The human genome: metaphysical object and political icon

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Abstract

In recent years, the Human Genome Project has generated many questions on the ethical value of the human genome as such. Is there a right to an "non manipulated" genome? Should the genome be protected against what and in what sense? Is genetic diversity in humans a value to be treasured if so, what is its ethical relevance? What connection does it have (if any) with the value of genetic diversity in animals and plants? To dispel some of the bewilderment raised by these questions, one must address a set of more basic conceptual problems: how do we understand the relationship between the genome of a cell, the genome of a person and "the Human Genome"? What stakes can one legitimately have in one's genome? What stakes can humankind have in "the Human Genome"? In Switzerland, a specific and rather unique context is given to these debates because there is a special "genetics and reproductive technology" article in the Constitution, which specifies that gene technology must respect "the dignity of creatures". Not surprisingly, this article can be interpreted in contradictory ways, but it provides an interesting focal [...]

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1.4. The human genome: metaphysical object and political icon

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In recent years, the Human Genome Project has generated many questions on the ethical value of the human genome as such. Is there a right to an "non manipulated" genome? Should the genome be protected against what and in what sense? Is genetic diversity in humans a value to be treasured? If so, what is its ethical relevance? What connection does it have (if any) with the value of genetic diversity in animals and plants? To dispel some of the bewilderment raised by these questions, one must address a set of more basic conceptual problems: how do we understand the relationship between the genome of a cell, the genome of a person and "the Human Genome"? What stakes can one legitimately have in one's genome? What stakes can humankind have in "the Human Genome"?

In Switzerland, a specific and rather unique context is given to these debates because there is a special "genetics and reproductive technology" article in the Constitution, which specifies that gene technology must respect "the dignity of creatures". Not surprisingly, this article can be interpreted in contradictory ways, but it provides an interesting focal point for the dilemmas of "genomic metaphysics".

1. Introduction

The starting point of my presentation is a humorous drawing published earlier this year in a Swiss daily (although the cartoon is presumably of American origin). Several scientists in lab coats are gathered around a microscope. On the wall, there are diagrams of "the alcoholism gene", "the homosexuality gene", "the obesity gene". Another researcher rushes in through the door, gleaming with the joy of having made a major discovery: "I have discovered the gene that makes us believe that everything is in the genes!"

This drawing expresses with elegant precision my main message. This message is not that "everything is in the genes", but rather that in our present culture, genetic explanations have acquired the status of common-sense beliefs. It is my contention that the rise of what is usually called, somewhat misleadingly, "genetic reductionism" is closely connected to the rise of what I call the "genomic perspective". By this term, I mean a perspective on medicine and on human nature based on the centrality of the genome as the hard-core of the human essence and the repository of a person's identity, long-term traits and destiny. Today, the genome is much more than just a biological entity. In the words of the American sociologist Dorothy Nelkin, it has become a "cultural icon", a concept that underpins many ideas and dilemmas about health and disease, and more broadly, about what it is to be human, about the diversity of individual characteristics, issues of equality and inequality. The genome is not only an object of scientific investigation, it has become an ideological construct or even a metaphysical concept.

The initial step in this metamorphosis belongs to the history of molecular genetics. Since the nineteen-eighties, the Human Genome Project has added a new perspective in genetics in which the genome itself -rather than the genes or DNA- has become a distinct object of scientific investigation. It is significant that the term "genomics" was soon coined to specify a particular aspect of molecular genetics, namely the study and mapping of genomes, human and otherwise (This is also the title of a journal devoted to these studies, and historians of science know how useful titles of new journals are in tracking the twists and turns of scientific progress). Today, the genome of several organisms has been sequenced in full, and many more genomes have been mapped to a high level of detail. The Human Genome Project is well on its way to provide a
reasonably complete picture of the more important regions of our genome within the first few years of the next century. Furthermore, it does without strong central planning but relies on a multiplicity of co-ordinating bodies. The Project really stands for a flurry of variegated efforts that converge towards the goal of deciphering the human genome and these endeavours are difficult to demarcate precisely from the usual business of basic research in molecular genetics, although they are increasingly using specific, industrial-scale, methods. Yet it is fascinating to see how, in the public eye, the Human Genome Project retains the misleading image of an unified, deliberate Promethean effort to uncover the « Book of Man », the « Holy Grail » of the Genome, supposedly encapsulating the human essence.

