
The Making of a Biobank. The Case of the Estonian Genome Project

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Abstract

Large, sometimes population-based biobanks, have by now become a commonplace feature of the research landscape. My focus will be on the Estonian Genome Project (hereafter EGP) – its main characteristics, the legal framework and recent changes to its status and objectives. My main interest is to analyse the discursive context of the establishment of the EGP, looking at the ways in which scientific, political, economic, nationalist and other discourses influenced the coming into existence of a large complex socio-technological artefact. EGP is an undertaking the inception and development of which provide an explicit case study for the analysis of the essential engagement and co-production of politics, market and research interests. My focus will be on the discursive level – on the rhetoric employed by various actors, on the explanations and arguments put forward in the media, as well as implicit premises of the policy moves and targeted legislation.

Biobanking and the Estonian Genome Project

Large collections of human biological data (or henceforth biobanks) are hoped to become an important resource for biomedical research. Biobanks usually contain genetic data and often also medical data, additionally newer collections aim to gather information about the lifestyle and environment of the donors or participants. *Genetic database* has been defined as:

a collection of data arranged in a systematic way so as to be searchable. Genomic data can include inter alia, nucleic acid and protein sequence variants, mutations, and polymorphic haplotypes. The work associated with a database includes collecting, annotating, curating, storing, validating and preparing specific sets for transmission. (HUGO 2002, HUGO Ethics Committee Statement on Human Genomic Databases 2002).

Human biobanking has taken off internationally and indeed, globally. Recently large consortia of biobanks have been formed to provide a basis for research cooperation among those structures (for example the P3G – Public Population Project in Genomics, or the more recent European initiative BBMRI – Biobanking and Biomolecular Resources Research Infrastructure etc.). Research is increasingly focusing on causes and cures of common complex diseases (in comparison to the earlier focus on the more easily detectable monogenetic diseases), on the relationship between genotype and environment and lifestyle, and on gene-drug interaction (pharmacogenetics). For all these purposes, and potentially many more, biobanks are a suitable research tool.

My case study is that of Estonia, a small country with a population of 1.3 million that regained its independence from the Soviet Union in 1991. Estonia may well be characterised as a ‘techno-optimistic’ country where the possibilities offered by various new technologies have been embraced with eagerness and often pride. This ‘Estonian techno-optimism’ is not limited to medical technologies but is also manifest in mobile technologies (e.g. car parking by phone was first introduced there, m-banking or banking by mobile phone is also offered), information technologies (the national Tiger Leap project provided IT infrastructure for all schools), widespread e-banking, e-governance (internet-based communication with local and national government) and recent e-voting.

One of Estonia’s most renowned and ambitious engagements with science and technology has been the Estonian Genome Project (EGP) that initially aimed to collect genetic and medical data from up to 1 million people. EGP will permit the implementation of studies to identify genes causing and influencing the development of common diseases (the database will include information on both phenotypes and genotypes). A longer term goal of the project has been the practical implementation of gene-based medicine to public health on a massive scale. Although the project was initially started in 2000 it has since undergone several important changes. But let’s first look at the initial setup and distinguishing characteristics of the EGP.

One of the most significant traits, especially in comparison with other large population databases, was the promise of possible feedback of indi-

vidual genetic data to participants – in other words, participants are to be informed of their genetic risks (if they so wish, they also have a right not to know). Secondly, the database was to be literally population-based, aiming to include up to 1 million samples from a population of 1.3 million. This would have made the collection similar to the Icelandic case where almost the entire population (of little over 300,000) was to be included. However, in Iceland the population was included by default and people had the option of opting out of the health sector database. In contrast to the Icelandic project, the EGP requires the explicit voluntary consent from participants.

