

# Human genome and philosophy: What ethical challenge will human genome studies bring to the medical practices in the 21<sup>st</sup> century?

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**Abstract** – A human being or person cannot be reduced to a set of human genes, or human genome. Genetic essentialism is wrong, because as a person the entity should have self-conscious and social interaction capacity which is grown in an interpersonal relationship. Genetic determinism is wrong too, the relationship between a gene and a trait is not a linear model of causation, but rather a non-linear one. Human genome is a complexity system and functions in a complexity system of human body and a complexity of systems of natural/social environment. Genetic determinism also caused the issue of how much responsibility an agent should take for her/his action, and how much degrees of freedom will a human being have. Human genome research caused several conceptual issues. Can we call a gene ‘good’ or ‘bad’, ‘superior’ or ‘inferior’? Is a boy who is detected to have the gene of Huntington’s chorea or Alzheimer disease a patient? What should the term ‘eugenics’ mean? What do the terms such as ‘gene therapy’, ‘treatment’ and ‘enhancement’ and ‘human cloning’ mean etc.? The research of human genome and its application caused and will cause ethical issues. Can human genome research and its application be used for eugenics, or only for the treatment and prevention of diseases? Must the principle of informed consent/choice be insisted in human genome research and its application? How to protecting gene privacy and combating the discrimination on the basis of genes? How to promote the quality between persons, harmony between ethnic groups and peace between countries? How to establish a fair, just, equal and equitable relationship between developing and developed countries in regarding to human genome research and its application? © 2001 Académie des Sciences/Éditions scientifiques et médicales Elsevier SAS

human genome project / genomics / genetic essentialism / genetic determinism / eugenics / informed consent / informed choice / gene privacy / gene discrimination / benefit-sharing

## **Résumé – Génome humain et philosophie : quels défis éthiques vont engendrer les études du génome humain dans l’exercice de la médecine au XXI<sup>e</sup> siècle ?**

Il est impossible de réduire l’homme ou une personne à sa seule dimension génomique ou à ses gènes. L’essentialisme génétique est une erreur. L’entité concernée, en tant que personne, doit, en effet, disposer d’une conscience et d’une capacité d’interactions sociales qui se développent dans le jeu des relations humaines. L’idée de déterminisme génétique est également fautive, car la relation existant entre un gène et un trait phénotypique ne répond pas à un modèle linéaire de causalité, mais s’inscrit bien plus dans la complexité. Le génome humain doit être compris comme un système complexe

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fonctionnant au sein d'un système de complexité d'ordre supérieur qu'est le corps humain, et dans la complexité des systèmes qui régissent la nature et la société. La notion de déterminisme génétique implique aussi celle de responsabilité d'un individu vis à vis de ses actes et celle de la liberté intrinsèque à la condition humaine. La recherche sur le génome pose plusieurs problèmes conceptuels. Peut-on parler de bon ou mauvais gènes, de gènes supérieurs ou de gènes inférieurs ? Doit-on considérer un enfant porteur du gène de la chorée de Huntington ou du gène de la maladie d'Alzheimer comme un patient ? Que devrait signifier le mot eugénisme ? Quel est le sens des termes thérapie génique, traitement, stimulation d'expression et clonage humain ? La recherche sur le génome humain et ses applications sont et seront source de questions éthiques. Peut-on les utiliser dans une perspective eugénique ou doit-on les réserver à des fins thérapeutiques et préventives ? Doit-on poser avec force les principes du choix et du consentement éclairés dans les domaines de la recherche sur le génome humain et de ses applications ? Comment protéger l'intimité génétique et combattre une discrimination fondée sur les gènes ? Comment promouvoir des rapports de qualité entre les personnes, l'harmonie entre les groupes ethniques et la paix entre les pays ? Comment établir des relations raisonnables, justes et équitables entre les pays développés et les pays en voie de développement en ce qui concerne la recherche sur le génome humain et ses applications ? © 2001 Académie des Sciences/Éditions scientifiques et médicales Elsevier SAS

**projet génome humain / génomique / essentialisme génétique / déterminisme génétique / eugénisme / consentement éclairé / choix informé / intimité génétique / discrimination génétique / partage de bénéfice**

## 1. Introduction

The greatest scientific achievement in the 20th century is that the secret of the atomic nucleus on the one hand and cell nucleus on the other began to be unveiled. Since the discovery of DNA structure made by Watson and Crick, great progress in genetics has been made. Now a cross-century worldwide project is going on. It may be the greatest in size and the longest time-consuming scientific project in human history. That is Human Genome Project (HGP). The success of mapping and sequencing of human genome opens up a wide range of prospects for the use of genetic knowledge to benefit all people in the world. A revolution in medicine may happen in the 21st century: medicine will become a 'root' treated (not merely symptom focused), predictive and really prevention oriented medicine. Meanwhile, a lot of philosophical issues have been raised and debated.