2. The Genome and the Self

The increased emphasis on the genome as a whole has had major effect in the public perception of genetics and of its ethical aspects. To mention but one example, « genome talk » has partly substituted « genetics talk » in non-specialist discussions of these topics, especially in the German language. The new word Genomanalyse (genome-analysis) has been invented by lawyers and ethicists as an overarching concept including genetic diagnosis and screening, as well as their ethical and legal implications. This shift illustrates how basic notions of self and identity tend increasingly to be understood in genomic terms. The notion that the genome is the ontological hard-core of humanhood, the bearer of individual identity as well as that which makes a human being human, has become a common-sense idea and yet it is a largely unexamined belief.

This « genomic metaphysics » (1) has both an individual and a species-specific component. On the individual side, the popular notion that the genome contains genes for specific diseases, and also for obesity, homosexuality and the like, has fostered the notion that the genome is the repository of the most important and stable characteristics of an individual. In Aristotelian terms, one might say that the genome is the formal principle that makes a person what she is. As I have discussed elsewhere (2), this Aristotelian reference has to be taken fairly literally. For instance, it is highly significant that in debates on the ethical standing of human prenatal life, those who support the view that the embryo is a fully-fledged person ever since fertilisation, typically believe to be vindicated by the discoveries of modern biology: the fact that a new stable diploid genome arises at fertilisation and remains ever after the genome of that particular individual is taken as the true and definitive marker of personal identity and individuality, i.e. the character of an entity that is distinctive and separate from others (the problems with this view, as well as the relative character of biological individuality are discussed in (2)).

The main difficulty with this genomic metaphysics is that the genomic notion of identity that it entails presupposes a specific metaphysical framework: roughly speaking an essentialist understanding of identity. But this framework is not necessarily correct, nor is it supported by modern science. The latter is much more congruent to an alternative philosophical tradition going back to John Locke, in which identity is not grounded in any essential predicates that apply to an individual but in the continuity of certain contingent properties (such as specific mental states for example). In the article mentioned above, I have explained why I believe that biological data do not support an essentialist conception of individuality. If this analysis is correct, genomic metaphysics is not vindicated by current science, nor is it an appropriate framework for assessing the philosophical and ethical implications of genome research and genetics.

A similar hypothesis of the genome has occurred on the level of species. Because the notion of the Human Genome as Holy Grail and Sacred Book alluded to before, the collective genome of humankind becomes the defining principle of humanhood. To be human is to partake in the Human Genome and conversely, organisms having a human genome are, by definition, human. It is as though genomes had a special species-defining quality, a quality that not only marks genomes as wholes but also extends to their component single genes. In public debates on the ethics of gene technology, I have often noticed that people resent the introduction of human genes in, let us say, pigs, more than they would the transfer into pigs of genes from some other, non-human, organism. The « moral » transgression appears to be more serious because a little bit
the human essence would be mixed with the porcine essence and that would be an « unholy » hybridisation (I was asked several times the question how many human genes one has to reduce in the pig genome to make it human, a question that is revealing of the essentialist core of the popular species concept).

Again the problem here is that the underlying, popular, species concept is typological and essentialist. People without knowledge of evolutionary biology do not realise that this way of thinking about species has been abandoned ever since Darwin. The modern biological species concept is highly complex and partly controversial. But at its core, one finds the notion of interbreeding potential (ref. 3, chapter 1,6). « Species-belongingness » is a relationship determined by an individual’s capacity to produce fertile offspring within a population of a given species. This is a rational property that cannot be logically reduced to a list of essential properties, a reduction it would be basic to a typological, pre-Darwinian species concept (this is one reason why it has been suggested that species are logical individuals rather than logical classes (ref. 4, chapter 4)). Furthermore, reproductive isolation is necessary for a new species to arise and there are many mechanisms that can result in this reproductive barrier, from modifications in chromosomal structure to behavioural and ecological changes. Because of this pleiotropic nature of species barriers, there is no predefined amount of genetic change, valid across all organisms, that would be necessary and sufficient to give rise to a new species. This exemplifies how misleading it is to define the genome as a species determinant in the way that the Human Genome is increasingly being received as the Sacred Recipe to produce a human being. If one goes from the level of the genome to that of individual genes, the category mistake is even more obvious: there is nothing essentially human about the human insulin gene or the human alpha-haemoglobin gene. Human beings are contingently, not essentially human, and mixing them with the genome of a pig does not add the metaphysical implications that are popularly imputed to genetic engineering. If a human being successfully implements a necessary biological function in a pig and contributes to its life as earthly normal pig, that is all there is to it. Even though the pig hosts a bit of human DNA, no tule of humanity has heightened its porcine nature. In a certain sense, there is no such thing as a human gene.

