

Doc. 8901
6 December 2000

Protection of the human genome by the Council of Europe

Report
Committee on Science and Technology
Rapporteur: Mr Valentino Martelli, Italy, Group of the European People's Party

Summary

The « Human genome » international research project, which results should be made available to all, will have numerous and unimaginable consequences for the social and medical fields and will raise new questions regarding human dignity. It promises many important advances in medicine made possible through the understanding and manipulation of gene mechanisms, but it also raises new fundamental ethical questions regarding further research and applications in this area. It is therefore proposed that each Council of Europe member state should create a mechanism (national authority) to survey and monitor developments relating to the human genome, and that on a European level using the Council of Europe structure a European authority be set up to co-operate with national bodies. These co-operating agencies should also be used to familiarise the European health professions and the public with the new possibilities for progress in genetics in terms of information and technology. This could involve the promotion of campaigns.

I. Draft recommendation

1. The Council of Europe's Parliamentary Assembly notes that the Human genome international research project, in view of the numerous and unimaginable consequences that it might have for medicine and for the animal and plant world, conjures up scenarios for all humanity that raise numerous ethical questions, while holding out the promise of enormous improvements in the quality of life.
2. The protection of human dignity should be the guiding principle for the handling of the Human Genome Project.
3. The genetic age will dawn with the completion of the project: diagnosis will become objective, and it will be possible to identify the presence of genetic disorders or a genetic predisposition to illnesses at an early stage. In many cases, gene therapy will become possible, and this will basically give rise to a form of genetic engineering designed, for instance, to avoid the development of a tumour in an individual found to be at risk. It might also be applied to other illnesses, such as hypertension, diabetes, Alzheimer's disease, osteoporosis, certain psychiatric disorders, etc.
4. At the same time, the Assembly is aware of the enormous ethical implications of further research on the human genome, including some of a negative nature. These include questions regarding the cloning of cells, the conditions ruling genetic testing and the divulgation and use of obtained information.
5. In this connection, the Assembly is fully aware of the now well-known fact that laboratories, with their associated data banks, which conduct experiments specifically on DNA separation are already actively at work in certain European countries and enjoy, among other things, the financial support of prominent pharmaceutical companies.
6. It is also aware that substantial economic interests are at stake in the Genome Project, by virtue of the very fact that it might hold out incalculable opportunities for preventing illness and improving treatment, as it involves many public and private research centres to which considerable financial resources will be allocated.
7. The Assembly is of the opinion that the results of this grandiose research effort – in which the United States has the lead over Europe – must be made available to all, genetic information being a common human heritage, as set out in Article 1 of the Universal Declaration on the Human Genome and Human Rights, adopted at Unesco in Paris on 11 November 1997. The Assembly in particular refers in this context to the Council of Europe Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine – Convention on Human Rights and Biomedicine (European Treaty, Series No. 164) as well as its own Recommendations 1425 (1999) on biotechnology and intellectual property and 1468 (2000) on biotechnologies.
8. In particular, the Assembly is aware that the prospects opened up by the discoveries associated with the Genome Project pose a whole series of ethical problems, essentially concerning such fundamental issues as the use of genetic information for preventive purposes and possibly a presumed right, at a later stage, to take preventive action when certain genetic information is obtained. However, a crucial question will be the assessment of *who will have*

which rights to use the information: the case of insurance companies, employers, parents, schools, etc.

9. The Assembly calls, inter alia through the establishment of a Euroforum on Human Genetics, for the widest possible participation by citizens in the discussion on the human genome through the involvement of the European media and suitable and accurate information by the Council of Europe.

10. In view of the above, the Assembly recommends that the Committee of Ministers:

i. invite every Council of Europe member state concerned to set up, under its own domestic legislation, a national authority having the express task of monitoring, informing and advising on the compliance of research on the human genome with universally recognised ethical and moral principles of respect for life and human dignity;

ii. set up, at European level as well, and more specifically in the context of the Council of Europe, a body or authority to fulfil on a permanent basis the task of monitoring the development of the Genome Project research process, ensuring respect for ethical principles in the context of research on the human genome, assessing the effects of such research also regarding health risks, and giving thorough consideration to all the ethical aspects of the project and consider in this context the role of the Steering Committee on Bioethics (CDBI);

iii. ensure that these bodies for monitoring research on the human genome will familiarise the European public with new possibilities for progress in genetics in terms of information and technology and also serve to promote campaigns to inform and educate the public, in particular the health professions;

iv. make sure that consultation of the European authority be mandatory, and that it formulate an opinion when conventions are drafted on this subject in the context of the Council of Europe and codes of ethics produced; such a body should also have free access to important information on genetics and be able to carry out its own inspections of public and private European research institutes.

v. ask member states to sign, ratify and implement the Convention on Human Rights and Biomedicine”.