Thirdly, the recruiting process takes place at the general practitioner's office, this is also where the informed consent is signed and the blood sample together with health and genealogical information is collected. It is debatable whether the form signed in the context of biobank research qualifies as informed in the sense of the Helsinki Declaration. Quite clearly it is impossible to provide sufficient data about the expected research – since biobanks exist for the very reason of providing a resource for different and sometimes unforeseeable research needs. Discussions are ongoing regarding the ethical dilemmas such research participation involves and about the possible future of informed consent as 'open consent', 'proxy consent', 'authorisation' etc. (Nõmper 2005).

Last but not least, a special law, the Human Genes Research Act, was passed by Parliament in December 2000 to govern the setup and management of the Foundation. The Act determines the rights of gene donors, data processing and protection in the bank and prohibits discrimination in employment and insurance relationships or any other discrimination based on the structure of a person's DNA and his or her genetic risks.

To reiterate, the main distinguishing characteristics of the EGP at its inception were the following:

- population-wide
- opt-in system
- promise of personal feedback
- involvement of GPs
- dedicated legal framework in place.

Perhaps the most significant of these was the EGP's promise to return information about genetic data and risks to all those participants who wished to know. This important population-wide aspect, together with the potentiality of almost everybody actually knowing his or her genetic constitution, formed the background for several health care ethics issues. For example, how is such a large-scale feedback effort organised? Is appropriate genetic counselling provided (and who is paying for it)? Is the right not to know respected in an environment where people might be increasingly perceived as responsible for their health? How would that personal responsibility for health reflect in the provision and allocation of health care resources?¹ But time has shown that at least some of these issues will not become burningly relevant, at least for some time.

Several important changes have taken place in the EGP over the last years. In 2004 the private investors behind the EGP grew impatient with the low speed of gathering donor samples and disagreed with some of the research foci in collecting phenotypic data. This eventually led to their disengagement from the project and the Estonian government had to step in with investments to save the project. Since 2007 the EGP is formally part of the University of Tartu (the oldest research institution in Estonia) and is funded publicly by the state and university as well as through several international research cooperation projects. The objectives of the project have changed somewhat (now the aim is to collect 100,000 samples) and the rhetoric has also and quite significantly followed a change of course (feedback has been forgotten and the language of altruistic donorship has taken over). However, due to space restrictions I have limited my focus to the original setup of the project and the initial discursive tactics employed there.

The discursive setting of any large national public undertaking is multi-levelled. Such a grand project is never easily categorisable – while promoted as a medical research tool, it is simultaneously many other things – a public health initiative, a for-profit business enterprise, a national undertaking, a politico-legal event etc. Needless to say, none of these 'appearances' are distinct but they provide a number of storylines for the governance and marketing of such a complex sociotechnical artefact. I will analyse how the EGP was portrayed as simultaneously delivering

scientific progress and potential public health benefits, as well as being an economically lucrative business project with national and international significance. How are these multiple 'tasks' or 'capabilities' brought about and explained?

The EGP as a public health initiative

Participation in large scale biomedical research projects like population biobanks has been under scrutiny lately (Tutton 2007), and the very word 'participant' evokes a sense of reciprocity and 'being part of' some common project. Participation can of course be conceptualised in various complementary or conflicting ways. While there has been a lot of discussion in science communication debates about the necessity to provide much-needed legitimacy in areas previously 'ruled by experts' (e.g. science and its various applications) through increased collective public engagement, it is simultaneously essential to motivate participants as individuals in their projects of self-rule and responsible management.

In the Estonian case the discourse of 'gene donors' was utilised, which places the endeavour within traditional medical research settings. Almost the entire population was to be voluntarily involved, with everyone being promised feedback on the genetic risks and the database was to be continuously updated later with medical histories. The EGP thus hoped to become one of the first large-scale playgrounds for active symbiosis of genetic research with the public health care system through the intimate involvement of GPs in the project (they were to collect the samples and fill in the questionnaires). The initially expected scope of involvement of the EGP in the lives of most Estonians was thus rather grand.