An additional problem is the complex relationship between the Human Genome (i.e. the HGP will uncover), the gene pool (as understood by population geneticists), the genome of human individuals, and the actual genome present in a human cell. As I have discussed another earlier article (5), whenever defending the integrity of the genome is being proposed as ethical objective, there is no clear account of what is actually being protected, and against what it stands to be protected. Furthermore, there is a potential contradiction between protecting the Human Genome or the Gene Pool as compared to protecting the interests and rights of human individuals. Historically, the sacralisation of human nature as an overarching collective resource to be defended against individualistic rights and interests of persons has been a hallmark of classical eugenics (ref. 6, chapter 4). This shows that defending the integrity of the genome could conceivably work against personal rights and in itself be counterproductive as regards the ethical values that most of us agree must be upheld in this field of genetics. More recently, the discovery by the media (if not by historians) of the active programme of sterilisation of the « unfit » in Sweden has highlighted the collectivist and individualistic ethos that has always been typical of eugenic enterprises everywhere. Conversely, it suggests that in order to mount an effective anti-eugenic ethical case, one must go at it on an uncompromising defence of individual rights, not merely on an abstract consideration of the common good of groups, societies or genomes.

What are the ethical stakes?

We have seen that, for current conventional wisdom, the genome has come to be derstood as the formal principle underpinning personal identity as well as species identity. In effect, one could say that the genome has become the secular equivalent of the soul. This shift in teleological representations would merely be a curiosity for historians of mentalities if it weren’t for the strong influence it has on ethical discussions on genetics. Because of it, ethical attention
has occasionally moved from the scrutiny of concrete practices (gene therapy, genetic diagnosis, screening, genetic discrimination etc.) to an attempt to define human rights vested in the Human Genome, or the genomes of humans. This move is exemplified by two declarations of international importance, the oldest of which is Recommendation 934 of the Parliamentary Assembly of the Council of Europe that asserted in 1982 « a right to inherit a genetic pattern that has not been artificially changed », although it also foresees exceptions for bona fide therapeutic purposes (7). The second is the International Declaration on the Human Genome and Human Rights that has been discussed by UNESCO’s International Bioethics Committee for several years (8). This text states that the human genome is the common heritage of humanity and makes the genome the basis for the unity of human kind and the dignity of persons. It then goes on to specify various rights and principles for future legislation which on the whole are quite well-taken. The basic problem is that the connection is not obvious between these quite reasonable directives on one hand, and the initial solemn pronouncements on the genome as common heritage on the other. The Committee has reacted thoughtfully to initial criticisms. For instance, it has included a statement that « The human genome, which by nature evolves, is subject to mutations ». Presumably, the idea here is to avoid giving the mistaken impression that the genome is taken as a kind of fixed and eternal benchmark of humankind. The Committee has also changed the title by adding an explicit reference to Human Rights, and implemented other changes that aim at making it clearer that individual rights are to be protected rather than jeopardised by the Declaration. Interestingly, it has resisted suggestions to make human genetic diversity a value to be protected, correctly stating that it simply represents an empirical fact, not a valuable resource per se in the same way as the biodiversity of animal and plant species would be. These changes have gone a long way towards the right direction. However, the basic problem still remains, namely that no logical link can be discerned between what is in effect a series of "factual" statements (the genome is this or that) and the normative statements that follow, which, as basic human rights, could stand « on their own feet ». In my opinion, the initial genomic statements are still basically redundant, and liable to elicit grave misunderstandings.

In addition, a focus on the integrity of the genome could be distracting attention from the more pressing ethical dilemmas in genetics, which lie in the field of predictive genetic testing rather than genome modification (such as gene therapy). In genetic analysis, one does not touch the genome, one only looks at it, as it were: and yet this is where the most contentious issues arise. Progress in genetic diagnostics is far quicker than in therapy and we are already witnessing conflicts of values both at the individual level (right to know/not to know, sharing genetic data in families, invasive prevention for high-risk patients) and the collective level (the role of commercial pressure in implementing genetic tests in medicine, genetic discrimination in employment and insurance) (9, 10). It is true that the public is more sensitive to issues involving « gene manipulation », possibly because it evokes deeply entrenched cultural icons : Faust, Frankenstein, the Sorcerer’s Apprentice and his bumbling intervention in the natural order. However, it is the role of international bioethical debates to prioritise our anxieties and to show which problems lie far in the future and which ones are already with us and need our immediate attention. If leading bodies in bioethics simply went along public instincts in this matter, they would in effect be failing their political and didactic duties.