II. Explanatory memorandum by Mr Martelli

1. The **Genome Project** is an international research project launched in 1986 at the suggestion of the Nobel Prize winner Dulbecco. Its purpose is to compile and complete the map of the human genes, which number around 36,000. A knowledge of the role of each gene will open up unimaginable prospects for current medicine. The ensuing gains in terms of prevention and healing will be enormous.

2. The Genome Project is being conducted by a large number of countries (currently 18, namely Australia, Brazil, Canada, China, Denmark, European Union, France, Germany, Israel, Italy, Japan, Korea, Mexico, the Netherlands, Russia, Switzerland, the United Kingdom and the United States). The work is being led by two public institutions and one private institution, including the US National Institute of Health and the Department of Energy. The participating research centres have joined together as partners in the Human Genome Project, which aims to make the results available to the whole of humanity. However, an interest is also being taken in the matter by numerous private firms which frequently exploit the above partners' findings for their own advantage. One of these firms, Celera Genomics (Maryland, USA), announced recently (6 April 2000) that it had completed the characterisation of the sequence of bases - there are about three billion of them - composing the genome. The first phase of the task is thus complete. The aim now is to identify the portion of this chain to which every individual gene corresponds and determine each gene's role. This is the most difficult part. Meanwhile, the United States Congress has started to examine possible legislation.

The nature of the genome

3. The genome is the whole of our genetic heritage present in each of our cells. It represents the sum total of the instructions necessary for "reproduction" - but also for the development and creation of the linked structures constituting our bodies - and for our proper functioning as organisms.

4. The genome is initially composed of 46 chromosomes, 23 of which come from the maternal ovule and the remaining 23 from the male gamete. Chromosomes are therefore sub-cellular structures located in the cell nucleus which constitute the first units that link genetic information together.

5. At a more microscopic level, chromosomes are further composed of genes, which number around 36,000 and are the smallest units of significance for the transmission of genetic information. Each gene specifies the ways in which a particular gene is synthesised. Proteins are the basic components in the make-up of organisms and in the unfolding of the vital processes, representing as they do both the structural component of cells and tissues and the key that triggers the biochemical reactions leading to life.

6. Genes (and therefore chromosomes as well) are physically composed of DNA (dioxynucleic acid). Each chromosome consists in fact of a single long DNA molecule; a gene is a small portion of such a molecule.

7. DNA is a large molecule composed of two strands of helical form made up of sugar and of molecules of phosphate aligned inversely to each other; this is the famous double helix discovered by Watson and Crick in 1953, who were awarded the Nobel Prize as a result. The joining together of the two strands gives rise to two pairs of bases: particular chemical compounds whose succession along the DNA molecule is the secret of the genome and its individuality (this is the sequence recently mapped by Celera Genomics). The bases are four in number - adenine, guanine, cytosine and thymine - and what is fundamental is therefore only their spatial arrangement.

The advantages of the Genome Project

8. The potential advantages of the Genome Project are incalculable.

The first discipline to benefit will undoubtedly be medicine, which will undergo radical change the moment we truly enter the *genetic era*. Diagnosis will become completely objective, at least for certain diseases. It will be possible to reach an early diagnosis of the presence both of genetic and hereditary diseases which are difficult to spot (perhaps because they appear at an advanced age or state of development) and of genetic *predispositions* to diseases. In such cases, the therapeutic side among other things may undergo improvements unimaginable today: possible scenarios will include the rational preparation of drugs and more deliberate action aimed at eliminating a faulty gene. We shall thus practise genetic correction for therapeutic ends; such action may be limited to modification of the somatic line - in which case only the characteristics of the individual concerned are altered, and not his genetic heritage - or may be directed at the germinal line - in which case the characteristics not only of the individual but also of his descendants are transformed.

9. Once research has identified the exact molecular alphabet which permits the presence in a human being's genome of a predisposition to a certain type of tumour, it will be possible to intervene much more effectively in order to prevent such a tumour from appearing in a subject found to be at risk. Genetic engineering seems perfectly desirable in such cases. But the knowledge that a predisposition exists is in itself a positive factor that by itself reduces mortality (obviously assuming a strict regime of preventive controls).

10. Furthermore, since the majority of the diseases in question are not caused entirely by genetic factors but have to be triggered by, amongst other things, environmental factors, a knowledge of one's predisposition to disease may be useful in planning, in co-operation with an expert medical team, a lifestyle or a manner of conducting certain specific activities designed to minimise the risk of stirring the predisposition into harmful life. Diseases to which this remark applies include hypertension, diabetes, psychiatric disorders like schizophrenia and manic depression, cardio-vascular defects, tumours, Alzheimer's disease, rheumatism and osteoporosis.

