations on how genome-based knowledge can responsibly and effectively be integrated into public health," said Helmut Brand, MD, MSc, project leader and director of the Institute of Public Health of Nordrhein-Westfalen, in Bielefeld, Germany. The PHGEN is essentially run by the Institute, the German Center for Public Health Genomics (also in Bielefeld), and the Public Health Genetics Unit, in Cambridge, England.

To date, public health policies and practices have mainly dealt with environmental determinants of health and disease, and little attention has been paid to targeting individuals according to their genetic profiles. But knowledge of gene variants that affect an individual's susceptibility to a disease or response to therapeutic and preventive interventions is fast increasing. For example, the discovery of gene variants that can affect an individual's risk of developing coronary heart disease make it possible

to identify individuals who are at increased risk of developing the condition and to quantify the effect of such variants. However, it's also important to understand and quantify the effect of important modifiable risk factors for coronary heart disease, such as smoking and hypertension. "Based on the knowledge of these attributable risks, sound policies and effective interventions can be made," said Angela Brand, MPH, PHGEN speaker at the German Center for Public Health Genomics.

The network intends to work out how EU policies apply to advances in public health genomics. "It is a task of PHGEN to identify what regulations, or not yet regulated fields, are relevant to public health genomics," said Helmut Brand. He added that PHGEN will monitor the integration of genome-based evidence with public health by working with other networks established by the European Commission (EC), including EuroGen-test (which addresses questions about the clinical validity and utility of genetic tests), EUnetHTA (which works on health technology assessment), Orphanet (European network on rare diseases), and NuGo (European network on nutrigenomics).

"We are excited about the European initiative because realizing health benefits from genomic research depends above all on collaborative efforts," says Marta Gwinn, MD, MPH, of the Office of Genomics and Disease

Prevention at the Centers for Disease Control and Prevention in Atlanta.

The EC is providing \$1.6 million to fund the project until the end of 2008. At that time, it will consider a second proposal to fund the project for another 3 years.

During PHGEN's first 3 years, 100 experts from 34 countries will provide an inventory of public health genomics issues and priorities in Europe while identifying legal differences and barriers in a cross-border market. "Plus we will work on the institutionalization of public health genomics in the different European countries," Helmut Brand said.

Speaking at the PHGEN's first network meeting in February, in Bielefeld, Dolores Ibarreta, PhD, of the EC Joint Research Centre, said that PHGEN may help fill in some gaps in genetic testing in Europe, where pre- and post-counseling are not always adequate and about half of the laboratories performing genetic testing have never been officially inspected. Besides genetic testing and gene therapy, the PHGEN will assess policy pertaining to pharmacogenomics and nutrigenomics, recommendations for principles governing biobanks, and practices of preimplantation genetic diagnosis in the EU.

"In a final report, we will make recommendations to the EC, which might then suggest legislation relevant for public health genomics," said Angela Brand. □

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