4. Politics and the genomic mystique : the Swiss case

These may seem rather theoretical and far-fetched issues and yet the have received a highly political focus in Switzerland. In 1992, the Swiss people voted in a nation-wide referendum in favour of a Constitutional amendment (article 24novies, Federal Constitution) that asserts a wide range a legal principles as regards genetic engineering, human genetics and reproductive technologies. Especially notable are the following provisions:

a) interventions affecting the genetic heritage of human gametes and embryos shall not be admissible;
b) the germ-cell and genetic heritage of non-human species may not be transferred to the human germ-cell, nor fixed with it;
§3 The Confederation shall issue provisions concerning the use of germ-cells and of the genetic heritage of animals, plants and other organisms. It shall thereby take into account the dignity of creatures and the safety of humankind, of animals and of the environment. It shall also protect the genetic diversity of animal and plant species.

The language of genomics metaphysics is pervasive in this text. For instance, it could have simply and straightforwardly prohibited the production of chimeras involving human material. Instead, it chose to outlaw the mixing of human and non-human « genetic heritage »: clearly, the human genome is valued as such and protected against the quasi-religious transgression of its purity. §3 is notable as it introduces the notion of « dignity of creatures ». For the first time, the concept of dignity is applied in the Constitution to non-human entities, a move that was welcomed by « biocentric » ethicists that are currently in fashion in Central Europe but which I find unfortunate and dangerous for our humanistic tradition of ethics and political philosophy. Also significant is the context in which this notion appears in the Constitution, i.e. genetic engineering. This more or less suggests that the dignity of creatures is somehow more at risk through genetic engineering than through, say, industrial farming or the transportation of farm animals in inhumane conditions. In other words, it leads rather seductively to the idea that dignity of animals is basically connected with their genome rather than with their welfare or their protection from harm and suffering.

Needless to say, nobody is quite sure what the « dignity of creatures » is all about and this notion has been debated ever since. Philosophers and theologians have tried to flesh out this concept (11, 12), as have bioethical bodies such as the Joint Commission for Animal Experimentation of the Swiss Academies of Medicine and of Science (13). Civil servants are trying to figure out how the new dignity principle might inspire future legislation on genetic engineering. But given the influence of a vulgarised Naturphilosophie in the dominant culture of German-speaking peoples, one often encounters the idea that interfering with the genome affects the dignity of organisms and is therefore ethically suspect. Not surprisingly, a coalition of environmentalists and other opponents of gene technology have recently campaigned for a ban on most of genetic engineering. In the Swiss system of direct democracy, any group of citizens can cause a constitutional amendment to be submitted to a popular referendum if they manage to collect the requisite number of citizen’s signatures. This has happened as regards genetic engineering, so that the Swiss people will decide next summer on a constitutional amendment that would contain the following prohibitions:

- the production and use of transgenic animals is absolutely forbidden (even for purely research purposes);
- release of any genetically-modified organisms is absolutely forbidden;
- patents on organisms, products and processes in animal and plant genetic engineering are outlawed (for information on this upcoming referendum and the current stand of the debate, see (14) and links therefrom).

Considering the importance of biological research (both academic and industrial) in Switzerland, the adoption of this amendment would have truly cataclysmic consequences and would constitute a momentous turning point in the country’s history. There is little doubt that it would usher in a major political crisis. What is significant as regards my present topic is that the proponents of this amendment present it as a logical extension of the present article 24movies. They contend that they simply want to implement in more concrete terms the directive already contained in the Constitution as regards the dignity of non-human organisms. This shows how pervasive the sacralisation of the genome has become and how powerful an influence it represents in shaping public perceptions of genetics and political attitudes towards it.

E.T. Juengst has noted that these sorts of debates move through three stages: romantic, precise, generalising (15). The genomic perspective tends to pin down debate at the initial romantic stage and inhibit a more precise, pragmatic and focused discussion of the issues. In this sense, the Swiss debate illustrates a specific form of intellectual disability, namely the inability to move away from the romantic questions such as « Are we playing God? », « Is it permissible to
modify human nature?», towards more down-to-earth questions such as: «how are we to protect patients’ rights in gene therapy experiments?» or «what are the rights of clients of life-insurance companies as regards the privacy of genetic data?». Yet at the end of the day, these are the questions that really matter. They are the issues that make a difference to the rights and interests of real people, rather than abstract entities such as communities, societies and gene pools.

5. Conclusion: ethics is primarily about human persons

The Human Genome project is an epoch-making project of immense promise and historical significance. Furthermore, there is no denying that it has brought a great deal of interest and resources to the study and debate of many ethical issues linked to human genetics. This is as it should be and my criticism is only directed at one particular side effect of these efforts, namely the sacralisation of the genome. This I believe to be an unfortunate ethical debate because it side-tracks ethical debate away from pragmatic and potentially resolvable problems and moves it towards mythologising genetics, fostering quasi-religious fears and fuelling anti-science demagoguery, as the Swiss situation eloquently shows. The human person, not genomes, nor «creatures» large and small, should be at the centre of ethical reflection.

References