



CNECV – NATIONAL COUNCIL OF ETHICS FOR THE LIFE SCIENCES

**DOCUMENT ON THE HUMAN GENOME
(31/CNECV/2000)**

English version: José Henrique Neto

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INTRODUCTION

The human genome has been big news these past few weeks. It was publicised worldwide that, after nearly 10 years of laborious and highly expensive research, participated by several countries, a near-complete sequence of the human genome has been established. This achievement opens the way to relevant scientific and technical advances, but it will also raise new ethical problems.

The *National Council of Ethics for the Life Sciences* (CNECV) foresaw long ago the problems that would arise from this discovery, wherefore it organised in 1997 a public Seminar entitled "The Powers and Limits of Genetics". This Seminar brought together fifteen national and foreign specialists, to dwell on the issues that emerge from the analysis of the human genome. The Council has published the Proceedings of this Seminar².

The CNECV is preparing now an updated Report and Opinion on the matter. Since such work will necessarily take some time to complete, the CNECV decided that the following notes, a brief and preliminary working document, should be made available for immediate use.

The National Council of Ethics for the Life Sciences

Lisbon, the 12th of July 2000.

Prof. **Luís Archer**
President of the National Council of Ethics
for the Life Sciences

¹ English version: José Henrique Neto

² CONSELHO NACIONAL DE ÉTICA PARA AS CIÊNCIAS DA VIDA (CNECV), *Poderes e Limites da Genética, Actas do IV Seminário (1997)*, Presidência do Conselho de Ministros, Lisboa, 1998.



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I – SCIENTIFIC SIGNIFICANCE

What, then, has been discovered? Man's innermost chemistry has been revealed – the 50 or 100 thousand genes found in each human cell that largely determine the course of its life and diseases. Still, all that we possess in concrete terms, for now, is a sequence of some 3,000,000,000 characters (each representing one chemical compound), without punctuation or paragraphs, or even separation between words. Only now may begin the systematic work (already done in a few isolated cases) of separating words, sentences and paragraphs, and of translating through complex experiments the meaning of each word – which proteins each produces and how these proteins act on the organism. It will take several decades to complete these tasks.

One of the uses of this discovery is that it provides a solid basis on which to begin the long work of establishing in full detail the genetic errors that cause many infirmities. Once we are in possession of such data, we shall be able to identify many more individuals who bear a predisposition to diverse common diseases (which nevertheless will develop only under certain circumstances). We will also be able to predict, decades ahead, the occurrence of monogenic late-onset diseases, some of which are still incurable.

There is hope that this knowledge will enable us one day to cure such infirmities, by several means, including the introduction into the organism of therapeutic genes (gene therapy). In the case of late-onset diseases, it will be possible eventually to perform such gene therapy preventively, that is to say, before the disease develops. Another way to effect prevention, in cases where the genes predisposing to a specific disease have been identified, is to create environmental, lifestyle and monitoring conditions that will prevent the predisposition from becoming actual infirmity.

Knowledge of the genome is also useful in that it makes it possible to produce, with the help of computer-aided biotechniques, personalised medication. As an example, people affected by high blood pressure, for instance, will be prescribed different medication, according to the different genes that, in each person, are involved in the particular affection. This search for personalised medicines is now referred to as "pharmacogenetics". The July 2000 edition of *Scientific American* estimates that, by the year 2005, pharmacogenetics may command an 800 million-dollar market. Among the advantages to the patient is a reduction in medication overload, which for some patients is ineffective and may cause adverse, toxic or allergic reactions. The fact that the highly reputed *Nature* has dedicated its edition of June 15, 2000, to this theme attests to the importance of pharmacogenetics and pharmacogenomics in the present panorama of Biology.

That is not all. Genomics is already taken for granted. Now the attention of the industry is turning to what is being dubbed "proteomics". What is really interesting, it turns out, are not the genes but the proteins they produce. Highly sophisticated techniques are being used to characterise in a short time the structural shapes of the proteins and the way they interact, in order to speed up the design and construction of medication able to either activate or block natural proteins, as the case may require.