11. Risk assessment is a chapter that medicine has still to write and completion of the genome project could certainly constitute an epoch-making event in this connection. It would become possible to estimate the damage to health caused by smog, radiation and chemical and toxic agents; such damage factors could be calibrated and calculated for any specific individual.

12. Thanks to new advances in therapy, in particular with regard to genetic therapy and predictive medicine, it will be possible to find new therapeutic applications for diseases that until now were incurable. However, the progress of genetics does not obscure but, on the contrary, emphasises the necessity for consideration of the environmental factors involved in disease. If attention to the incidence of specific environmental components can become one of the most important aspects of future medicine, the study of microbic genomics will bring about new environmental-policy prospects and put them on a rational footing. It is worth recalling that the genome of the common fruit fly, *Drosophila melanogaster*, has already been completed.

13. Regarding other than strictly medical applications, it is impossible to ignore the future spin-off from the genetic sciences onto other sectors of life, for example food production. Crop raising and animal production will be genuinely and favourably altered technologically by genetics, which can indicate the timeliness of action to increase productivity whilst simultaneously enhancing product quality. It is also becoming a matter of urgency to distinguish between harmful and non-harmful genetically modified foods, the latter representing at the very worst a real improvement over the original varieties. Distinctions of this type are only possible with a mature and complete science of genes. Ever-present in our minds must be the fact that every minute 10 children die of hunger, that every hour hundreds of human beings are killed by negative reactions to medicines, that the Aids virus reproduces itself in a sufferer's body a billion times a day.

14. History, archaeology, anthropology and the cognitive and social sciences will receive a fundamental boost from the completion of the map of the human genome. It will be possible to resolve age-old philosophical debates about the innate or acquired nature of certain abilities - first among them all, the faculty of language; it will be possible to adopt a new approach to the study of questions like the evolution of the human species by exploring the more mysterious historical processes through the heuristic filter of the genes (human migrations, bio-archaeology), as has been done for some time by one of our best scientists, Luca Cavalli Sforza, in his masterly crossing of genetics and linguistics which may well be worthy of a Nobel Prize.

Ethical problems raised by knowledge of the genome and necessity for a monitoring body

15. The problem posed by the increasing availability of information of as great a moral and economic value as that concerning the human genome undoubtedly gives rise to unavoidable ethical tensions. It is predicted that by 2009 sales of DNA-based products and technology will exceed 90,000 billion dollars in the USA alone. The financial interests involved are enormous but the specific nature of the information makes the question extremely sensitive. First and foremost, should this information be considered to constitute the common heritage of humanity, or should it belong to the person who holds it or who discovered it? Article 1 of the UNESCO Declaration on the Human Genome and Human Rights explicitly states that the genome is "a common human heritage", but the implementation of this formula must be continuously monitored and such implementation is often debatable. The decision by Celera Genomics to make the recently completed map of the bases available to anyone over the Internet but only for payment has naturally provoked controversy. As far as the Parliamentary Assembly of the Council of Europe is concerned, it has voted two important recommendations in this context. Recommendation 1425 (1999) on biotechnology and intellectual property specifically asks for improvements of international

legislation in this field and stresses the importance of considering the ethical aspects of the patentability of inventions involving biological and, in particular, human material. Recommendation 1468 (2000) calls for a new international convention on the use of living matters. It opposes on ethical grounds Article 5, sub-paragraph 2 of the Directive 98/44/EC of the European Parliament and Council of the European Union of 6 July 1998 on the legal protection of biotechnological inventions which states that “an element isolated from the human body or otherwise produced by means of a technical process, including the sequence or partial sequence of a gene may constitute a patentable invention, even if the structure of that element is identical to that of a natural element”. This makes the first sub-paragraph inoperative which states that the human body and its parts cannot constitute patentable inventions.

16. However, this discussion represents only the tip of the iceberg. What will become increasingly crucial is the question of *who has the right to use* the information.

- Under which conditions has an insurance company the right to know the results of genetic tests on potential customers with a view to requiring a higher premium from individuals at risk from or predisposed to a large number of serious disorders? Or does this amount to discrimination against the less fortunate?
- Does a parent or couple have the right to subject its/their child to genetic tests of any kind? Have they the right to *compel* the child to undergo such tests? Has anyone the *right* to impose this on anyone else? (A person on his or her own spouse? A child on his or her parents?)
- Are employers, firms, universities or schools entitled to select their employees or students according to considerations connected with examination of the genetic code? It is, of course, unnecessary to *demand* a test in order to use it as a basis for discrimination, since it is sufficient to accept only those who have “freely” undergone the test with results considered positive by the selector.
- Has anyone the right to divulge genetic information relating to another person? (Suppose the information concerns the presence in an individual of a genetic threat to that person’s descendants, the individual being the healthy carrier of a serious genetic disorder. Do we have a *duty* to warn the person’s partner? Do we have, in addition, a duty to *prevent* that person from having children?)
- Does one have the right, in some cases, to demand privacy regarding information on one’s own genome? If so, when?
- Does the state have the right to force its citizens to take genetic tests?
- Prenatal tests. *May* or *must* parents have them carried out? What results could entitle them to take corrective action? What results could justify a decision to prevent the embryo from being born?
- In what cases could a child born with genetic malformations or disorders take legal proceedings against parents or doctors who fail to act despite awareness of the facts?