In general, databases have primarily been perceived as potentially powerful research tools and their relevance and impact on public health (of donors and their contemporaries) has therefore been rather indirect or at least a rather long-term one. For example, the UK Biobank collects data from those aged between 40 and 69 and the benefits are to be expected mostly for the next generations.²

The EGP organisers quite straightforwardly linked the project to the Estonian public health system, primarily by means of two measures. The data was to be collected by GPs, thus making use of the existing public health infrastructure. Secondly, all donors had the right (written into law) to learn of their genetic information. While the details of this information-giving process remained vague, the main idea behind this rather unusual promise was to engage people actively with the project, promise them individual benefits and feed relevant information back into the health care system. The initiators of the project have insisted that allowing decoding of genetic data will give donors the possibility of directly profiting from the project, as they can take preventive measures according to the risks revealed. The gene donors were promised a personal 'gene card' giving them an opportunity to make use of personalised medicine.

Our sociological study showed that the main motivation for people who had decided to participate in the project was the wish to obtain a personal gene card (among those who definitely planned to participate, 96 % wanted the 'gene card').³ Although the so-called gene card was a popular aspect of the project among both the promoters and the public, in reality it never became a very realistic entity even on the discursive level, not to mention the many difficult scientific, ethical and financial factors related to it.

The EGP as a research tool

Sociologists have established that in Estonia there is generally a positive and trusting attitude towards scientists and a strong belief in scientific progress (Korts 2004, 247). Several explanations can be relevant here. Firstly the experience of Soviet valorisation of science – in the pursuit of communism, science and technology assumed centre stage in providing for the major expected breakthroughs. Also, the smooth uptake of rather controversial research applications can be attributed to an underdeveloped socio-critical discourse. While modernity has viewed science and also technology as tools for social progress, it has also been marked by a more critical stance towards the risks that our increasing reliance on science and technology bring along. But while Soviet modernity had applauded the rationalisation of

technology and the application of scientific achievements in practice, it had suppressed the parallel process of emancipation, critical reflection and mobilisation that characterised Western modernisation. For example, in relation to trusting the statements about the EGP, over 80 % of people tended to trust most of all geneticists and the employees of the EGP (Korts 2007, 50). This does not necessarily reflect optimism as such but rather a lack of scepticism about potential problems (Korts 2004, 249).

So, generally speaking, the Estonian public still seems to trust their scientists and their projects as beneficial and progressive by default. However, the actual role of scientists in 'selling the EGP' to the public (as well as to the politicians and the investors) departed from the modernist traditional position of neutrality and autonomy of science. Engaging actively with future health care needs and potential development trajectories in relation to applications of genetics, new categories of actors emerged – scientists as experts, not only in their specific fields but also as public policy visionaries and promoters of the project as a national centrepiece. The trust towards scientists as experts is transformed into political gain as the project becomes 'national' (despite the heavy involvement of foreign investors).

The debate largely took the form of weighing costs and benefits within the traditional framework of technology as 'applied' to society. This approach to the ethical and social weighing of new scientific developments allows for international comparisons, where Estonia seems to be sharing a standpoint with the utilitarian calculus of the USA, and thus differs from the expert-based complex debates of Germany as well as from the more transparent working groups and ambitious public consultations of the UK (Jasanoff 2005).

From the very beginning, the EGP was very much inspired by and linked to similar research internationally – the biobank bandwagon was moving and Estonia had a chance to join this process while it was still considered relatively new. When the project was still mostly privately funded, the plan was to sell access to the data for companies and researchers internationally. Later, when public funding was increased and the private funders withdrew, the EGP collaborated closely with other biobanks in the framework of research projects.

The project was seen as instrumental in pooling Estonian specialists and researchers in the area – both providing jobs for those in Estonia and as a motive for returning scientists (biology, especially molecular biology, genetics and biotechnology are considered some of the most successful disciplines in the Estonian academic world, in terms of publications and international cooperation). A similar ‘reversing of the brain-drain’ argument to the Icelandic case is discernible here.