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II – SOME GENERAL ETHICAL PERPLEXITIES

1. The foremost danger is to presume that, in Man, genes are everything. To believe that knowledge of the genome will yield the complete cartography of our temperament and character; that the genome alone explains intelligence, love, religious belief, crime and anti-social behaviour; that Man is fatally programmed by his genome; and that freedom and guilt are delusions of the pre-genetics era. All this is tantamount to dangerous ignorance of scientific data, and may lead to fatal discrimination. The human person and human dignity transcend the genes.

2. Without the protection of patents, the large-scale industries would not invest in research on the human genome, and that would obstruct advances in medicine. Yet, if patents are granted to any and every sequence of the human genome, the spirit of patents will be perverted and the scientific community will rise in protest, because this would affect the fundamental right to freedom of information and research. The issue is complex. It has been studied in Opinion 7/CNE/94, on *The Juridical Protection of Biotechnological Inventions*³, and Opinion 18/CNECV/97, on *The European Parliament's and Council of the European Union's Draft Directive for the Juridical Protection of Biotechnological Inventions* (COM (95) 661, 13 Dec. 95)⁴, whose Rapporteur was Dr. Paula Martinho da Silva.

The fact is that, by and by, Science is losing its innocence regarding the candid search for truth and good, and letting itself be seduced by economic power. Not accidentally, the special feature on the human genome in the July 2000 edition of *Scientific American* is entitled *The Human Genome Business* – it describes the tug-of-war between the large economic powers. In fact, the human genome has also become a *business*.

Even more seriously, the commercialisation of genetic tests leads to their being performed without the indispensable genetic counselling and psychosocial support, and this may result in grievous consequences to the individual.

3. The discovery of the genome feeds humankind's old dream of the perfect child, bearing a genome without blemish or imperfection; the dream of being able to choose every trait in our children, of creating a new Man, pre-programmed and risk-free. This dream is not only scientifically utopian but also an attempt against human dignity. Life would no longer be generated, it would be produced: parents and children would no longer relate as people would but as producer and product. There would be no more expectancy in the face of chance, nor the alternatives of freedom.

4. There is a group of other questions relating to the problem of who will be entitled to have access to genetic testing and to test results. Informed consent, the preservation of privacy and the right not to know one's own genetic traits must all be respected. Some questions, usually answered in the negative, remain controversial. Thus: is it to be accepted lightly and indiscriminately that a youngster should undergo a test designed to predict a late-onset disease that is presently incurable? Is it permissible, in every case, that parents may demand such a predictive test for their minor child? What must one do if an individual does not wish to communicate his/her positive test result to those of his/her relatives who need

³ CNECV, *Documentação*, Vol. II (1993-1994), Presidência do Conselho de Ministros, Lisboa, 1995, pp. 97-144 (the English version was published in Special Reprint VII, 1994).

⁴ CNECV, *Documentação*, Vol. IV (1997), Presidência do Conselho de Ministros, Lisboa, 1998, pp.13-18 (the English version was published in Special Reprint XI, 1998).



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such information for clinical purposes? Is it to be accepted that insurance companies may hang the acceptance of insurance contracts on the submission of predictive tests? Is it fair of employers to hang job contracts on predictive tests?

The latter question appears to have particular relevance in Portugal, for it stirred some controversy recently. For that reason, it will be developed at more length below.

III – GENETIC TESTING IN THE WORKING PLACE

Distinction must be drawn between 1. **genetic monitoring**, 2. **screening of predispositions** and 3. **pre-symptomatic diagnosis**.

