



FINAL REPORT

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Project Acronym : EUHEALTHGEN

Project Title : Harnessing the potential of human population genetics research to improve the quality of health of the EU citizen

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Title of Report : From Biobanks to Biomarkers – translating the potential of human population genetics research to improve the quality of health of the EU citizen

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1. Executive Summary

- 1.1 The main objective of EUHEALTHGEN was to stimulate greater awareness and greater co-ordination of the activities of biobanks across Europe and elsewhere. It did this through the organisation of an international conference which took stock of developments in the field and the level of their application. This report should be read in conjunction with the published report of the conference and the corresponding lay summary report given in Annexes 6 and 7 respectively.
- 1.2 EUHEALTHGEN was successful in bringing a significant number of researchers (over 200) from diverse backgrounds and countries of origin (32) together to discuss how population genetics research could improve the future healthcare of the EU citizen (Annex 2 of the conference report). It promoted the development of collaborations amongst the existing biobanks across the European Union and has drawn attention to the current (see Annex 3 of the conference report) and future resources available (notably through FP7) for population genetics research across the ERA.
- 1.3 The conference report and its lay summary has been distributed to over 500 individuals from the scientific community, the lay public and both national and international politicians. The intention is to inform not only key stakeholders within the constituent scientific community but also policy makers within the European Commission and national governments and to highlight the key role that human biobanks will play in future developments of improved healthcare delivery across Europe. This will ultimately play a significant role in changing the health delivery paradigm from one of disease treatment to one of disease prevention.

2. Introduction

- 2.1 The progress in human population genetics made during the past two decades has emphasised the importance of developing collaborative partnerships to address major research topics that, because of funding or population constraints, cannot be accommodated by single countries alone. A key common challenge for all programmes on human population genetics is to identify ways in which research outputs can be used to improve public health and preventative healthcare. The implicit assumption is that this will eventually enhance industrial competitiveness and wealth creation.
- 2.2 Common diseases of major public health importance are phenotypically complex with many having a heritable component. Population genetics can be used to characterise and stratify these complex diseases. However, this requires access to databases

containing genotypic, clinical, environmental and lifestyle information on individuals along with corresponding clinical specimens (biobanks). These databases can be used for :

- Linkage studies to identify gene sequences associated with inherited diseases.
- Association studies to correlate a genetic change with a specific disease when there are no obvious patterns of inheritance.
- Investigating how genes, environmental factors and lifestyle act independently or in combination to influence susceptibility to disease and how, once a disease develops, it may progress.
- Investigating the diversity of genetic risk factors in different European populations.
- Pharmacogenetic analyses to determine the genetic basis of drug metabolism and induction of adverse drug reactions.
- Reclassifying disease phenotypes based on the genetic/biological background.

2.3 Several such databases already exist in, or are being planned by, a number of Member and Associated States and the Candidate Countries but at present there is little collaboration between them. This is largely due to difficulties in sharing or exchanging material and/or information and to the lack of standardised and quality controlled protocols for data collection, sample storage, analysis, and access etc. Furthermore, few of these databases provide for prospective studies of disease outcomes and, even then, there are considerable restrictions to access.

2.4 Significant advantage could therefore be gained if there was greater co-ordination of the activities of biobanks across Europe and elsewhere. It is this opportunity that EUHEALTHGEN planned to address. It aimed to promote the development of a large multi-national programme to harness the potential of human population genetics research with a view to promoting a paradigm shift in healthcare.

2.5 This would see healthcare moving from disease diagnosis and treatment to the identification of personal disease risk and the development of appropriate personalised prevention strategies. By adopting this strategy it is assumed that individuals would benefit through a reduction in their disease burden and that communities would benefit by reducing overall lifetime healthcare costs, leading to a stimulation of economic growth and industrial competitiveness across Europe.