## 2. Genetic essentialism and determinism

The philosophical issues that are involved in human genomics and its application include metaphysical, conceptual and ethical problems. The first metaphysical problem is whether a human being or person can be reduced to a set of human genes, or human genome, or whether a human genome constitutes the essence of a person.

In the West, there is a long tradition to think of 'being', and hence the 'person' in terms of 'substance'. The conception of substance as the primary instance of real being, and hence the person can be traced back to the thoughts of Aristotle. As a primary instance of real being, this power to exist in itself as an ultimate subject of action and attribu-

tion and not as a part of any other being is perhaps the most important thing one can say about substance. Without the 'in itselfness' of the substance, every instance of real being can be a part of, or inhering in, some other being, then the necessary fulfilling conditions for any being to exist would be infinitely deferred and theoretically never fulfilled. Substance with its 'in itself' characteristic is necessary for making a being to be called an identity. Philosophical circles of the modern West have largely retained this 'substantialist' tradition. There are those who define human personhood in terms of the body, genome or the development of the human brain. René Descartes (1596-1650), the 'father of modern thinking', the 'self' as "*cogito ergo sum*" – "I think therefore I am". For many modern people, "my genome constitutes what I am", or in other words, the essence of a human being is her/his genome. This concept of personhood is called genetic essentialism.

Genetic essentialism is fundamentally deficient in its description of the human nature. We have to take in account the social and relational basis of personhood. Traditional Chinese philosophy recognizes that human life is made up of two essential components: '*xing*' or '*ti*' (body, form) and '*shen*' (psyche, spirit) and is concerned with the way that they exist in a harmonious psychosomatic unity. And in the Confucian holistic concept of personhood it is emphasized that moral character of a person has been perfected. Since no one is born as a sage, to become a sage means that the person is a being always in the process of becoming. This implies that all the three aspects of the person: the psyche (*shen*), the vital force (*qi*), and the physical body (*ti*) are imperfect, waiting to be perfected. The emphasis of the physical body and the

understanding of the human person as a psychosomatic unity points to the next characterization of the human person as a social and relational being. For Confucianists human beings are distinguishable from animals because of their relational/social capacity. In the Confucian tradition, a person is never seen as an isolated individual but is always conceived of as a part of a network of relations. Even though it may seem to many that the bulk of Confucian ethical teaching pertains to the self-cultivation of an individual person to become a sage, one must not lose sight of the fact that the whole process is carried out in and through the social context and for the purpose of fulfilling social responsibility rather than self-actualization per se. The Chinese understanding of self-cultivation is never on the establishment of the independence or individualisation of the subject, but on the promotion and maintenance of the collective harmony of the community. To actualize man's nature is to fulfill man's human relatedness; a person is always a 'person-in-relations'. So the ability to have human relationships is what differentiates human persons from animals, and what is crucial for a human person is to be self-consciously aware of 'a keen insight' in human relationships. In Confucianism, relations are of the ontological category, in that they both constitute and complete personhood. [1] So a person cannot be reduced to her/his genome, as pointed out by a Jewish scholar: "If I am only my genes, what am I?" [2].

In close relationship with genetic essentialism some people claim that all human traits, diseases, mental abilities, behaviors and characters are determined by human genome. It is labelled genetic determinism. Any action of using genetic knowledge is predicated on the presumptions of these dichotomies, such as nature vs. nurture, biological determinism vs. social determinism etc. With the development of human genetics and the success of genome mapping project, more weight seems to be put on the genetic side of the balance, and revives ancient debates. People are talking about selfish genes, aggressive genes, altruist genes, homosexual genes, gambling-maniac genes, alcoholic genes, crime genes, improvement of human species etc. American philosopher R. Nozick imagined that the future parents could buy the genes that codify the traits of their child which they want [3]. These talks imply a theoretical presumption that everything of a human being is determined by genes, and leaves no room for environment and individual's free will. However, except a few human diseases and traits, many of them are the result of interaction between multi-genes and environment. For some of them, such as mentality, the role of socio-cultural environment cannot be ignored. Although the genes which cause the onset of some cancers have been known, nobody can deny the role of the human behaviors or environmental factors play in it, such as smoking, radiation and cancerogenic chemicals.

