“Human Genetic Databases: Towards a Global Ethical Framework”

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1. Genetic research challenges traditional ethical principles

In the past decades, the collections of biological samples and genetic data have raised a substantial ethical discussion on issues that had been commonly considered well understood. [1]. These issues were framed in terms of either individual rights or collective interests, with a prevailing impression that the distinction between the two is easy to draw.

Genetic data that have contributed to scientific knowledge ought to be considered public goods [2, 3]. This position—which a large part of the community of scientists and bioethicists hold [4]—reflects what the sociologist of science Robert Merton called “communalism” when referring to the public ownership of the fruits of scientific investigation [5]. Stimulated by the free circulation of information, ideas and experiences, scientific knowledge is by nature allergic to secrets and appropriation. On the other hand, personal genetic and genomic data pertain to the private sphere of individuals, thus reclaiming a role for two of the cardinal principles of medical ethics: the obligation to keep those data confidential and to protect the rights of the individuals involved.

What the evidence suggests, however, is that the issue lies not on genetic data or genetic knowledge themselves, but rather on the way genetic/genomics knowledge and data are obtained. How can we apply the basic principles—expressed by the Nuremberg Code, the Helsinki Declaration and other international instruments—in the context of genetic and genomic knowledge? Moreover, before undertaking biomedical research with human subjects, the prior, informed consent of participants is required in order to respect the autonomy of the research subject and in order to protect the subject himself or herself. The dual significance of the requirement for informed consent stems from the fact that two distinct ethical principles require it. Firstly, informed consent is the expression of the deontological obligation to respect the person and, secondly, of the utilitarian idea assuming that each individual is the best judge of his or her interests.

Although the requirement for informed consent and its moral foundations have been discussed thoroughly, genetic/genomic research colors these ethical obligations with implications that are controversial although they have been debate for many years also in this specific context.

2. Ethical issues with genetic data

Because of its nature, genetic data have implications that go beyond the individual concerned thus reaching family, community, and ethnic group to which he or she belongs. Each family shares not only emotions, experiences and beliefs…but also genes. And sharing genes is inescapable! What we learn about our genes allows us to know something about the genes of our blood relatives, whether we like it or not. Genetic data have also implications that go beyond the scope of a single study. A DNA sample is a source of virtual information. Certain data may be derived from the sample as part of a specific study—and there is an obligation to inform the sample sources about that. However, DNA also carries virtual information that may be pertinent to questions that today's researchers are not asking but that perhaps tomorrow's researchers would like to investigate. Preventing future investigation sounds unreasonable if not absurd. Yet the scope of the informed consent given at the time the samples were collected is controversial. Unfortunately, at this stage traditional bioethics principles do not offer clear responses [6, 7].

The traditional doctrine requiring prior informed consent of participants in biomedical research protects two interests: the autonomy of the research subject and his or her ability to protect himself or herself against possible risks. It is certainly difficult to define how to protect the research subjects' autonomy with respect to questions that will only be investigated in future. In fact, if we consider future developments, it is quite unclear what risks must be communicated to research subjects at the time their consent is taken. Shall information include both individual and collective risks? Both short and long term risks? Because genetic data spread their effects overtime, they have long term implications that, as in practice, neither not researchers can easily foresee.

Genetic data also raises other important issues, most of which remain controversial: whether any feedback should be provided to the research subjects, under what circumstances (if any) is it appropriate to re-contact them, and whether the benefits of the research must be shared with the participants [8]. Among the possible benefits, one must account the benefit for an individual to have access to a treatment that is inspired by genetic research performed in the future on the sample or data provided by him or her. The traditional solution to these issues is anonymizing of data [1]. However, one cannot have it both ways—benefit sharing and protection from the risk being re-contacted. In fact, anonymizing data protects research subject against unforeseen risks, yet it prevents them from enjoying the - possibly unforeseen

- benefits of the research. Anonymization may be still adequate solution under certain circumstances. However, one must also be also aware of the implications for research participants.

3. The study

Facing these issues, it is not surprising that the policy instruments that have been drafted in the last few years are either contradictory or inconclusive. International law instruments, national laws, ethical guidelines, professional guidelines and other kinds of less formal instruments reflect a debate that is characterized by perplexity and controversy. Within this context, the project entitled "Genetic Databases-Towards a Global Ethical Framework,"—carried out by Department of Ethics, Trade, Human Rights and Health Law at the World Health Organization (Switzerland), the Institute of Bioethics of the University of Geneva (Switzerland), and the Institute of Medical Ethics, Charité in Berlin (Germany)—aims at studying the conditions under which genetic databases can be established, kept, and used in an ethically acceptable way [9]. This study unfolds along two lines of investigation. First, it compares the existing policy instruments of different countries from a cross-cultural perspective. Second, the project investigates the opinion that experts have of these issues. To this end, a number of experts of different professional and cultural background form around the world are interviewed to obtain their opinion on this topic. From a conceptual point of view, the project adopts a framework shaped on the three stages by which genetic knowledge is obtained: collecting, storing, and using samples and data. For each of these stages, the project asks the following questions:

- Who collects, stores and uses samples?
- Why are samples and data collected, stored and used?
- Under what conditions are samples and data collected, stored and used?

For instance, collecting samples and data raises the important question of the nature and the scope of the informed consent of the research participants. Storage raises the policy question of the legal status of genetic databases and the ethical question of whether and how to implement the participants' right to withdraw their consent. Finally, the use of samples and data raise issues of benefit sharing and feedback to participants.

The debate on the ethical and policy conditions under which genetic databases can be established, kept, and used is dense with substantial controversy and uncertainty. In even

worse, it is sometimes unclear whether controversy and uncertainty are genuine disagreements based on a thoughtful understanding and reasoning about the ethical issues that are involved or whether they entail misunderstandings. Therefore, although the ethical issues that the collections of biological samples and genetic data raise have been debated for a long time, further empirical research and ethical discussion is needed.

[*] This is the English version of an excerpt from Alexandre Mauron. « Recherche en génomique: droits, intérêts et perspectives futures » in : Knoppers, B. and Nootens, S., *La recherche en génétique et en génomique : droits et responsabilités* (Montréal, 2005) (forthcoming).

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