In contrast to most similar research undertakings, the project’s set-up was characterised by the focus on self-interest as a motivating factor in participation for the people involved. How can this rather straightforward departure from the more traditional ways of doing research be explained? Traditionally the guiding motivational basis for research participation has been altruism – participating so as to further research and help future generations as well as humanity as a whole. Indeed, personal gain has been seen as unfitting and corruptive in this domain (e.g. Titmuss 1997).

One of the things to consider is the neoliberal political climate in Estonia focusing on individual responsibility and entrepreneurial spirit in general (see the section below on politics). Another is perhaps a less local and more global phenomenon – namely the rhetoric of bioethics. This interdisciplinary field has been crucial in conceptualising and reflecting upon the many novel issues and entities that have made their (re)appearance within the past couple of decades of biotech research. While bioethics often has local / national undertones in terms of debated topics and preferred solutions to problems and dilemmas, there are also global attributes. For example, bioethics as an institution has been criticised as too complacent and too industry-driven, almost a by-product of the biotech development. It is seen as instrumental in turning patients into consumers and of fetishising choice in all areas and in all situations as it is overwhelmingly focused on individual interests, to the detriment of communal or social interests and needs (e.g. Kerr & Shakespeare 2002). Some of that criticism might well have its merits, especially when we think about the bioethics of the 1990s. In the present climate, however, there has been quite a significant turn towards communal values like solidarity, reciprocity and collective interests (Chadwick & Berg 2001; Knoppers & Chadwick 2005). Also, to be fair, bioethics of the earlier

days was largely concerned with avoiding and warning against the excesses of the eugenics movement, hence the extra effort to ensure respect towards individual rights and privacy.

Bioethics has been a tool in producing order and decreasing ambiguities but we must not assume that this has been done from a neutral basis. It is illustrative for example how bioethics has been subservient in national debates in conceptualising the central worries and risks of new biomedical technologies in very distinct ways (in the USA for example, the economic interests of individuals and groups are often articulated in the language of ethics – privacy, personal choice, corporate responsibility, see Jasanoff 2005). Ethics and bioethics in this case are in the service of powerful national narratives – law and order in Germany, securing the sanctity of research space in UK and so forth. So let's look more closely at the politics around the EGP.

The EGP as a political project

I take the term 'political' to have two meanings in my overview. Firstly, it denotes the straightforward public activities of politicians in promoting the project and the legislative efforts made to ensure that the EGP got off to a successful start. Secondly, I am aiming to capture politics in a more inclusive sense of the word – the often tacit presumptions in conditioning the set-up of the EGP, its rationale, the marketing and possible future uses of the project. In other words – what are the underlying and often implicit assumptions about the role of such an institution? What kind of arguments are seen as motivating for potential participants? What sort of premises and values are present in visions for the future of health care in relation the EGP. The special focus here is on the symbiosis and cooperation between politics and science.

There is a certain parallel here with Sheila Jasanoff's notion of political cultures and civic epistemologies – the tacit but nonetheless powerful routines by which collective knowledge is produced and validated in countries. There are always different ways of dealing with novel technologies and different countries have taken different routes in introducing,

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debating and embedding these technologies. Yet my focus is less on the mechanisms of governance and problem-solving in the face of new technologies and more on the ideological content of such activities and the values they are reinforcing.

While the economic interests and the subject matter of competitiveness of national research are important concerns everywhere, these considerations have rarely taken such centre stage in political life. The consensus on the necessity of going forward with the project was buttressed through the smooth drafting and passing of the special Human Genes Research Act in 2000 that set out the governance of the database. It is especially significant to note that the extreme *laissez-faire* approach and the stringent delineation of state politics from the business and/or research activities usually characterising Estonian policy-making was reversed in this case as politics took active interest in conceptualising the project as a 'national undertaking'.

Thus, from the very beginning the EGP was perceived not only as a research and health care infrastructure project but also as a national initiative with both domestic and international significance. This is not surprising as a degree of nation-building characterises most contemporary applications of novel technology on such a scale.