Genetic determinism also leads to medical solution of social problems. If all human behaviors are predetermined by genes, then not only at risk behaviors but also unethical and illegal actions can be explained by abnormal genes

and can be corrected by gene therapy, but not by health education, moral education or correction institutes. It will also raise some ethical and legal issues on whether offenders should be responsible or accountable for their unethical or illegal actions, because they have no choice by their free will. Genetic determinism in general, radical version in particular, will produce over-expectation to genetic knowledge and over-ambitious genetic program, and in turn make genetics be discredited as eugenics has been.

Underlying this theoretical presumption is a linear model of causation. One of the characteristics of the linear model of causation is that it assumes that an effect is caused by one factor, called monofactorial theory. It ignores downward, upward and lateral causation. The other is that in this model there is only one way causation: cause is always cause, and effect is always effect. And the linear causation model is predicated on a conception that human genome is merely a simple system. However, human genome is better to be viewed as a complexity system. So the causal relationship between a gene and its expression, or between genotype and phenotype is much more complex that it cannot be grasped by this simple linear model of causation. That a human being has a certain gene means he/she has the predisposition to have a trait which the gene leads to. The predispositional property is expressed only in an adequate context: a fragile grass is broken only when it is dropped on the ground. Not alone those complicated traits such as disease, mentality, behavior and character. Perhaps, we need an alternative model of causation, for example, a web model of causation that has been worked out by the *zangfu* theory of traditional Chinese medicine. In this non-linear model of causation all causes from different directions and multiple levels are considered, and an effect may be caused by a cause but in turn it can become a cause of other effect or even of the cause in a chain of events. There are so many variables that are involved in the expression of a gene that there may be no determinism at all, each of them make its contribution to the expression of a gene (R. Wachbroit). Actually, we are faced with three complexity systems: human genome, human body and environment. Human genome is a complexity system and functions in a complexity system of human body and a complexity system of natural/social environment. Any action we take on human genome may cause an unexpected consequence somewhere, as the so-called butterfly effect shows. In some cases an apparent linear causation may be viewed as a special case of this non-linear model of causation.

### 3. Conceptual issues in medicine

Some conceptual issues have been raised in human genome research and its possible application. Human genome research seems to challenge some well-received basic concepts of medicine, such as health, disease, patient, therapy or treatment, eugenics and others.

First, if a boy is detected to have the gene of Huntington's chorea or of Alzheimer disease, can we still say the

boy is healthy, he does not suffer disease or we should say he is ill or he is a patient? If we say he is ill or a patient, it seems not convincing, because there is no symptom then, those diseases will be set on in the age of 40 or 70 years. If we still say he is as healthy as other boys who have no such disease causing gene, it is not convincing too, because a disease causing gene is in his body. It seems to me that we have to find some term to describe the condition which is in between health and disease/illness.

Secondly, now we use the term genetic therapy to refer both the genetic interventions that are aimed at the cure of a disease or relief of some symptom and those that are aimed at changing a trait without connection to disease. The term therapy means “the treatment of disease or of any physical or mental disorder by medical or physical means, usually excluding surgery.” (*Webster’s New World Dictionary*). However, any sense of enhancement, even the enhancement of immunity is not therapy in the sense mentioned above.

Thirdly, the various meanings of the term eugenics still caused confusions and debates. When Chinese geneticists supported ‘yousheng’ (well born and well bear), it was taken as an evidence that they supported the eugenics practiced in Nazi Germany. When China’s law was unfairly criticized as eugenic because there is some article that could be understood as a program imposed by the state, those laws with explicit eugenics in other countries are immune from this criticism. In some Western media or even some professional journals the practice for meeting the parents’ wish to have a healthy child was also charged as eugenics. Although the 18th International Congress of Genetics in 1998 reached the consensus in which a suggestion to not use the term eugenics in scientific literature was made, but the *American Journal of Human Genetics* still published a hostile article to attack Chinese geneticists. The author of the article manipulated the figure in an international survey on ethics and genetics, and made fraud to cheat readers, but the editor never sent the article to authoritative scholars to be reviewed [4].

