

## UK Biobank: Good for Public Health?

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24 July 2003

*GeneWatch UK might just be the kind of 'genetic union' Mike Fortun had in mind when making the case for 'genomic solidarity'. Here, their Deputy Director scrutinises Biobank UK, its aims, cost, science and commerce, foregrounding the questions GeneWatch wants answered. She makes the case for a democratic debate which alerts the public to such unknown variables as the research aims, database assessments and issues of donor anonymity involved, before they give their consent.*

The UK Medical Research Council (MRC), the Wellcome Trust and the Department of Health announced the allocation of £45 million start-up funding to the UK Biobank in April 2002. The project "will be the world's biggest study of the role of nature and nurture in health and disease". Yet, before a single sample has been collected, the UK Biobank is mired in controversy about its aims, its costs, its underlying science and its relationship to commercial exploitation.

UK Biobank aims to collect DNA samples from 500,000 volunteers between the ages of 45 and 69. This genetic data will be linked with lifestyle information taken from an initial questionnaire together with details about subsequent sickness, medication and causes of death taken from the volunteers' medical records.

The Department of Health sees UK Biobank as a pilot project for a national genetic database, potentially including the whole of the National Health Service (NHS). This idea (partly inspired by the DeCODE database in Iceland) was first proposed in mid 1999 by

Dr (now Sir) George Poste, then Chief Science and Technology Officer of the pharmaceutical company, SmithKline Beecham. Poste's proposal for a "public-private partnership" was discussed with ministers in December 1999, a "call for proposals" for large DNA collections was issued by the MRC in January 2000, and the Government allocated £20 million to the MRC (its share of the funding for the UK Biobank) in November of that year. GeneWatch UK's key concerns are that:

- UK Biobank's aims are controversial: prediction of future common illnesses by testing people's genetic make-up is unlikely to be a successful or cost-effective means of disease prevention.
- UK Biobank's science is highly questionable: the serious limitations of its design mean that genetic factors in disease or drug response will be hard to identify correctly, and spurious links between genes and diseases may be made.

- UK Biobank will not contribute directly to the development of new medicines, but may allow commercial companies to go “gene fishing”, patent gene sequences and gain excessive monopolies over future treatments.
- There is a lack of legal safeguards to protect participants and others from future misuse of their genetic information: there are no laws to prevent genetic discrimination by insurers or employers, and inadequate controls on access by the police or by commercial companies.

All these concerns raise underlying issues about public involvement in decision-making. Why have the implications of UK Biobank for health policy never been debated? Why has the process of scientific peer review been so secretive? What is the relationship of UK Biobank to commercial companies? What has happened, what will happen to ‘informed consent’?

What does the UK Biobank mean for health?

There has been no democratic debate about the health strategy that underpins the UK Biobank. Its main aim is to identify genetic and environmental factors predisposing individuals to common diseases, such as heart disease, cancer and mental illness. The Medical Research Council (MRC) has claimed that the UK Biobank will lead to “individualised risk assessment and preventative advice or treatment” and “a major shift in emphasis from treatment towards prevention”. It has stated that the understanding developed using the Biobank “will be used to predict the likelihood that an individual will develop a disease so that medicines can be used to prevent its onset rather than as a treatment for symptoms once a disease develops”. Lifestyle advice could also be targeted at those identified as ‘genetically susceptible’ to future illness.

However, genes are poor predictors of future common illnesses and this approach to disease prevention is highly controversial. It has more to do with increasing the market for genetic tests, and associated ‘preventive’ medicines or supplements, than with reducing the incidence of cancer, heart disease and diabetes. Biotech and pharmaceutical companies have recognised that genetic tests, reaching the market far earlier than new treatments, could provide a means of

generating “near term revenue”, while also allowing expansion of the pharmaceutical market to healthy people identified as “genetically susceptible” to future illness – sometimes called the “healthy ill” or “worried well”. The former Chairman of GlaxoSmithKline, Sir Richard Sykes, has predicted that within 20 years most people in developed countries will receive ‘pre-symptomatic treatment’ while they are still healthy.