As discussed above, the EGP was marketed among Estonians as providing personal gain. My hypothesis is that this unique project design in the medical sphere that is usually ruled by altruism and gift-relationships, is at the same time very much in unison with the political climate of Estonia. In other words, the foci on individualism and personal responsibility characterise both the values of the ruling political elites as well as the arrangements of the EGP. Estonia has for the past 15 years been a country governed by very strong neoliberal policies. The move towards right-wing rhetoric and practices in Estonia has been quite exceptional even among the countries of Eastern and Central Europe, where the pendulum generally swung to the right after the fall of Communism. This ideology focuses first and foremost on the responsibilities and rights of the individual, and the disparaging comments regarding notions like solidarity and equality have been rather common among Estonian politicians.⁴ The way the EGP has been tailored to fit

the apparent ideological preferences of politicians (and of the public?) is instructive and highlights the way in which the political climate can negotiate the seemingly universal application of principles of medical and research ethics.

As the preparation for the EGP was not accompanied by active public discussion but could rather be characterised as a one-way advertising campaign (EGP sponsoring radio and TV programmes, newsletters etc), this raises additional questions regarding the legitimisation of such 'national' endeavours. An author of a discourse analysis of the EGP media debates has straightforwardly concluded that there was too little rational argumentation available to actually come to a reflected decision about the project (Hallap 2004, 219). Media representation of science is always selective in many ways but a strong pro-science rhetoric and even 'pseudo-argument' characterised the debates around the EGP (Hallap 2004, 238). As a result the press was full of reports and comments on the impossibility of stopping the progress of science (and therefore futile attempts to oppose the EGP), promises of the 'personal gene card' and the chance of national competitiveness that the project would provide.

Was the overwhelming approval of the EGP a positive practical acknowledgement and embracing of the eventuality of the 'knowledge society' in which acceptance and support for such highly complicated research tools and large-scale pursuits are necessary for national development or even survival? Or, in a more critical view, were traditional politics swiftly appropriated by (increasingly commercialised) science and research interests? The lack of long-term democratic governance traditions coupled with the 'hands-off' neoliberal ideology offer the potentiality of interpreting this case as the ultimate blurring of science and politics. If science has traditionally been insistent on a separation from politics, then we are now widely witnessing a reciprocal acknowledgement and instrumentalisation of both of these institutions in their quest for normative power. Legitimation processes go both ways here – a population-based genetic database embodying the idea of a modern nationhood, and vice versa, the EGP being popularised and becoming a potent symbol through references to national identity (Tamppuu 2004, 213). Albeit tacitly, the EGP can well be seen as a case study of the state using and appropriating science

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for governance purposes. The opponents of the EGP are even seen as a threat to national unity: *One uses the project of the gene bank to scare people and to divide the nation* (Metspalu 2002).

To conclude, I find the EGP to be an interesting case study of how science as a social practice interlocked with the neoliberal political tendencies to produce a novel socio-scientific arrangement and carved out a space for the launching of the database. The boundaries became blurred between science and politics as a nationalist discourse was appropriated by scientists and the values of political ideologies took centre stage in shaping the framework of the biobank.

Notes

- ¹ For an overview of issues see Sutrop and Simm (2004).
- ² See UK Biobank web-site at <http://www.ukbiobank.ac.uk/assessment/takepart.php>. Downloaded 10.11.2008.
- ³ A nationally representative survey in relation to the EGP and surrounding attitudes was carried out in December 2002. Some research results have been published in Korts (2004).
- ⁴ One prime minister has publicly ridiculed the idea of solidarity and social justice in the media, the other has complained that any desire towards equality is always unfair and indeed, attempts to achieve equality are the very source of social tensions (interviews with Juhan Parts in *Tages-Anzeiger Magazin* (12.06.2004) and *Eesti Päevaleht Mõte* 2008, article by Siim Kallas in *Postimees* 12.11.2003).

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