Finally, can we call a gene good or bad, superior or inferior? Some gene may cause disease, then we call it bad gene or inferior gene. However, even this disease causing gene may protect the body from other disease. So in one context it is a disease causing gene, and in another context it becomes a disease preventing gene. Moreover, the term bad gene or inferior gene may cause genetic discrimination. So it should handle very careful to call a gene bad or inferior. And we have less reason to call such genes bad or inferior that cause short length or colour skin, as we should call those that cause tall length or blonde as good or superior just because they are undesirable to some people.

#### 4. Some ethical issues related to human genome

Because of the limit of length, this paper cannot discuss all ethical issues that are involved in human genome

research and its application. Instead, the paper will be focused on the following issues, such as: what is the purpose of the application of human genome knowledge for the treatment or prevention of diseases or for eugenics? Is the principle of informed consent/informed choice indispensable in human genome research and its application? How to deal with the access to genetic information and the discrimination on the basis of genome, and how to deal with the relationships between persons, ethnic groups and countries human genome research and its application? Recently the Ethical, Legal & Social Issues committee (ELSI) adopted a statement in which it announced that the committee reached the following consensus:

- the research on human genome and its application should be focused on the treatment and prevention of diseases, but never on eugenics;
- in the research on human genome and its application, the principle of informed consent or informed choice should be adhered persistently;
- in the research on human genome and its application, the privacy of individual genome should be protected, and the discrimination on the basis of genome should be combated;
- in the research on human genome and its application, efforts should be made to promote the equality between persons, harmony between ethnic groups, and peace between countries.

#### 5. Treatment/prevention of diseases/eugenics

It was claimed that the new genetics (on the basis of human genomic research) can improve human race by eliminating inferior traits and enhancing superior ones, or make ‘supermen’. These talks remind us of the atrocities in Nazi Germany. Of course, those who talk about eugenics are not Nazis or fascists, but they don’t know that there is a slippery slope along which people may slide into immoral abyss. Eugenic programme is wrong both scientifically and morally. It is scientifically wrong because it is impossible for any society or any country to hold back or prevent the birth and existence of disabled since there are recessive genes and natural mutation of genes is unavoidable. It is morally wrong because disabled and non-disabled are equal, they enjoy equal rights, there is equality between people in same ethnic group and between different ethnic groups, nobody can be said inferior or superior. The marriage or procreation of anybody is a voluntary choice based on informed, rational decision, and cannot be decided by any other people. Any society or country has no right to take disabled or people who suffer from genetic disease as a burden that want be got rid of, instead, it has obligation and responsibility to develop their potentials, and make efforts to enable them to live as good as other people.

It is tempting for scientists to use gene therapy for the enhancement of human traits. But it does not raise less

issues. First, which kind of human traits should be enhanced, it will be a problem to which there will be no agreement among human beings. Second, when we reach an agreement on which kind of human traits should be enhanced, it may not be avoidable that people with the opposite and undesirable traits which are not to be enhanced will be stigmatized or discriminated. Third, there is no guarantee that the enhancement of human traits will not lead to eugenic practices such as in Nazi Germany [5].

So it should be emphasized that the human genome research and its application should be focused on the treatment and prevention of diseases, and the improvement of the quality of life.

## 6. Informed consent/informed choice

Informed consent/choice is a central principle of Nuremberg code which is based on the historical lessons of human experimentation in Nazi Germany. Unfortunately, those who initiated and drafted Nuremberg code ignored the same lessons of possibly more cruel and inhumane human experimentation done by Japanese doctors in 731 Team. And many Chinese doctors did not know much about Nuremberg code. In a Confucian community with paternalistic tradition both in society and medicine individual autonomy is easily not paid attention to. In my opinion, although medical decision is made by the family as routine in Chinese community, it is helpful to make a more reasonable and considerate decision, but individual preference should still be seriously considered, and incorporated into family decision. A Chinese geneticist was wrong when he said that informed consent needs not be taken seriously, we can sign the form for the subjects. Such a geneticist is not qualified to do research on human subjects. Some regulations laid down by provincial legislatures like “Regulations on limiting inferior birth” in Liaoning province or “Regulations prohibiting the reproduction of seriously mentally retarded” in Gansu province openly violate the principle of informed consent.