- There are cases in which it is difficult to tell the difference between a disease and an imperfection. How will we distinguish between therapy and simple improvement? Regarding the latter, supposing parents wish to programme the colour of their child's eyes and technology makes this possible, are we to permit it or not?
- Technology has a cost which the individual user might be made to pay in its entirety or which could be met in whole or in part by the public health authorities.
- Might adoption of the concept of "genetic predisposition" lead to a weakening of the idea of moral responsibility? How threatening does the idea of genetic determinism become? What are its implications for the great ideas of liberty and equality?

17 The legal system certainly already provides for means and solutions – e.g. the constitutional prohibition (Article 32) on compulsory medical treatment – allowing some of these questions to be answered. However, there is no doubt that certain of the issues concerned may be affected by the Genome Project in currently unimaginable ways. It is accordingly necessary to create a monitoring body which will act as a reliable point of reference in the confused discussion that otherwise awaits us.

18. The challenges posed by technical knowledge and resources are already upon us: they cannot be evaded but must be confronted as calmly and thoughtfully as possible, setting aside prejudices, untruths and unverified inferences.

19. The nature and density of the material and the sensitivity and number of the consequences make it urgently necessary to create a monitoring body whose special task will be to keep a critical eye on matters connected with the human genome and related problems. If the century ahead is really to be the century of biology, a permanent body will have to be set up to monitor one of the key subjects in the development of the biological sciences on which the country's legislators and philosophical and legal authorities have yet to pass decisive judgment, namely genetics.

Tasks of the monitoring body

20. The monitoring body must study and take decisions on all matters relating to human and non-human genetics submitted to it. The ethical status of its task will be reflected in the planning of public and medical policies. In particular, the monitoring body will examine:

- the process of completion of the Genome Project and its consequences;
- the possibility of perceiving new implications of the concept of "injustice" in the scenarios opened up by the availability of genetic information and genetic technology;
- the process of integrating into current medical practices, the new medical treatment and policies, hospital environments, and new information and technological facilities resulting from progress in genetics;
- novel bioethical issues emerging in the genetics field;

- novel sociological and political issues arising out of the new light thrown by genetics on seemingly unrelated social and political phenomena (interaction between racial, ethnic and socio-economic factors);
- the advisability of promoting information, education and training policies and campaigns for the public and the health profession;
- management of prevention of the risk of genetic weapons falling into the hands of enemy or terrorist states, a matter recently raised by Neal Lane, scientific adviser to the White House.

Reporting committee: Committee on Science and Technology.

Reference to committee: Doc. 8773 and Reference No. 2520 of 26 June 2000.

Draft recommendation unanimously adopted by the committee on 29 November 2000.

Members of the committee: MM. *Rakhansky* (Chairman), *Birraux* (Vice-Chairman), *Tiuri* (Vice-Chairman), *Lekberg* (Vice-Chairman), *About*, *Asciak*, *Bartos*, *Brunhart*, *Bulic*, *Cherribi*, *Cioni* (Alternate: *Martelli*), *Colla*, *Cunliffe* (Alternate: *Etherington*), *Damanaki*, *D'Aron*, *Dolazza*, *Duka-Zolyomi*, *von der Esch*, *Mme Faldet*, *M. Fayot*, *Mrs Fernandez Capel*, *Gligoroski*, *Guardans*, *Ivanov*, *Jackson*, *Kitov*, *Kittis*, *Kjaer*, *Kolb*, *Kurucsai*, *Leers* (Alternate: *Dees*), *Lengagne*, *Libane*, *Liiv*, *Lotz*, *Maass*, *Marmazov*, *Mateju*, *Melnikov*, *Minarolli*, *Monteiro*, *Moreels*, *Moynihan-Cronin*, *Mozgan*, *Mrs Nabholz-Haidegger*, MM. *Niculescu*, *Nigmatulin*, *Nikologorsky*, *Patarkalishvili*, *Pawlak*, *Plattner*, *Mrs Ragnarsdottir* (Alternate: *Olrich*), MM. *Rapson*, *Raskinis*, *Riccardi*, *Roseta*, *Scheer*, *Steolea*, *Tanik*, *Turini*, *Wittbrodt*, *Wurm*, *Yürür*, *Zissi*.

N.B. The names of members present at the meeting are printed in italics.

Secretaries of the committee: Mr Lervik, Mr Torcătoriu.