1. The genetic monitoring of workers aims to detect the possible genotoxic effects of substances or other factors present in the working place. It has a bearing on the workers' health, on the employer's obligation to ensure work safety and on society's duty to promote health, and it must be compulsory when the case justifies it. Nevertheless, if a worker refuses consent, after being fully informed, the worker's decision must be respected and must not lead to losing the job on those grounds.

It is ethically imperative that the biological materials (blood or other) obtained from the workers for monitoring tests be not used for any other tests, and that they be destroyed immediately after the prescribed utilisation.

In the case of a general positive result, revealing harmful effects of the working environment on all workers, the employer is ethically bound to improve work safety conditions.

It may happen, however, that exposure to the working environment affects only a fraction of the workers, due to the presence of genes predisposing them or making them more susceptible to the diseases caused by the working environment. In such cases, there must be a screening as described in the following paragraphs.

2. Screening of genetic predispositions. Its aim is to prevent workers who have such a genetic predisposition from taking on jobs entailing higher risk of exposure. This limitation of work rights is justified by the workers' health rights. Yet, even in such cases tests are to be performed after free consent given by the worker, and his/her full information. If the worker refuses to undergo the test, the worker's contract must remain in force.

It is important in such cases to consider the disease as being job-related, for, in fact, its agent resides in the working place. Such genetic tests must not in any way degenerate into tests for detection of general hereditary diseases, which might lead to discrimination among workers. Even more important is to prevent the selection of workers who are resistant to the disease, with the purpose of saving the costs of improving safety conditions at the working place. It would be unjust to tolerate low levels of safety at the expense of excluding a sector of the working population.

The health rights of society must also be considered. In some jobs, an unexpected deterioration of the worker's health might affect the safety of the public or of the worker's colleagues. In this case, genetic predictive tests may help prevent serious accidents, and then they are ethically justified. Still, there is a danger of generalising this principle excessively and therefore using predictive tests with unjustified frequency. For instance, no



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ethical reasons justify excluding a person who bears the gene for Huntington's disease from being an air pilot, so long as the disease is not expressed⁵. Legal norms and mechanisms should be established to create competent entities charged with restricting the application of safety-related predictive tests, so as to keep them within proper proportions, and with defending potential or real workers' rights no less than society's right to safety.

3. Pre-symptomatic diagnosis of late-onset diseases that are not caused or influenced by the working environment but will lead in future to disablement of the worker. Employers are interested in obtaining such test results in order to predict the useful life of job candidates. In fact, it is in the employers' interests to reduce production costs and increase the profitability of investments, i.e. to avoid workers' inefficiency or absenteeism, loss of investment on their training through premature disablement, and participated costs pursuant to disease, disablement or death.

Such employers' interests are legitimate, within the context of a market economy that accepts the system of free competition.

Nonetheless, these legitimate rights ought to be placed on only one side of the scale, and counterweighted on the other with the equally legitimate interests of workers and society.

Workers have several interests and rights, such as the right to autonomous decision and informed consent, the right not to know (although it is important that this right be exercised with fully informed awareness of the risks connected with not knowing), the right to privacy, the right not to be discriminated on the basis of genetic traits, and the right to work. The latter constitutes a fundamental element of many modern Constitutions and international agreements⁶.

Work is necessary to self-fulfilment and integration into society; moreover, for the vast majority of the population, it constitutes the main source of income and subsistence. To deny access to work for reasons not connected with inability but with mere prediction of future diseases or predisposition thereto would represent a strong discrimination. It would be even more unjust than withholding from handicapped people the special protection of their right to work prescribed by the Law of most countries.

Job placements must not be denied because of a prediction of possible future diseases. Otherwise, this will create a new class of people, which, albeit able and fit to work now, would be barred from any employment. Such discrimination would be unjust to individuals and economically onerous to society.

For all these reasons, the prevailing ethical position today gives precedence to the worker's rights over the employer's rights. Employers may make the placement depend only on the candidate's present health situation, not on a future health situation. To exclude people from job opportunities on the basis of genetic tests appears to be ethically acceptable only if this is proved to be absolutely necessary to safeguard the worker's health or the safety of third parties⁷.