## 7. Protecting gene privacy and combating gene discrimination

The reality is, we live in a society full of various discriminations, including gender discrimination, age discrimination, discrimination against homosexuals. The discrimination on the basis of traits exists too, such as people with short length are looked down on. Once these not favoured traits are known relevant to genes, these discriminations will be the ones on the basis of genes, or gene discriminations. When we apply the results of human genome research for eugenics, no matter it is genetic intervention for enhancing traits or capabilities in somatic cells and germ line cells, or ‘Sperm bank from famous persons’ or ‘Egg bank from models’, it will cause the discriminations

against those with the traits that are not enhanced. Apart from that we should emphasize it is wrong to label some genes good, superior, others bad, inferior, one precautionary measure is protecting the confidentiality of individual genome that is the core privacy of individuals. It needs to draft the law to prevent and combat gene discriminations and to protect gene privacy.

## 8. Equality between persons and ethnic groups, peace

We should insist that persons within a nation as well as persons between different ethnic groups are equal. Human genome diversity project shows that “we are all brothers and sisters in the four seas”, and that the difference between whites, yellows and blacks is only on skin. In the human genome research all information should be shared by all countries and all peoples. The developing countries/communities should not be taken as cheap providers of DNA samples. The fair, just, equal and equitable relationship should be established between developed and developing countries. It cannot be denied that there are some cases which can be called gene-piracy or gene-colonialism.

– Case 1:

US researchers study genetics of nicotine dependence in an isolated village in China. The funding agency, the US National Institutes of Health, required, in the request for applications (RFA), as a condition for awarding the grant, that researchers bank their samples permanently and make the samples available, on request and without charge, to other researchers, including commercial entities, though without providing individuals’ names. The RFA also required that information from the samples be put on a website. The informed consents, as required by the RFA, specified that the samples could in the future be used by commercial companies. The researcher who took the original samples was an academic, basic researcher, who found several promising markers for nicotine dependence. Subsequently, a pharmaceutical company, using the researcher’s samples, found ‘the gene’ and ten years later (pretty fast) developed an immensely profitable drug, after further research in the US and US-based clinical trials. Does the company owe anything to the Chinese village? The company argues that its finding was based on a chain of research, of which the original Chinese samples were only a part. The company also argues that the first group of researchers should have paid the community.

– Case 2:

Some geneticists in a Japanese university with the help of a Chinese graduate student went to Yunnan province disguised as tourists, and collected blood samples from a non-Han ethnic group without informed consent and benefit in return.

– Case 3:

Some US people went to a non-Han village in Yunnan province directly without the consent from the community

and collected blood samples at expense of US \$ 1 for one sample without informed consent.

– Case 4:

A US institute made a contract with the Institute of Nutrition in Beijing which is not qualified in genetic research. In the contract the Institute of Nutrition was required to provide half million Chinese blood samples to the US counterpart in exchange of money.

These cases show that there may be unequal, unfair and inequitable relationship between people, communities and institutes of developed countries and those of developing countries. The developing countries may be only the cheap providers of blood samples and the benefits are not received in return by the people who provided the sample and the community of which he/she is a member. In view of these cases, Chinese scientists and geneticists helped the government to draft “Interim measures for protecting human genetic resources” the underlying presumption and principles of which is: (1) Every country has the responsibility for protecting its own genetic resources; (2) the relation between both sides of cooperation should

be equal; (3) the benefits brought by the research should be shared by both sides; and (4) the informed consent is mandatory.

To establish a fair, just, equal and equitable relationship between developed and developing countries in regard to human genome research it is necessary to recognize the principle of benefit-sharing. A benefit is good if it contributes to the well-being of an individual and/or a given community (e.g. by region, tribe, disease-group...). Benefits transcend avoidance of harm (non-maleficence) in so far as they promote the welfare of an individual and/or of a community. Thus, a benefit is not identical with profit in the monetary or economic sense. Determining a benefit depends on needs, values, priorities and cultural expectations. The ethical justification of benefit-sharing can appeal to the ethical principle of beneficence, distributive and retributive justice and solidarity. I hope that the fair, just, equal and equitable relationship will be established between France and China in the field of human genome research.

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