

The new genomic patient

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Each genome, epigenome, metabolome, microbiome, etc. is, as we learn, an interactome. In the philosopher's imagination, the genome has lost its supremacy and at the same time won in significance by acknowledging its interrelatedness with those other parts and processes in the cell it once was thought to dominate. Dead is the "genetic programme" model of the genome. Long live the genome as an intricate information resource that successfully interacts with other components and processes of the cell and is used by the cell (rather than controlling it) in sometimes unforeseen and complicated ways, as they have emerged during evolution. In this present time, genetics has become even more interesting.

While this conceptual transformation in the natural sciences and in medicine unfolds [1], a different transformation takes place on the level of the social organisation of genomics. People, healthy and ill, become part of the research process. Hundreds of thousands of users of health-care systems join biobanks and allow researchers to use their genetic data and to put them into a relation with their 'phenotype'. These participants' phenotypes, however, are temporally extended, and is ultimately nothing else than their lived life. Of course, it is not their life in its whole complexity, which becomes interesting to the scientists; it is rather a selection: diseases, reactions to drugs, eating habits, behavioural particularities, biochemical data other than DNA sequences or single nucleotide polymorphisms, etc. From a superposition of different layers of data – genotypical, phenotypical and environmental – correlations and patterns emerge that can be used as hints leading to new hypotheses and discoveries. This new style of 'data driven' instead of 'hypotheses driven' research that characterises systems medicine only works with the involvement and participation of large numbers of people.

In the article "The Lausanne Institutional Biobank: A new resource to catalyse research in personalised medicine and pharmaceutical sciences" [2], one model for patient involvement is presented. Patients admitted to a large research hospital (the CHUV in Lausanne) have been systematically invited to provide general consent for the use of their biomedical data and samples for research, including a blood sample for DNA sequencing, and also to be recontacted for clinical trials. The acceptance rate is remarkable; over the first 18 months, 76% of patients contacted agreed. This model is highly significant as a proof of feasibility;

nonetheless, it also shows where the challenges of this new form of 'biosociality' (Paul Rabinow) are.

The style of research that we are talking about here is different from clinical studies such as drug trials. Participants are not asked to take part in a medical experiment. They are asked to allow researchers to use their data – genetic, non-genetic and medical – in order to feed a huge data resource that then will be managed by bioinformaticians and used by scientists. It can be exploited for many research purposes like good university libraries or archives can also be exploited for many different research purposes. The ethical issues are less the potential risks involved in the experiment but the confidentiality of data, which, since the genome is personal, can never be anonymised irreversibly and be perfectly safe. People, in principle, could always be tracked down, if somebody is not daunted by the effort that it needs. However, perhaps the confidentiality issue is not the key issue here, because only some information may prove sensitive for the research participants. Quite to the contrary, the disclosure rather than the containment may be the much more tricky issue to solve by good biobank governance i.e., the handling of health related "additional" and "incidental" findings. How much information feedback is fair to the participants and how should communication in both ways be organised?

The two basic ethical questions that are raised here are these: (1) Do researchers have a moral duty to tell the participant in case they find predictive genomic information that could be used by the participant to prevent a disease? If there should be such a duty, it is still to be decided whether it outweighs the patient's right not to know and how the patient can exert her or his right not to know without knowing its content. (2) Does the participant have a moral duty to know health related predictive genomic information that could be used to prevent a disease? If there should be such a duty, we need to clarify under which conditions it exists and on which ethical base it rests.

What is at stake here is the communication regime around biobanks. We need to understand the normative contents of and the values involved in the relationship between the researchers and the participants of biobanks. They might not be the same as in imaging studies, where, for instance, in a computed tomography scan of the brain a tumour could be discovered. This tumour then needs immediate treatment, it is a definite disease but the patient is in fact un-

aware of it, whereas the gene variants are not diseases, not even undiscovered germs for diseases. They are variants that might *lead* to a disease in the future – with a certain probability. The communication regime; therefore, also includes questions of participation. How much should the participants be able to shape the terms and conditions of disclosure, i.e., the rules of genetic transparency [3] with regard to their own lives? Additionally, how can they make their own ‘good’ decisions about which kind of information they want to have disclosed and which kind they want to ignore?

In systems medicine, therefore, large groups of people are becoming involved in this new type of research. Regular patients all become genomic patients. While ‘free and informed consent’ was the key for organising clinical studies ethically, here, new and appropriate modes of recognising the participants as subjects, i.e., new ways of participation and partnership with the participants need to be found. Informed consent will still be important, but as a stand-alone principle it is insufficient. The questions to be answered first appear on a meta level, where the terms and conditions of this genomic information partnership are negotiated. Several models are possible, ranging from no disclosure of research grade information, partial disclosure on demand, mandatory partial disclosure, complete disclosure on de-

mand, finally to participants’ open access to raw data [4]. – I hope for a broad and open debate to clarify which one is best in particular types of cases and in a particular society. *That* decision cannot be taken just from the therapeutic ethos of “doctor knows best”.

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