No public assessment has been made of the relative costs and benefits of a genetic approach to disease prevention, compared to alternative population-based measures such as banning the advertising of unhealthy foods to children. The growing epidemic of obesity, for example, is not caused by increases in ‘genes for obesity’ but by unhealthy diets and lack of exercise. There is an urgent need for more public and parliamentary debate on the potential implications of genetic ‘prediction and prevention’ – for society and for the NHS.

Will the UK Biobank achieve its aims?

Concerns about the science of UK Biobank are closely linked to doubts about its aims. Finding genetic factors in common diseases can lead to benefits to health – by identifying new biological mechanisms or pathways. However, this usually means identifying new links

between genes and diseases (in the hope that it might prove possible to design a new drug to target the gene) or using genes to find clues to other factors that are amenable to intervention (intermediate traits, such as cholesterol levels). Both these aims require much more detailed information than will be collected in the Biobank, not to mention careful design to minimise spurious associations between genetic variations and disease. Biobank’s main aim, in contrast, is to quantify the risk associated with having particular common genetic variations (called ‘polymorphisms’) and being exposed to certain environmental factors (such as smoking).

Not only are such individual risk assessments of questionable value to health, there are also serious doubts about the science underlying this approach. The majority of genetic associations are never replicated, and the underlying rationale that a few common genetic variations lead to most common diseases is in doubt. The limitations of the data –

*“[Biobank UK] is a big gamble... People who opt into this study have to know exactly what is being done with this DNA. They need to know its relationship to any industrial exploitation.”*  
*Professor Sir David Weatherall, Oxford University.*

particularly the use of medical records for follow-up – has also led to claims that “garbage in” means “garbage out”. Biobank’s draft scientific protocol includes only one paragraph on the complexity of interactions between multiple genetic and environmental factors and no analysis of how these might affect the outcomes. Misleading statistical conclusions could easily be drawn.

Finding new genes is best achieved by studying high-risk families, where the effect of a genetic factor is expected to be strongest. New traits cannot be found unless they are also measured – an issue that has led to the Biobank being described as “a poor vehicle for study of cardiovascular and metabolic disease”. Participants in the Biobank’s protocol workshop proposed studying a smaller group of 20,000 to 100,000 people in more detail, however funds for such a study have not been allocated. Some scientists have argued that funding this type of one-off smaller study would provide much better value for money and a better test of the causes of disease, obviating the need for a larger UK Biobank. However the MRC has not provided a comparative assessment of the costs and benefits of this type of approach. The full costs of UK Biobank have not been published – some scientists at the Workshop estimated that the true costs of meeting its objectives might be more than £500 million.

Political concern about the science has led to claims from the Government that it would not be appropriate to peer review the project like ‘any other grant proposal’. Yet the funders have previously claimed that the funding decision was based on “thorough peer review done according to well established principles”.

GeneWatch believes that the UK Biobank should undergo a new, transparent and independent scientific review process and assessment of its likely value-for-money. These issues could then be publicly debated and resolved.

### Fishing for genes?

The Biobank’s promotional leaflet claims that “in order to develop new treatments and drugs, scientists in pharmaceutical companies will need to have access to the information”. However, people are concerned about the potential for profiteering by pharmaceutical and biotechnology companies involved in UK Biobank research, including the patenting of genes.

In evidence to the House of Commons Science and Technology Committee, Sir George Radda, Chief Executive of the MRC, stated that according to Biobank’s policy, it must be possible to patent genes

with known functions as a safeguard for industry. Biobank’s new director recently denied this during a debate with GeneWatch – but a new policy has not yet been published.

The patenting of gene sequences is morally objectionable to many people and allows unprecedented monopolies over future genetic tests and treatments. There is growing concern – some of it in industry – about negative impacts on research and innovation. Current legislation does not require people to be informed if their genes are patented and there has been no public consultation on the approach Biobank should take.