3. Project Report

3.1 Project Objectives

- 3.1.1 The ultimate aim of this programme is to stimulate greater co-ordination of the activities of biobanks across Europe and elsewhere. This will involve complex negotiations as many scientific, technical, social, legal and ethical barriers need to be overcome. EUHEALTHGEN represented a crucial first step in this process and its chosen medium was the organisation of an International Conference for top scientists and other likely stakeholders to explore relevant issues associated with the creation and use of biobanks across Europe.
- 3.1.2 The Conference had multiple objectives. First, it was to take stock of developments in the field and thereby identify possibilities for harmonising data collection and use of genetic, disease end-point and environmental information across a number of biobanks. Second, it was to facilitate the development of specific guidelines, protocols, standards and recommendations across each Member and Associated State. Third, the conference aimed to help the integration of clinical, environmental and lifestyle data into an individual's medical record and so improve the cost-effectiveness of healthcare provision across Europe.

3.2 The Steering Committee

- 3.2.1 A Steering Committee composed of the consortium of stakeholders that developed the Specific Support Action proposal was established to guide the activities of EUHEALTHGEN. Its membership and the Institutional affiliations are shown at Annex 1. The Steering Committee was charged with the development of the Conference programme and, subsequently, the identification of a strategy for the better utilisation of the resources available for population genetics research. The Steering Committee met a total of four times.
- 3.2.2 The first meeting, held in Brussels on 21 – 22 September 2004, was used to establish the Committee and to agree a plan of action for the development of the Specific Support Action proposal. The Agenda for this meeting is shown at Annex 2. No minutes of the meeting were kept as the sole agreed action was to appoint Dr Peter Greenaway as the Project Coordinator who was then to produce the first draft of the required proposal. This was circulated to Steering Committee members for comment, modification and subsequent approval.
- 3.2.3 The second meeting of the Steering Committee was held on 12 April 2005 at the Wellcome Trust in London. It considered progress of the application for a Specific Support Action, the format of the proposed conference, possible speakers and other

participants and publicity issues. The minutes of this meeting (minus the draft conference programme and the list of possible participants) are given at Annex 3.

- 3.2.4 The third meeting of the Steering Committee was held on 22 June 2005 in Brussels to agree and endorse all the proposed arrangements for the conference and the subsequent publication of its proceedings. The minutes of this meeting are given at Annex 4. The final details of the conference, participant list etc were all agreed by email correspondence.
- 3.2.5 The final meeting of the Steering Committee was held after the conference in London on 14 March 2006 and not on 30 November as previously agreed. This was due mainly to delays in producing a draft report of the conference proceedings. The main business of the final meeting of the Steering Committee was to sign off this report and to agree the arrangements for its publication. The minutes of this meeting are given in Annex 5.

3.3 The Conference

- 3.3.1 The international conference organised on behalf of the EUHEALTHGEN consortium was held at the Wellcome Trust Conference Centre, Hinxton, Cambridge, UK on 20 – 22 September. The conference title was 'From Biobanks to Biomarkers : translating the potential of human population genetics research to improve the quality of health of the EU citizen. It was attended by approximately 200 researchers from 32 countries. The web co-ordinates of the published report of the proceedings of this conference is given in Annex 6; the web co-ordinates of a lay summary is given at Annex 7.

4. **Outputs**

4.1 Strategic impact on population genetics

- 4.1.1 The main objective of EUHEALTHGEN was to stimulate greater co-ordination of the activities of biobanks across Europe and elsewhere. It aimed to take stock of developments in the field and the level of their application. The International conference was the instrument by which this objective was achieved. An overview of the conference and a summary of the key presentations made are given in Chapter 4 of the conference report.
- 4.1.2 EUHEALTHGEN also aimed to facilitate the development of specific guidelines, protocols, standards and recommendations regarding the use of biobanks across Member States and it aimed to facilitate the integration of clinical, environmental and lifestyle data into an individual's medical record. These were long term aims and it is difficult to assess in the short term whether

Annex 6 : The published report of the proceedings of the conference entitled ' From Biobanks to Biomarkers : Translating the potential of human population genetics research to improve the quality of health of the EU citizen'

This is available at www.wellcome.ac.uk/assets/wtx032086.pdf

Annex 7 : A lay summary of the proceedings of the conference entitled ' From Biobanks to Biomarkers : Translating the potential of human population genetics research to improve the quality of health of the EU citizen'

This is available at www.wellcome.ac.uk/assets/wtx032985.pdf