⁵ K. Berg, "People who are healthy should be treated as healthy persons whatever their future disease risks are." in *Revue Internationale de Droit Économique*, no.1 (1993), p.131.

⁶ Carlos de Sola, "Privacy and Genetic Data. Cases of Conflict" (II), in *Law and the Human Genome Review* no.4 (1995), pp. 147-156.

⁷ *Genetic Screening: Ethical Issues*, Nuffield Council on Bioethics, 28, Bedford Square, London, WC1 3EG, 1993.



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This ethical position is in accordance with Article 12 of the recent Council of Europe's *European Convention on Human Rights and Biomedicine*⁸. It is also in accordance with Articles 6 and 10 of the *Universal Declaration of the Human Genome and Human Rights*⁹, with the even more recent *Final Report of the U.S. Task Force on Genetic Testing*¹⁰, and other documents. Last February, U.S. President Clinton signed an executive order prohibiting all federal employers from admitting, promoting or firing workers on the grounds of genetic information.

Patrick Nairne, "Genetic screening-1", in Wayland Kennet, & John Libbey (eds.), *Parliaments and Screening — Ethical and social problems arising from testing and screening for HIV and genetic disease*. Eurotext, Paris, 1995, pp. 59-64.

⁸ " 'Tests which are predictive of genetic diseases or which serve either to identify the subject as a carrier responsible for a disease or to detect a genetic predisposition or susceptibility to a disease may be performed only for health purposes or for scientific research linked to health purposes, and subject to appropriate genetic counselling.' Nevertheless, article 26 n. 1 admits exceptions," Paula Martinho da Silva, *Convenção dos Direitos do Homem e da Biomedicina*, Cosmos, Lisboa, 1997.

⁹ Article 6: "No one shall be subjected to discrimination based on genetic characteristics that is intended to infringe or has the effect of infringing human rights, fundamental freedoms and human dignity". Article 10: "No research or research applications concerning the human genome, in particular in the fields of biology, genetics and medicine, should prevail over respect for the human rights, fundamental freedoms and human dignity of individuals or, where applicable, of groups of people."

¹⁰ "No individual should be subjected to unfair discrimination by a third party on the basis of having had a genetic test or receiving an abnormal genetic test result. Third parties include insurers, employers and educational and other institutions that routinely inquire about the health of applicants for services or positions. Discrimination can take the form of denial or of additional charges for various types of insurance, employment jeopardy in hiring and firing, or requirements to undergo unwanted genetic testing." Neil A. Holtzman & Michael S. Watson (Eds.) *Promoting Safe and Effective Genetic Testing in the United States*, The Johns Hopkins University Press, Baltimore-London, 1998, p. 15.



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IV – CONCLUSIONS

1. The complete sequencing of the human genome is a highly important scientific achievement, which, within a few decades, will permit establishing the molecular characteristics and eventually the therapy of many more genetic diseases, as well as the construction of personalised medication.

2. However, the possibilities offered by these techniques are not unlimited. For example, the behavioural traits of a person (and even some physical traits) do not depend only, perhaps not even principally, on that person's genes. It is important to inform the public realistically of the many limitations of the new technologies.

3. The ethical principles of the dignity, autonomy and freedom of the human person, and of beneficence and equity, must be strictly respected in all applications made possible by knowledge of the human genome. By way of example, we mentioned the access to predictive genetic diagnosis and to its results in the workplace. Upon informed consent, such tests must be performed, and their results communicated to the employers, only for the sake of the health of the subject or the safety of third parties.

4. Debate must continue on the problem of the juridical protection of sequences of the human genome.

5. The themes broached in this working document will be developed further and completed in this Council's Report and Opinion, presently under elaboration.

Lisbon, the 12th of July 2000.

Rapporteur,

Luís Archer, Ph.D.