Genes identified in UK Biobank could be claimed in patents for “diagnostics” – that is for genetic tests to predict the risk of future disease. Although the link between a gene and a disease is really a discovery, patent offices have commonly allowed such links to be used as the basis of a claim for an invention. This type of patent is particularly controversial because it allows a company to assert rights over all future uses of a gene, usually for 20 years. The existence of such patents adds to concerns that companies may go “gene fishing” in the Biobank – claiming monopolies over future uses of gene sequences simply by finding a (possibly spurious) statistical association between a gene and a particular disease.

### What are participants consenting to?

Research teams based in universities or companies are expected to apply to use the Biobank. The plan is to seek a general form of consent to all future research. However, this risks a loss of trust in medical research if people later feel that they were misinformed. Is the jettisoning of ‘informed consent’ really desirable or necessary? It seems likely that any genuinely useful studies will require the collection of additional data to test specific hypotheses, and will involve renewed contact with participants for follow-up purposes, rather than relying on medical records. If this is the case participants could simply be asked for their consent to new studies as they arise, and given the recommended information regarding the aims, methods, sources of funding, potential conflicts-of-interest and anticipated benefits and risks.

It is not only in connection with patenting and profiteering that controversial commercial conflicts-of-interest arise. What happens if commercial companies seek to undertake research to identify those who are ‘genetically susceptible’ to diseases associated with their own products or pollution? These could include

tobacco companies, food companies, or employers seeking to identify those who are susceptible to hazardous chemicals or radiation. The funders' failure to rule out such studies means that people taking part will not be able to consider in advance whether research by particular commercial interests, including their own employer, is something that they find acceptable. New democratic mechanisms are needed to involve the public in setting the research agenda – perhaps also drawing up a 'code of practice' that sets the boundaries for what is acceptable before consent is sought.

Legal safeguards also need to be in place before volunteers are asked to give their data and their samples. This is important, not only to protect individual participants, but also to ensure that genetic tests developed in the Biobank are not used to discriminate against others in the future. People donating samples to the Biobank will hope that the research will benefit those who are susceptible to future illness because of their genetic make-up. Yet genetic tests developed using UK Biobank could be used in future to discriminate against these people – for example, by refusing them insurance or a job.

Organisations such as the Trades Union Congress (TUC) oppose genetic screening of the workforce as a false option in terms of controlling workplace risks. The workplace should be made safe for everyone rather than selecting out those workers considered to be 'genetically susceptible' to certain hazards. Yet, currently, there are no laws to prevent genetic discrimination, and the UK Government has not signed and ratified the European Convention on Human Rights and Biomedicine.

There is also no specific UK legislation for the protection of personal genetic information. Moreover,

the basis on which the police might be granted access to the Biobank, under warrant, remains unclear. Addressing this issue is particularly important because the Government sees the Biobank as a pilot study for a national genetic database, potentially including the whole of the NHS. This raises the prospect of future erosions of civil rights, by using DNA collected for medical or research reasons for citizen surveillance or forensic purposes.

A public assessment is also needed of the extent to which the data from UK Biobank, even when supplied on an "anonymised" basis, could be used to identify individuals. Recent research seeking to build up a "genetic photo-fit" from DNA samples left at crime scenes – including predicting surname or red hair colour – renders the idea of truly "anonymous" genetic information rather meaningless. Both postcodes and employment information are likely to be used by some third parties using Biobank UK – but they may also help reveal an individual's identity. Certain health events – such as adverse drug reactions (a topic likely to be of interest to pharmaceutical companies) – are also often rare enough to make access to specific information sufficient to identify an individual.

## Conclusions

There is a real need for public and parliamentary involvement in decisions about UK Biobank, including the ethics of diverting resources from other health research and 'streamlining' consent. Without proper legal safeguards and open debate, there is significant potential for a loss of public trust in medical research. Despite numerous consultation exercises, none of the issues outlined above has yet been democratically debated or resolved.

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